WEAVE SPRINT . PAPER III

Intellectual disability & autism

Intellectual disability from its definition to its measurement, its causes from the chromosome to the household, and autism from Kanner to the spectrum: criteria, recognition, the instruments and what they actually measure, the interventions that earn their place, and the Indian statutory route from assessment to entitlement.

CONTENTS

- Intellectual disability: the concept and its measurement
- The causes of intellectual disability
- Living with intellectual disability
- Autism: concept, causes and criteria
- The clinical picture of autism and how it is recognised
- Managing autism and its outcome

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PAPER III

Intellectual disability & autism spectrum disorder

Six bands . one complete notes document

Intellectual disability: the concept and its measurement

1 · The neurodevelopmental frame

Begin with the person, not the label. Intellectual disability and autism spectrum disorder are developmental presentations that demand careful formulation rather than rapid categorisation. The marks lie in showing that one can move from a concern raised by a family or school to a coherent account of abilities, everyday functioning, communication, behaviour, context and need. A diagnosis may organise that account, but it does not replace it.

This chapter is best read as a clinical sequence. It first establishes the language and measurement needed to describe intellectual disability, then considers causes, associated psychiatric and practical concerns, and care across settings. It then develops autism spectrum disorder from its diagnostic framework through recognition, assessment, intervention and outcome. The final task is discrimination: deciding which explanation best fits the observed developmental pattern and what the distinction changes for the person and family.

ID	ASD	PERSON
CONCEPT AND MEASUREMENT	RECOGNITION AND FORMULATION	FUNCTION BEFORE LABEL

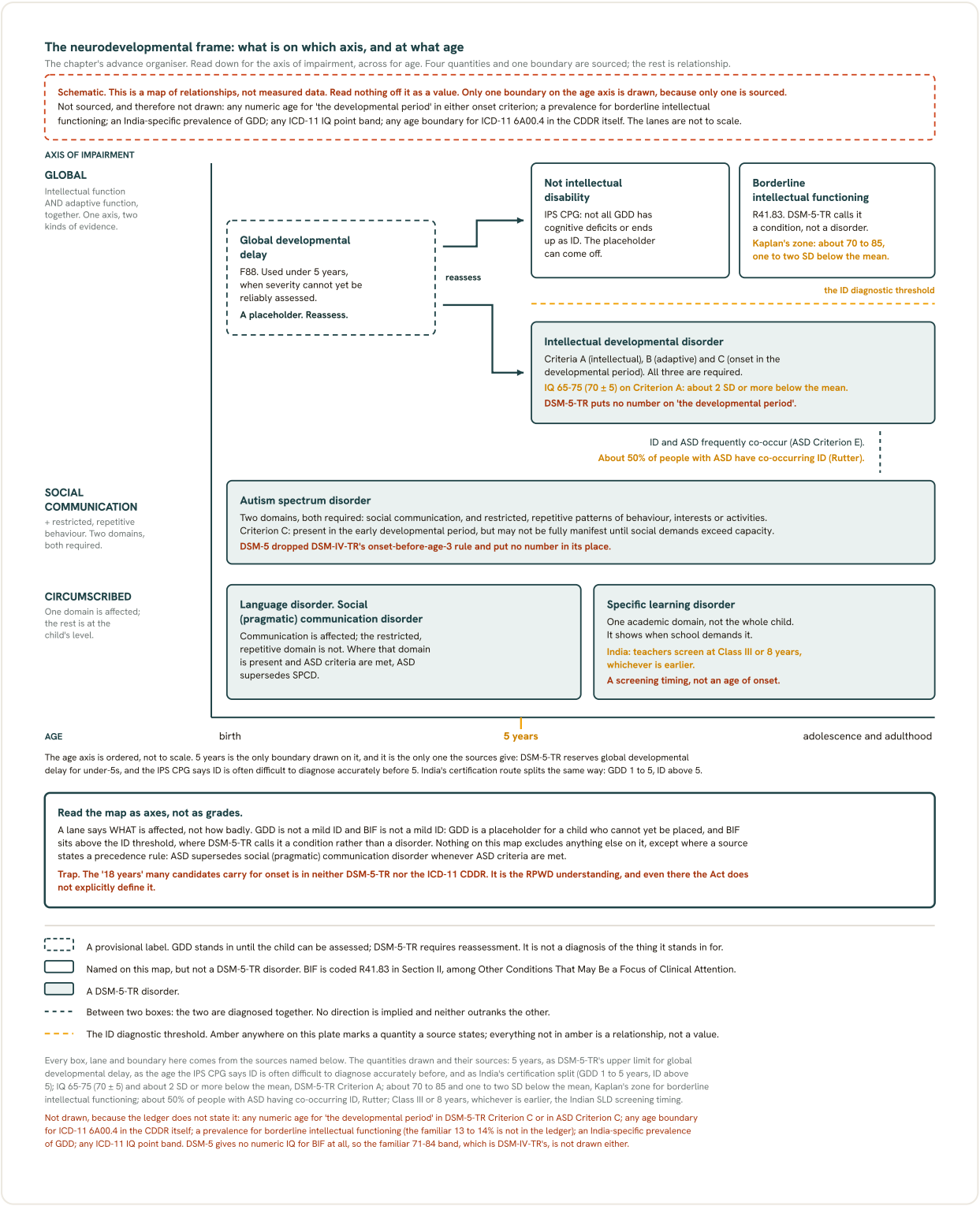


Fig 21.1 The neurodevelopmental frame. The chapter's advance organiser: age runs across, the axis of impairment runs down, and every entity the chapter teaches has a place on both. Global developmental delay is drawn with a dashed border because it is a placeholder, not a diagnosis, and it forks on reassessment; intellectual developmental disorder and autism spectrum disorder sit in different lanes and are joined by a dashed rule because they are diagnosed together rather than ranked; borderline intellectual functioning sits on the same global axis as ID but above the diagnostic threshold, where DSM-5-TR calls it a condition and not a disorder. The age axis is ordered rather than scaled, and carries the single boundary the sources give.

1.1 The organising clinical question

A **developmental formulation** is a working explanation of how a person has developed, what they can do in ordinary settings, what creates difficulty, and what may help. It is deliberately

broader than a diagnostic label. It prevents a clinician from mistaking a test result, a conspicuous behaviour, or a single historical detail for the whole clinical picture.

RULE **Formulate functioning in context before using a diagnostic label to organise the next step.** A label becomes clinically useful only when it clarifies the pattern of difficulty, the questions still to be answered, and the support that may be needed.

At the outset, hold several strands together:

- the **presenting concern** in the words of the person, family, school or referrer;
- a **longitudinal history** that asks how the current pattern emerged and changed;
- the person's **functional profile** across their actual environments; and
- the **contextual formulation** of relationships, opportunity, stress, culture and available support.

Developmental expectations belong in *the Normal child development and milestones notes*. A full child psychiatric assessment, service response and safeguarding approach belong in *the Child psychiatry: assessment, treatment, services and protection notes*. Those cross-references matter because a developmental formulation is only as good as the history and context on which it rests.

1.2 A map of the chapter

The chapter separates related questions so that each can be answered precisely. The following map is a reading aid, not a diagnostic algorithm.

CLINICAL QUESTION	CHAPTER FOCUS	WHY THE DISTINCTION MATTERS
How should intellectual ability and everyday functioning be described?	Concept, criteria, assessment and adaptive behaviour	Description needs interpretation in the person's life context.
What may account for the developmental presentation?	Causes and syndrome-specific discussions	Explanation can alter investigation, anticipatory care and counselling.
Is autism spectrum disorder the best account of the observed developmental pattern?	Diagnostic framework, clinical picture and assessment	Recognition requires a pattern-based judgement rather than an isolated feature.
What should change in the person's everyday life?	Interventions, health review, family support and prognosis	A **support plan** translates formulation into feasible action.

The questions interact, but they should not be collapsed. A person may need more than one explanatory frame, and the clinician must decide what each frame adds. Assessment methods, severity language, aetiological work-up and intervention are therefore developed in their own sections rather than treated as interchangeable.

TRAP **Examination trap:** do not let an early label replace description of the observed problem. Establish the pattern first, then use the relevant framework to explain it.

1.3 Reading findings without reductionism

GOOD TO REMEMBER

Interpret clinical information together: history may reveal a developmental trajectory; direct observation may show communication and response to change; assessment may characterise ability or practical skill; and collateral may reveal demands invisible in the consulting room. Check **ecological validity**: the account should fit the person's life outside the clinic as well as the clinician's impression inside it.

Use the chapter to ask progressively sharper questions: What is the pattern of strength, difficulty and participation? What information would materially change the formulation? Which co-occurring concerns require active recognition rather than attribution to the developmental diagnosis? What support is needed now, and what should be reviewed as circumstances change?

This approach resists **diagnostic reductionism**: the error of allowing one diagnosis to explain every difficulty, behaviour or need. It also makes room for the person's preferences and the family's realities. Good developmental psychiatry is exact about constructs while remaining practical about lives.

1.4 Master mnemonic: MAP

A compact way to retain the chapter. Use **MAP** as a sequencing aid when approaching a developmental presentation. It is not a substitute for assessment; it is a safeguard against beginning in the wrong place.

MNEMONIC

MAP

Map the developmental and functional story.
Ask which framework best explains the pattern.
Plan support around the person, family and participation.

The first step keeps the account anchored in lived function. The second directs the clinician to the chapter's conceptual and diagnostic material without forcing a premature conclusion. The final step prevents assessment from becoming an endpoint: the relevant outcome is a more useful, humane and workable response.

The durable answer in developmental psychiatry is a formulation that explains function, recognises pattern and directs support.

2 · Global developmental delay concept

Global developmental delay is a clinical description for an early developmental presentation, not a shortcut to a settled prognosis. It is used when a child has not acquired expected developmental abilities across several areas, but the clinician cannot yet make a reliable statement about the severity of intellectual impairment. The label is valuable precisely because it

preserves that uncertainty. It allows the record to acknowledge a real developmental concern, organise the next clinical steps, and communicate need without pretending that a premature measurement has answered the longer-term question.

The topic is frequently examined because it tests clinical reasoning at the boundary between observation and diagnosis. Candidates must distinguish a broad developmental delay from a definitive intellectual-developmental diagnosis, explain why early assessment can be limited, and avoid converting a description of current functioning into a prediction about lifelong ability. In DSM-5-TR, global developmental delay is recorded with the code **F88**. The code should serve the formulation, not replace it.

F88	below 5 years	between 3 months and 5 years	at least 2 domains
DSM-5-TR GDD CODE	DSM-5-TR AGE USE	INDIAN GUIDELINE WINDOW	OPERATIONAL THRESHOLD

2.1 What the label is for

Global developmental delay is reserved in DSM-5-TR for a child **below 5 years** when the clinical severity of intellectual impairment cannot yet be assessed reliably. This is not merely a matter of a child being young. The label is appropriate when the child has failed to achieve expected milestones in several areas of intellectual functioning and cannot undergo a systematic assessment that would permit a dependable severity judgement. Developmental variation, limited cooperation, sensory or physical difficulties, and the practical limits of formal assessment can all make an apparently precise conclusion unsound.

The key idea is **assessability**. A clinician may have persuasive evidence that development is not proceeding as expected while lacking a valid way to quantify the enduring intellectual and adaptive picture. The correct response is neither to deny the concern nor to force a severity label. Global developmental delay creates an honest interim formulation: there is a clinically meaningful developmental problem, its pattern must be described, and the formulation must remain open to revision.

RULE **Defining principle.** Use global developmental delay when delay is evident but a reliable developmental severity judgement cannot yet be made; make reassessment part of the diagnosis from the outset.

GOOD TO REMEMBER

Families often arrive seeking a definitive answer after a milestone concern, a referral from an educational setting, or an earlier clinical opinion. The psychiatrist must hold uncertainty without sounding evasive. Explain what has been observed, what remains difficult to judge, and what information will make the formulation more robust. A diagnostic label should reduce confusion, not offer a false certainty that later requires withdrawal.

Developmental expectations and their usual sequence belong in *the Normal child development and milestones notes*. The full approach to history-taking, examination, assessment planning, services and protection belongs in *the Child psychiatry: assessment, treatment, services and protection notes*. This section focuses on the conceptual work: when to use the label, what it means, and what it does not mean.

2.2 Why early developmental uncertainty is clinically real

Early childhood is a period of rapid acquisition, uneven performance and changing demands. A child may demonstrate an ability in a setting but not another, or show an emerging skill that cannot yet be relied upon in everyday life. Conversely, a striking delay may be clear before the clinician can determine whether it reflects a broad and persistent intellectual-developmental difficulty, a transient lag, a limitation of the available assessment, or a combination of these. The purpose of global developmental delay is to represent this clinical state accurately.

IN INDIA

The Indian guideline makes the same practical point: although intellectual disability may be recognisable early in development, it is often difficult to diagnose it accurately before 5 years of age. This is why the guideline uses global developmental delay as a **surrogate marker between 3 months and 5 years**. "Surrogate" should be read carefully. It means the term can organise recognition and follow-up during a period when the final intellectual-developmental formulation is not yet reliable. It does not mean that the two labels are interchangeable.

Diagnostic uncertainty is not diagnostic inaction. A useful **developmental formulation** states the abilities that are emerging, the activities that remain difficult, the contexts in which difficulty is most evident, and the collateral information needed to clarify the pattern. It also records uncertainty explicitly. This protects against a common clinical drift in which a provisional descriptive term is treated as though it had already settled questions about intellectual ability, adaptive functioning, cause or prognosis.

A child who cannot complete a systematic assessment should not be described as "untestable" and then left outside a formulation. The more accurate question is what has prevented a valid assessment on this occasion and how the clinician can obtain a better account of function over time. That may involve observation in familiar settings, informed collateral accounts, repeated contact, and attention to communication, motor, sensory and behavioural barriers. The methods and interpretation of intelligence testing are dealt with in the chapter's assessment material; the present point is that a score is not required before a developmental concern can be named responsibly.

TRAP **Examination trap:** do not write that global developmental delay is simply "intellectual disability in a young child". It is the appropriate label when the developmental concern is clear but the enduring degree of intellectual impairment cannot yet be judged reliably.

2.3 Operationalising a broad developmental delay

The Indian guideline cites **Shevell et al. (2008)** for an operational definition. In that approach, significant delay must be present in **at least 2** developmental domains. This is an operational framework for describing global delay, not a substitute for the diagnostic rationale about why a reliable severity assessment cannot yet be made. In an answer, it is good practice to name the framework being used rather than silently combining criteria from different systems.

The listed areas form a **developmental profile**, not a checklist to be ticked without context. A delay in a domain may affect performance in another. For example, limited motor opportunity can alter what is observed in self-care, and reduced communication can obscure social participation. The clinician's job is to decide whether the observed pattern reflects broad developmental delay rather than merely adding isolated concerns together.

DEVELOPMENTAL AREA	QUESTION FOR THE CLINICAL FORMULATION	COMMON REASONING ERROR TO AVOID
Gross and fine motor	What movement and hand-use abilities are established in ordinary routines?	Assuming that limited opportunity and intrinsic delay are automatically the same.
Speech and language	How does the child understand, express and use communication in context?	Equating a language difficulty with global delay without reviewing the broader profile.
Social and personal functioning	How does the child participate with familiar people and manage age-expected interactions?	Explaining every social difficulty through a single diagnostic label.
Cognition	How does the child learn, solve everyday problems and generalise an acquired ability?	Treating an isolated observed success or failure as the whole cognitive account.
Activities of daily living	What can the child do in dressing, feeding, routines and other everyday tasks?	Ignoring environmental support or opportunity when judging independence.

The table is deliberately descriptive. It does not convert the domains into a severity scale, an intelligence test, or a statutory disability assessment. Those are different constructs and are treated elsewhere in the chapter. For this topic, the useful result is a coherent account of which developmental abilities have not emerged as expected, which strengths are already present, and why the available evidence supports a broad developmental concern.

- Describe abilities in the child's actual routines rather than using only a global label.
- Identify whether delays are spread across the developmental profile or confined to a more limited concern.
- Record factors that may make observed performance underestimate the child's capacity on that occasion.
- State what should be reviewed when more reliable developmental information becomes available.

2.4 A provisional formulation, not a forecast

Global developmental delay is often clinically adjacent to intellectual developmental disorder, but adjacency is not identity. DSM-5-TR notes that some children younger than 5 years whose later presentation meets criteria for intellectual developmental disorder first meet criteria for global developmental delay. This is an account of a possible developmental pathway, not a rule that every child with global delay will later receive that diagnosis.

The Indian guideline is explicit about **prognostic non-equivalence**: global developmental delay is not an inevitable precursor to intellectual disability. Some children described as having global delay will not have cognitive deficits that persist as intellectual disability. This caution is more than a prognostic nicety. If the early label is treated as a permanent verdict, the family may be given an unnecessarily narrowed view of the child's possibilities, while the clinician may stop looking for change, emerging skills or a more differentiated explanation.

Equally, the opposite error is harmful. Because some developmental delays may change over time, clinicians should not minimise a broad delay in the hope that it will resolve without review. A longitudinal formulation balances both risks: it recognises current impairment and need, avoids premature permanence, and specifies the questions that subsequent assessment must answer. It is a way of being precise about the present without claiming ownership of the future.

MNEMONIC

PAUSE when the early developmental picture feels uncertain

Profile the child's functioning in ordinary settings.
Ask whether current assessment is genuinely valid.
Use uncertainty honestly rather than forcing a severity label.
See whether delay extends across developmental areas.
Evaluate again as the profile becomes clearer.

This is not a substitute for assessment. It is a guard against the familiar tendency to make the label carry more certainty than the evidence can support. The careful clinician does not wait passively for certainty; the clinician makes the uncertainty visible, develops a plan to reduce it, and documents the child's current needs in the meantime.

2.5 Reassessment is part of the diagnosis

The DSM-5-TR conditions of use for global developmental delay include **reassessment** after time has passed. That is not an administrative afterthought. The diagnosis has been chosen partly because present methods cannot support a reliable severity judgement; repeating the same unsupported conclusion at later contacts defeats its purpose. Reassessment asks whether the developmental trajectory has clarified, whether systematic assessment has become feasible, and whether the child's everyday functioning now supports a more definite formulation.

Good documentation makes this possible. It separates what was directly observed from what was reported, identifies the developmental areas of concern, records strengths and contextual supports, and states why the assessment could not yet be definitive. It also avoids language that

freezes a child into an early label. "Current presentation is consistent with global developmental delay; reassessment is planned as developmental information becomes more reliable" is clinically more useful than either an unsupported categorical diagnosis or a vague note that offers no plan.

The review is not solely a search for a new diagnostic name. It should ask whether the formulation has become more specific, whether the child's functional needs have changed, and whether the initial pattern needs to be reconsidered. A broad developmental concern can coexist with communication, motor, sensory, medical or contextual contributors that alter what is seen. The fuller developmental and psychiatric assessment belongs in *the Child psychiatry: assessment, treatment, services and protection notes*; this label should prompt that work rather than close it down.

TRAP **Examination trap:** "provisional" does not mean "wait and see without a plan". It means document the current developmental concern, address present need, and arrange the reassessment that the provisional formulation requires.

2.6 Coding boundaries and classification caution

Classification systems do not always provide a direct vocabulary correspondence for the same clinical presentation. DSM-5-TR supplies global developmental delay as F88 for the early-childhood situation described above. By contrast, the **ICD-11 CDDR** contains no category named global developmental delay. The safe clinical conclusion is not to invent a correspondence. Describe the child's developmental presentation clearly, use the classification framework required in the setting, and record why diagnostic certainty is limited.

A related DSM-5-TR category is **Unspecified Intellectual Developmental Disorder**, coded **F79**. It is reserved for exceptional circumstances in an individual above 5 years when locally available procedures cannot establish the degree of intellectual developmental disorder because associated impairments or severe problem behaviours make this difficult or impossible. It also requires reassessment. This is a different problem from the expected early-childhood uncertainty represented by global developmental delay, and the age boundary should not be reversed in an examination answer.

CLINICAL SITUATION	LABELING LOGIC	ESSENTIAL CAUTION
Early childhood with broad developmental concerns and unreliable severity assessment	DSM-5-TR global developmental delay, F88, for a child below 5 years.	It is a provisional developmental formulation that needs review.
Later assessment made impracticable by associated impairments or severe problem behaviours	DSM-5-TR unspecified intellectual developmental disorder, F79, in exceptional circumstances above 5 years.	Do not use it as a routine substitute for a proper assessment.
ICD-11 terminology	The CDDR has no category named global developmental delay.	Do not assert an unverified direct counterpart.

Kaplan and Sadock's summary describes an ICD-11 provisional disorder-of-intellectual-development category for a child below 4 years when there is evidence of disorder but it is difficult to know whether observed impairments may be transient. This is a useful reminder of the underlying clinical problem, but it does not resolve the naming discrepancy in ICD-11. The relevant **provisional category** reflects uncertainty about whether early impairment will persist, rather than a licence to treat every early delay as a definite disorder of intellectual development.

2.7 Clinical communication and the referral-to-support link

The value of recognition is practical. Identifying global developmental delay gives the clinician a shared language for referral, follow-up and communication with caregivers. It tells other professionals that developmental concerns extend beyond an isolated complaint and that the child needs an organised account of function. It also preserves the imperative to review the formulation as the child becomes more assessable. That combination of recognition and openness is the clinical strength of the term.

Conversations with families should be clear about what is known and what is not. Avoid implying that a descriptive early label proves a fixed lifelong outcome. Avoid the opposite reassurance that a broad delay needs no active response because the future is uncertain. Explain that the current pattern warrants attention, that abilities and needs should be described in everyday terms, and that later reassessment can refine the understanding. The family's observations of what the child can do, what needs assistance, and which settings make participation harder are central clinical data, not decorative collateral.

Why identifying global developmental delay matters for referral and **early intervention** is developed in the management section of this chapter. The present diagnostic task is more limited: make the developmental concern visible early enough that appropriate support is not postponed while diagnostic certainty is still evolving. The service model, its evidence and its Indian delivery context belong to that owner section and should not be inferred from the label alone.

- Communicate the present developmental profile in plain, functional language.
- State clearly that global developmental delay is an interim formulation rather than a prediction of a fixed outcome.
- Document the reason a definitive severity judgement is not yet reliable.
- Build reassessment into the plan and record what information should clarify the next formulation.

> Global developmental delay names a genuine broad developmental concern while protecting the child from a severity judgement that the available evidence cannot yet support.

3 · Definition and diagnostic criteria of intellectual disability (DSM-5 and ICD-11)

The diagnostic task is to establish a developmental disorder of functioning, not to attach a label to a low test score. Intellectual disability is diagnosed when limitations in intellectual

functioning, adaptive functioning, and developmental onset form the same clinical picture. The criteria are deliberately conjunctive: each protects against a different error. Intellectual testing without functional assessment can medicalise a score; functional difficulty without developmental onset may represent an acquired disorder; and a developmental history without evidence of current limitation does not itself establish the diagnosis.

For examination answers and clinical records, the useful sequence is therefore simple: establish the nature of the intellectual limitation, establish its consequences for everyday independence and social responsibility in the person's sociocultural setting, and establish that the pattern began in development. The DSM formulation calls the condition **intellectual developmental disorder**, with intellectual disability retained in parentheses; the ICD formulation uses the equivalent term disorders of intellectual development. The difference in wording should not obscure their shared clinical core.

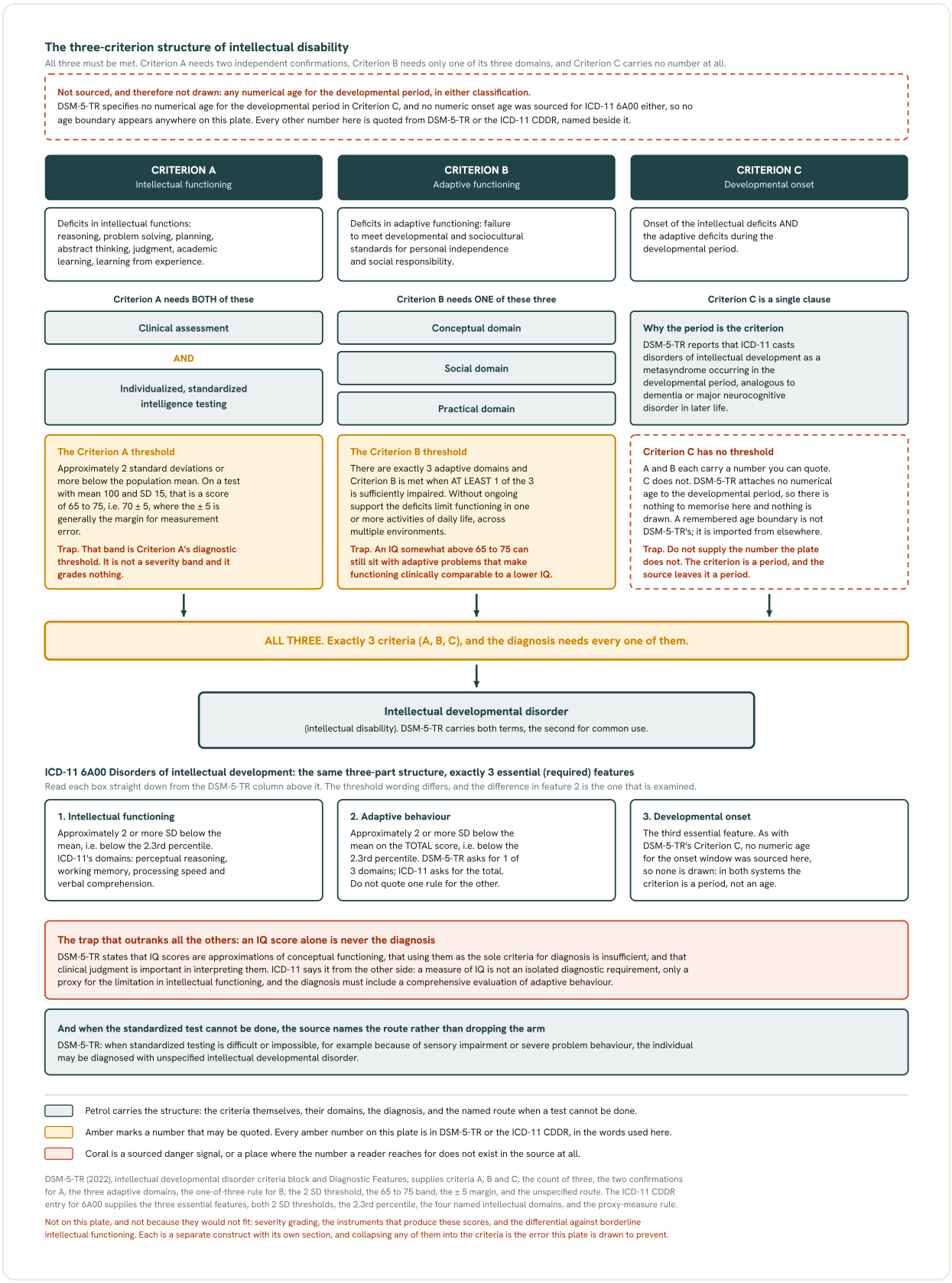


Fig 21.2 The three-criterion structure of intellectual disability. Criteria A, B and C are joined by a conjunction, not a menu: DSM-5-TR requires all three, and the two internal rules that decide A and B are examined more often than the criteria themselves. The ICD-11 6A00 strip reads straight down from each DSM column so the one real divergence stands out.

3.1 What the diagnosis is designed to capture

The diagnosis is about a persistent limitation in the person's capacity to understand, learn, reason and manage the ordinary demands of life, where that limitation has developmental origins. It is

neither a synonym for educational underachievement nor a shorthand for disadvantage, illness, sensory disability, behavioural disturbance, or inadequate opportunity. Each of these may complicate assessment, and some may impair performance in a testing situation, but none can substitute for the full diagnostic formulation.

DSM expresses this through **three** criteria that must all be met. Their structure is clinically elegant. The intellectual criterion asks whether the relevant cognitive functions are impaired. The adaptive criterion asks whether that impairment matters in real life. The onset criterion prevents an acquired cognitive disorder from being placed in a developmental category. The diagnosis consequently rests on convergent evidence, not on one encounter or one instrument.

RULE **Diagnostic principle:** diagnose intellectual disability only when intellectual limitation, adaptive limitation and developmental onset are demonstrated together.

This formulation is especially important where a family requests a certificate, school accommodation, or diagnostic clarification after a long period of difficulty. The clinician should keep the diagnostic question separate from the administrative question of whether services are known to the family or available locally. Service contact may reveal need, but it does not create the disorder. Conversely, absence of prior contact does not negate a developmental disability.

3.2 DSM diagnostic criteria: the diagnostic formulation

Criterion A concerns deficits in intellectual functions. In practice, the clinician looks for difficulty in reasoning, problem solving, planning, abstract thinking, judgment, academic learning, or learning from experience, and confirms the finding through both clinical assessment and individually administered standardised intelligence testing. The word “both” matters. A formal result gives disciplined comparison with an appropriate normative group, whereas the clinical assessment asks whether the result is coherent with the history, observed performance, language, education, sensory status and opportunities to learn.

Criterion B concerns **adaptive functioning**. The relevant question is whether deficits lead to failure to meet developmental and sociocultural expectations for personal independence and social responsibility. Without continuing support, the deficits must limit activity in everyday life across multiple settings. This moves assessment from an abstract estimate of ability to the person’s actual functioning at home, in education or work, and in the community. The criterion can be met where there is sufficient impairment in at least one adaptive domain; the domains are conceptual, social and practical.

Criterion C requires onset of both intellectual and adaptive deficits during the **developmental period**. This is a historical criterion, so it requires collateral. Developmental records, school reports, informant accounts and prior assessments may be more informative than a patient’s retrospective account alone. The question is not whether the person was ever labelled, but whether the characteristic pattern was present in development.

- Ask whether the cognitive difficulty is broad enough to affect reasoning and learning rather than reflecting a circumscribed academic problem.

- Ask what the person can do independently, what they can do only with prompting, and what remains unsafe or unachievable despite ordinary opportunities.
- Seek a developmental history from someone who knows the person's earlier functioning, while recognising that documentation may be incomplete.

The criteria are not a checklist to complete mechanically. A person may perform poorly because of an acute mental state, language mismatch, sensory limitation, poor engagement or an unsuitable test. Equally, a person may show apparently acceptable performance in a familiar, highly supported routine while being unable to generalise skills or assume responsibility in less structured settings. Clinical synthesis is what decides whether the three criteria describe one developmental condition.

3.3 Interpreting the intellectual-functioning criterion

Criterion A is met when intellectual functioning falls **two or more standard deviations below the population mean**. That is a diagnostic threshold, not a severity grade, and it must not be turned into one: this chapter grades severity by adaptive functioning, and the reasons are set out above. What an IQ score actually is, how far it can be trusted, and what makes an obtained score misleading are the business of the clinical-assessment section, which takes the normative distribution, the error margin around a score, the **Flynn effect**, practice effects and the instruments that cannot carry a diagnostic opinion in turn. Read it before you act on any number.

TRAP **Examination trap:** an IQ score is evidence for Criterion A, not a stand-alone diagnosis and not a substitute for adaptive assessment.

One consequence belongs here rather than there, because it is a statement about the criterion itself and not about the instrument. Discordance between intellectual testing and everyday performance deserves active thought rather than reflexive exclusion: DSM recognises that a person whose score lies somewhat above the conventional Criterion A band may nonetheless have adaptive problems so substantial that their actual functioning resembles that of people with lower scores. The response is not to lower a threshold by intuition. It is to review test validity, collateral information, the quality of adaptive evidence, and the developmental course before reaching a defensible formulation. The criterion is a floor for evidence, not a verdict delivered by a number.

3.4 Adaptive functioning and onset: turning scores into a diagnosis

Adaptive functioning is the bridge between ability and lived consequence. It asks whether the person can meet the expectations of independence and social responsibility that are reasonable for their developmental stage and sociocultural context. It is assessed through functioning, not inferred from social class, literacy, diagnosis already recorded in a file, or the wishes of a caregiver. The clinician should distinguish lack of opportunity from inability, and a task performed under extensive supervision from a task performed independently and reliably.

GOOD TO REMEMBER

Adaptive functioning asks what the person can do in everyday life across settings without ongoing support. Assess the quality and consistency of functioning, the scaffolding supplied by relatives or institutions, and what happens when that scaffolding is withdrawn. Measurement of adaptive behaviour belongs alongside, rather than in place of, this clinical enquiry. The framework specifies three domains: conceptual, social and practical.

Developmental onset has a second purpose beyond classification. It forces a chronological formulation. If the observed cognitive and functional decline is new, fluctuating, or clearly follows a later medical or psychiatric event, the clinician must consider an acquired cause rather than assume an intellectual developmental disorder. If early difficulties were present but were never assessed, the absence of a historic score should not itself defeat the diagnosis. What matters is whether reliable developmental evidence can establish the required pattern.

MNEMONIC

Think "mind, mastery, maturation"

Intellectual functions describe the mind; adaptive functioning demonstrates mastery of everyday demands; developmental onset establishes maturation as the time-course. The elements must point in the same direction.

In documentation, state the evidence for each criterion separately before writing the diagnosis. This prevents a report from using one broad phrase, such as "poor functioning," to conceal which element has actually been demonstrated. It also makes the reasoning reviewable when informants disagree, records are sparse, or testing conditions were imperfect.

3.5 ICD diagnostic framework

ICD-11 groups the condition under **6A00 Disorders of intellectual development**. It describes the construct as a **metasyndrome** occurring in the developmental period: a clinically useful reminder that the diagnosis identifies a pattern of developmental functional impairment, not a single aetiology. The aetiological formulation must therefore remain open and be pursued separately when indicated.

ICD specifies three essential features: significant limitation in intellectual functioning, significant limitation in adaptive behaviour, and onset during the developmental period. Its intellectual-functioning feature explicitly includes perceptual reasoning, working memory, processing speed and verbal comprehension. These named areas should not be substituted for the DSM Criterion A list, which is framed differently. The two systems overlap clinically but phrase their intellectual evidence in distinct ways.

DIAGNOSTIC QUESTION	DSM FORMULATION	ICD FORMULATION
Intellectual limitation	Deficits in intellectual functions, supported by clinical assessment and individual standardised testing.	Significant limitation in intellectual functioning, including the specified cognitive areas.
Functional consequence	Adaptive deficits that compromise expected independence and social responsibility.	Significant limitation in adaptive behaviour.
Time-course	Intellectual and adaptive deficits begin during development.	Both limitations have developmental onset.
Use of IQ	Interpret in clinical context; never use as the sole basis.	Treat as a proxy for intellectual limitation, alongside comprehensive adaptive evaluation.

ICD indicates significant limitation in intellectual functioning at approximately **two or more standard deviations below the mean, or below the 2.3rd percentile**. It uses the same statistical convention for a total adaptive-behaviour score. These thresholds describe the boundary with expected functioning; they do not remove the requirement for comprehensive clinical interpretation.

3.6 Assessment at the boundary: reliability, culture and unavailable tests

Both systems reject the seductive but unsafe idea that IQ alone separates disorder from normality. ICD states that IQ is a proxy measure rather than an isolated diagnostic requirement at this boundary, and diagnosis must include a comprehensive evaluation of adaptive behaviour. This protects against the false certainty produced when a numerical score is detached from language, culture, education, health, developmental opportunity and everyday functioning.

Scores may vary substantially across development. ICD consequently advises testing on repeated occasions across the developmental trajectory when a reliable estimate is required. A child can appear to meet diagnostic requirements at one assessment and not another; the clinician should interpret change before declaring either recovery or error. **Reassessment** is particularly valuable when the initial assessment was constrained by unfamiliarity, illness, poor engagement, communication difficulty, or an unstable environment.

Where appropriately normed standardised tests are unavailable, ICD places greater weight on **clinical judgement**, including behavioural indicators supplied in its diagnostic guidance. This is highly relevant to heterogeneous linguistic and educational contexts. Clinical judgement here means explicit, accountable reasoning from history, observation and functional evidence. It does not mean replacing assessment with a casual impression, assuming disability from poverty, or assigning a label merely because a formal test cannot be obtained.

- Record why a particular test result is credible, limited, or unavailable.
- State whose account supports developmental onset and what setting each adaptive observation represents.

- Describe the language and sociocultural context relevant to interpretation, rather than treating them as background decoration.

When standardised testing is difficult or impossible, such as in the presence of sensory impairment or severe problem behaviour, DSM permits the diagnosis of **unspecified intellectual developmental disorder**. This is a category for genuine diagnostic limitation, not a shortcut around careful clinical work. The report should make clear what cannot currently be established, what evidence is present, and what reassessment could clarify.

3.7 Terminology, prevalence and the distinction from service counts

Terminology has changed because diagnostic language affects how people are understood in families, services and law. DSM uses intellectual developmental disorder to align with ICD-11, while retaining intellectual disability because it remains widely used by professionals, advocacy groups and the public. In a clinical record, use respectful current terminology and describe the person's functioning and support context rather than allowing a historical label to stand in for a formulation.

Prevalence needs a denominator before it can be interpreted. DSM estimates overall general-population prevalence at approximately **10 per 1,000**, but figures derived from registers, schools or service rosters answer a different question from population studies. The concept of **administrative prevalence** concerns the people for whom a community would need services, and in practice often those whose needs are known to providers. It is useful for planning, but it should not be presented as an intrinsic biological rate.

10 per 1,000	16 per 1,000	9 per 1,000
DSM OVERALL ESTIMATE	DSM MIDDLE-INCOME ESTIMATE	DSM HIGH-INCOME ESTIMATE

IN INDIA

Reported Indian point prevalence lies between 1/1,000 and 32/1,000. A cited meta-analytic estimate is 10.37 per 1,000. In high-income settings, reported figures are 9.2 per 1,000 across all ages, 18.3 per 1,000 in child and young-person populations, and 4.9 per 1,000 in adult-only populations. The breadth of such estimates, including the reported Indian range, should make the reader ask how cases were defined, what population was sampled, and whether ascertainment relied on services, schools or household assessment. This is not pedantry. A prevalence figure used without its case definition and denominator can distort planning, obscure unmet need, or create a misleading comparison between settings.

Must remember: a low IQ score can support the diagnosis, but intellectual disability requires a developmental history and demonstrable adaptive limitation as well.

3.8 A defensible diagnostic conclusion

A high-quality conclusion names the diagnostic framework used, then shows how the evidence meets each required element. It identifies the source and limitations of intellectual assessment, gives a function-based account of adaptive impairment in relevant settings, and states the

developmental evidence. It should also acknowledge uncertainty when the evidence is incomplete. This is more clinically useful than a conclusion that merely reproduces a test result.

Do not use the diagnostic threshold to assign a severity category. The grading of intellectual disability is based on adaptive functioning and is addressed in the severity classification section. Do not confuse the diagnosis with borderline intellectual functioning, which is considered in the differentiation section. If the broader assessment has not yet been completed, say what is provisional and what collateral, functional assessment or repeat testing is still needed. The full assessment framework is covered in the Child psychiatry: assessment, treatment, services and protection notes.

4 · Severity classification (mild, moderate, severe, profound) by IQ and adaptive functioning

Severity is a description of lived support need. In intellectual disability, a severity label should help the clinician communicate what is difficult in everyday life, anticipate the breadth of assistance required, and organise a realistic plan with the person and family. It is not a shorthand for worth, potential, behaviour, or prognosis. The same label can conceal very different strengths, vulnerabilities, opportunities and barriers if it is treated as an identity rather than a clinical formulation.

The familiar grades run across **four** levels: mild, moderate, severe and profound. Their enduring examination relevance is that older systems linked them closely to IQ bands, whereas contemporary diagnostic practice grades severity through functioning in ordinary settings. Candidates need to be able to state both schemes, distinguish their purposes, and explain why the second is clinically more useful.

4	Adaptive	IQ	Supports
RECOGNISED SEVERITY GRADES	BASIS FOR CONTEMPORARY GRADING	SUPPORTING CLINICAL EVIDENCE	PRACTICAL CONSEQUENCE OF THE GRADE

4.1 What a severity classification is for

A **severity classification** is a communication tool, not a substitute for assessment. It gives a shared language for describing the extent of impairment, but the clinical account must still say what the person can do independently, what they can do with prompting or teaching, and where risk or dependence becomes important. A label that is not linked to concrete functional information has little value to the family, school, employer or care team.

Severity must therefore be used at the level of the individual. It should be informed by functioning across conceptual, social and practical domains. Conceptual functioning includes the use of knowledge in everyday demands. Social functioning concerns interpersonal judgement, communication in relationships and the ability to understand social situations. Practical functioning concerns self-care, routine, safety, work-related tasks and participation in community life. These domains may be uneven. A person may show a relative strength in one setting and

substantial dependence in another, particularly when a familiar and supportive environment masks difficulty that becomes apparent with changing demands.

The clinician's task is to translate this profile into a defensible grade without reducing the profile to the grade. Good documentation makes clear which examples support the judgement: what was directly observed, what reliable informants describe, and what the person manages when environmental scaffolding is withdrawn or new demands arise. It also separates lack of opportunity, sensory or motor limitations, language mismatch, physical illness and psychiatric symptoms from limitations attributable to intellectual disability. Otherwise severity can be overstated by deprivation or understated by unusually protective care.

RULE **Grade severity by the person's adaptive functioning, then use the IQ result as supporting clinical evidence.**

GOOD TO REMEMBER

This rule protects against a common distortion. A psychometric result can be highly informative, but it does not by itself show how the person manages money, relationships, self-care, safety, travel, routine, communication or unexpected change. Conversely, an apparently competent routine may depend on extensive family organisation and may not generalise beyond a familiar context. Severity is therefore a clinical judgement grounded in the person's actual life, not merely an arithmetic location on a test distribution.

4.2 The contemporary adaptive-functioning approach

Adaptive functioning is the organising principle of contemporary severity grading. The DSM-5-TR severity table describes each grade through conceptual, social and practical functioning rather than through an IQ column. This makes the table clinically legible: severity is not simply a description of test performance but a description of the gap between the person's abilities and the demands of their usual environment.

The reason for this shift is pragmatic as well as conceptual. Support needs are determined by what the person can and cannot accomplish safely and consistently in daily life. An IQ score contributes evidence about intellectual functioning, but it does not directly specify supervision, teaching, environmental adaptation, decision support or the degree to which another person must take responsibility for safety. In this approach, the grade predicts the nature of help that has to be planned, not a fixed ceiling on learning.

Adaptive information should be interpreted in context. The same skill may be performed independently in a familiar home and not be available in a more demanding environment. Families may compensate so effectively that important dependence is missed, while social exclusion or inadequate education may produce apparent deficits that are not a stable reflection of underlying capacity. The assessor should therefore ask about performance across settings, response to ordinary change, consistency over time and the amount of cueing, monitoring or physical assistance required. This is why collateral history is central rather than decorative.

The grading rule also prevents false precision. Two people with similar test results may differ materially in social judgement, practical safety, communication, co-occurring illness, sensory impairment, motor disability or access to support. Conversely, a person whose score is difficult to interpret because of language, behaviour or physical limitations may still have a clear adaptive profile. The clinical formulation should acknowledge uncertainty where it exists, rather than force a spurious numerical conclusion.

TRAP **Examination trap:** do not write that a contemporary severity grade is "based on IQ." State that severity is based on adaptive functioning, while IQ informs the wider assessment.

For DSM severity terminology, the crucial distinction is that the grade is decided from adaptive deficits. This is a useful sentence to carry into a long answer because it explains why the classification has clinical purpose. It also guards against a mechanical answer in which a candidate recites IQ ranges without demonstrating that they understand the modern framework.

4.3 DSM severity labels and coding language

The diagnostic labels are familiar, but they should be treated as severity specifiers attached to an individualised functional account. The codes below are useful for records and examinations. They do not replace a narrative description of the person's functioning, current supports, vulnerabilities or strengths.

The linked coding language is F70 for mild, F71 for moderate, F72 for severe, and F73 for profound. Pair the code and label with the adaptive profile and the help required in real settings. The code identifies a category; the formulation tells another clinician how to care for the person.

In clinical writing, the label should lead to action. A bare diagnosis may be sufficient for a register, but it is insufficient for care planning. The formulation should say which tasks need teaching, which require environmental adaptation, which need supervision, and which are currently managed with assistance. It should also identify how communication, choice and autonomy will be protected. Severity is most useful when it improves coordination between psychiatry, family, education, rehabilitation and community services.

Do not infer a person's adaptive profile from the label alone. Higher-grade disability can coexist with preferences, learning and meaningful participation, and lower-grade disability can be accompanied by marked risks in social judgement, exploitation or practical safety. The label alerts the team to likely breadth of need; the assessment tells the team what that need actually is.

4.4 IQ-based severity bands: their place and their limits

IQ-based severity bands belong to the older classification logic and remain examinable because they appear in legacy records, teaching material and administrative language. They are best presented as a historical mapping, then immediately qualified: a score range is not a contemporary severity judgement. The IQ construct, its interpretation and the choice of intelligence testing are addressed in the clinical assessment section; here the concern is only the mapping of an IQ result to a grade.

LEGACY GRADE	IQ BAND IN ONE REPORTED ICD-10 SCHEME	CLINICAL READING
Mild	**50-69**	Use as a historical IQ band, not as the sole current severity determinant.
Moderate	**35-49**	Interpret alongside the person's actual adaptive functioning.
Severe	**20-34**	Do not assume practical abilities from the band alone.
Profound	**below 20**	Functional observation remains indispensable, especially when testing has limits.

The legacy statement that the IQ cut-off is 70 is often quoted alongside the claim that ICD-10 categorised the grades by IQ. That statement accurately captures the older scheme, but should not be carried forward uncritically into contemporary diagnostic phrasing. The meaningful clinical question is not only where a score lies, but whether the individual can meet culture-appropriate everyday demands without assistance.

A source discrepancy deserves explicit caution. Another textbook presentation places the upper end of the mild band at 50-70, rather than at the upper boundary shown in the preceding table. This is not a reason to blend the two into an invented compromise. In an answer, name the classification framework being used, give its band accurately, and then state that contemporary severity is determined from adaptive functioning. That is safer than presenting a disputed edge value as universal fact.

The deeper lesson is that score bands create an appearance of sharp boundaries where clinical reality is continuous and context-dependent. Borderline intellectual functioning is a separate boundary concept and should not be folded casually into intellectual disability; its definition belongs in the differential-diagnosis discussion. Likewise, a test result affected by language, behaviour, fatigue, sensory limitations or unfamiliarity with the testing context should prompt careful interpretation, not an automatic severity label.

4.5 ICD-11: severity across ability and adaptive behaviour

The **ICD-11 approach** assigns severity according to the level in which the majority of the person's intellectual ability and adaptive-behaviour skills fall across conceptual, social and practical domains. This is an important refinement of single-score thinking. It accepts that a profile can be uneven, yet asks the clinician to identify the level that best represents the predominant pattern rather than choosing the most impaired isolated skill or the most successful isolated activity.

The numerical bands in ICD-11 are expressed as distance from the mean and corresponding percentile information. They should be read as psychometric context, not as a replacement for the adaptive judgement. The accompanying table is deliberately dense because the codes and

bands are examination material; in practice, the functional formulation remains the more clinically useful document.

ICD-11 GRADE	PSYCHOMETRIC BAND	CODE	KEY IMPLICATION
Mild	2-3 SD below the mean; approximately 0.1-2.3 percentile	6A00.0	Assign the grade with the adaptive profile in view.
Moderate	3-4 SD below the mean; approximately 0.003-0.1 percentile	6A00.1	Do not let the numerical band eclipse functional variation.
Severe	4 or more SD below the mean; less than approximately the 0.003rd percentile	6A00.2	Distinguish this grade through adaptive behaviour.
Profound	4 or more SD below the mean; approximately less than the 0.003rd percentile	6A00.3	Distinguish this grade through adaptive behaviour.

The apparent overlap between the highest-grade psychometric bands is deliberate and clinically instructive. Severe and profound are separated exclusively by adaptive-behaviour differences, because available standardised intelligence tests cannot reliably and validly distinguish people at the extreme lower end of intellectual functioning. This is not a defect to be repaired by making the score seem more exact. It is a recognition that the most meaningful discrimination at this level concerns what the person is able to do and the support required for comfort, safety, communication and participation.

ICD-11 also provides 6A00.4 for a provisional grade and 6A00.Z for an unspecified grade. These should be used thoughtfully when a definitive grade cannot yet be assigned, rather than guessing from incomplete information. A provisional label preserves diagnostic honesty while assessment continues. An unspecified label should prompt the clinician to record what information is missing and what will be needed to clarify the person's profile.

For longitudinal records that need a crosswalk to legacy coding, the mappings are 6A00.0 to F70, 6A00.1 to F71, 6A00.2 to F72, 6A00.3 to F73, 6A00.4 to F78, and 6A00.Z to F79. A crosswalk translates administrative language; it does not change the adaptive-functioning basis on which the current clinical grade is reached.

4.6 Reconciling discordant evidence in Indian practice

Apparent disagreement between intellectual testing and social-maturity findings is not unusual. It should not be treated as a nuisance to be averaged away. First check whether either result is invalid or unrepresentative because of communication barriers, behaviour, physical limitations, fatigue, unfamiliarity or an unsuitable testing context. Then return to the central clinical question: how does the person meet everyday demands in their own cultural and environmental setting?

IN INDIA

Where IQ and social-maturity results indicate different grades, the Indian guideline directs the clinician to decide in favour of the **social-maturity result**. The rationale is clinically coherent: social maturity reflects the degree to which the person meets culture-appropriate demands of daily life and generally better represents severity in ordinary circumstances. This is not an invitation to dismiss intellectual assessment. It is a reminder that the outcome of classification should describe functioning, not reward a deceptively precise test number.

The assessment method, the score it yields and its limitations belong in the adaptive-behaviour assessment section. Statutory disability quantification is a separate legal process and must not be confused with clinical severity or with adaptive functioning. In a clinical report, make the distinction explicit: describe the diagnostic severity formulation, describe the evidence used to reach it, and deal with certification through the relevant disability-certification pathway.

- Check that the intellectual and functional information reflects the person's usual, not merely best or worst, performance.
- Explain discordance rather than concealing it behind a single label.
- Prefer the evidence that best captures everyday functioning when a severity judgement is required.
- Record strengths, communication preferences and environmental supports alongside limitations.

4.7 From severity label to support profile

A grade becomes clinically useful only when it changes the plan. It should lead the team to ask what support is needed to enable learning, choice, health care, family participation, education, work, relationships and safety. The answer should be revisited as circumstances change. Severity is relatively stable as a description of pervasive need, but support can be improved through accessible communication, structured teaching, environmental adaptation and respect for the person's established competencies.

The **AAIDD** classification links the grades to a **profile of supports** instead, and is multidimensional rather than categorical. Its four intensities of support, in ascending order, are **intermittent, limited, extensive and pervasive**. Two qualifications keep that list honest. It belongs to the ninth edition of the AAIDD system and was not carried into the later editions, which found it lacked the standardised data to support graded intensities, so it is a historical scheme that remains examinable rather than a current classification. And it is a description of what a person needs, not a fifth way of grading them: the four intensities are often lined up against mild, moderate, severe and profound, but the mapping is a convenience and the whole point of a supports model is that two people at the same severity grade may need very different support. Reproduce the four-word list as written: one widely used textbook prints extreme supports at the severe tier, but the term in general use, and the one an examiner expects, is extensive. These words should not be used as a rigid prescription for any one person. They are a

reminder to specify intensity, consistency and context of help rather than to leave the plan at the level of a diagnostic adjective.

A support profile is also more humane than deterministic language. It turns the clinical focus from what the person lacks to the conditions under which the person can participate. It permits planning around autonomy as well as risk: who should provide information in an accessible form, how decisions can be supported, what routines promote competence, what supervision is proportionate, and how family burden can be recognised without erasing the person's voice.

MNEMONIC

GRADE

Ground the decision in daily functioning; Reconcile discrepant evidence; Avoid IQ-only classification; Describe supports; Explain the functional basis in the record.

The defensible severity statement is the one that explains the person's adaptive functioning and consequent support needs, not merely the one that reports an IQ range.

4.8 A disciplined answer and documentation approach

For a short-answer question, begin with the names of the grades, state the modern principle that adaptive functioning determines severity, and then contrast this with the legacy IQ-linked scheme. If asked for a classification table, present the requested framework cleanly. If the question is clinical, move beyond the table: identify the adaptive domains affected, state the basis for the grade, and outline the supports that follow. This sequence shows the examiner that the candidate understands both taxonomy and clinical reasoning.

For a case record, avoid language that makes the severity label sound final in isolation. State the diagnosis, the current grade, the evidence from functioning, the degree and type of assistance required, relevant contextual modifiers, and the plan to address identified needs. Be cautious when information comes only from one informant or one setting. A carefully qualified assessment is stronger than a confident but unsupported label.

The same discipline protects against two opposite errors. One is overmedicalisation, in which a score becomes the whole person. The other is under-recognition, in which a person's dependence is minimised because they have learned familiar routines or because relatives quietly perform the demanding parts of daily life. Both errors lead to mismatched care. Severity classification is valuable precisely because, done well, it keeps clinical language anchored to the reality of support.

5 · Clinical assessment and IQ testing in ID

Assessment is a clinical act, not a score-collecting exercise. In suspected intellectual disability, the psychiatrist has to decide whether an observed difficulty represents a broad limitation in intellectual functioning, a barrier to showing ability in the assessment setting, a circumscribed learning problem, or a mixture of these. The task is therefore to obtain a valid account of how the

person reasons and learns, and then to set that account beside history, observation, collateral information and adaptive-behaviour assessment. The numerical result matters, but it is never self-interpreting.

The marks in this area are usually won by showing disciplined interpretation. A candidate should be able to explain what an IQ score represents, why it is reported as an interval rather than treated as an infallible point, how a poor-quality administration can mislead, and why a patterned profile may be more clinically useful than an overall score. The assessment must also stay within its proper boundary: severity grading and the formal measurement of adaptive behaviour are separate tasks, addressed elsewhere in this chapter.

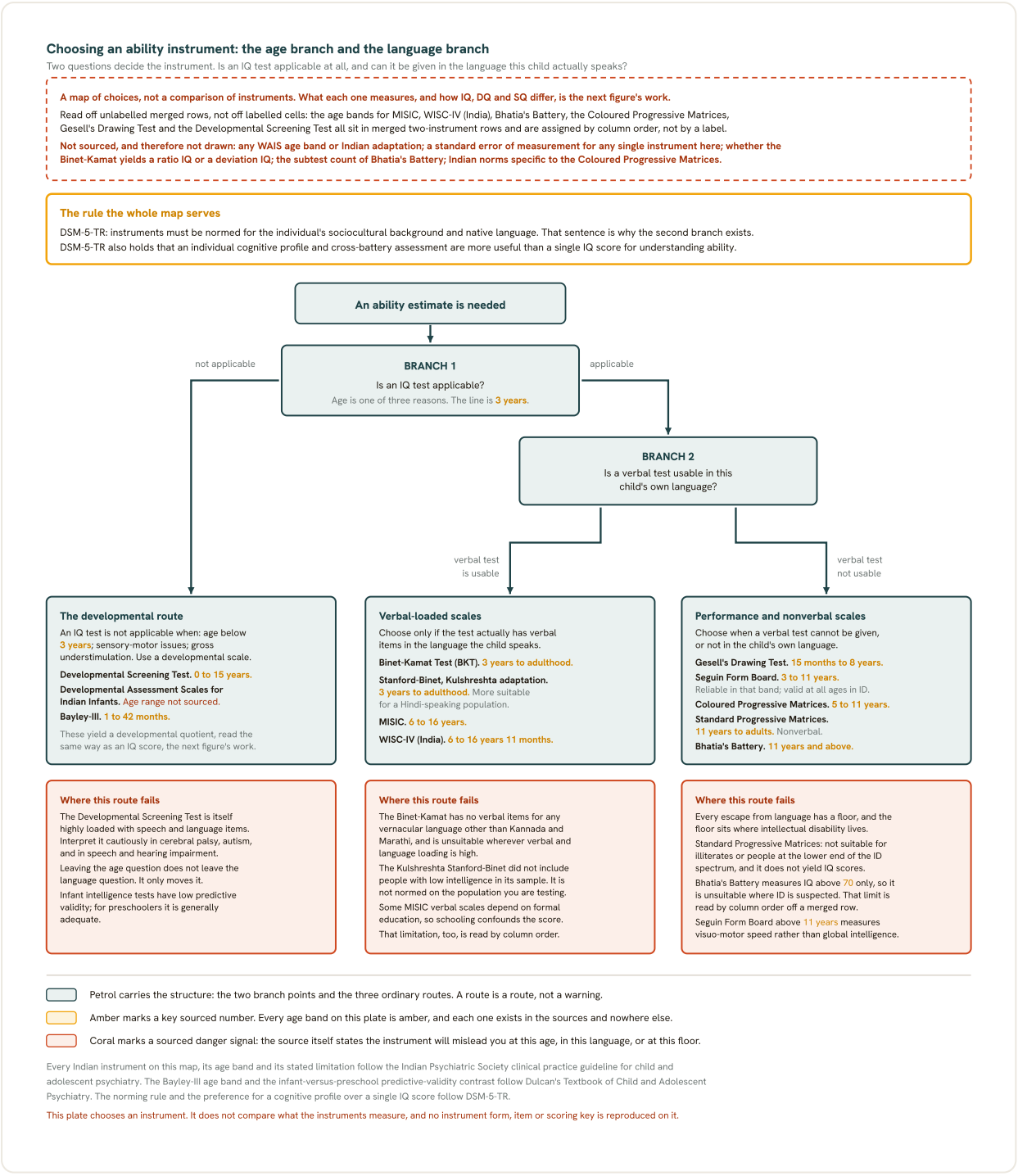


Fig 21.3 Choosing an ability instrument by age and by language. Two branch points, not a catalogue: whether an IQ test applies at all, and whether a verbal test can be given in the language the child actually speaks. Each of the three routes is named with its instruments and their sourced age bands, and each is drawn with the limit its own source states, because every escape from the language problem has a floor and the floor sits where intellectual disability lives.

5.1 The clinical question before testing

Begin with the referral question. “What is the IQ?” is usually an impoverished question. The useful question is: what decision will a careful estimate of intellectual functioning help the team make? The answer may concern the nature of a broad learning difficulty, the credibility of an apparent decline, the meaning of an uneven school record, the person’s capacity to engage with an explanation, or the supports that should be considered. Stating the question first protects the assessment from becoming an administrative ritual.

Intelligence can be understood clinically as a broad capacity to act with purpose, reason coherently and deal effectively with the environment. An intelligence test samples that capacity under defined conditions; it does not sample every ability a person possesses, every opportunity they have had, or every demand made by ordinary life. The psychiatrist should therefore ask what the test performance means in this person's lived context rather than asking the score to speak beyond its remit.

The first interview establishes whether testing is likely to answer the question. Clarify the presenting concern in the informant's own language, the course of learning and functioning, the educational and occupational setting, sensory or motor barriers, communication style, behaviour during unfamiliar tasks, and the person's current mental and physical state. Review available records for consistency rather than assuming that a label in a previous document is a conclusion. A recent deterioration in performance, acute distress, reduced alertness or inability to engage may change the meaning of a low obtained score. The full developmental history and child psychiatric assessment framework belong in the *Child psychiatry: assessment, treatment, services and protection notes*; here, their relevance is that they determine whether a test session is interpretable.

RULE **Use psychometry to refine clinical judgment, not to replace it.** A score becomes useful only when the referral question, the testing conditions and the person's real-world functioning have been considered together.

- Formulate the decision that testing is meant to inform before selecting the assessment.
- Establish whether communication, sensory, motor, emotional or situational factors could distort performance.
- Obtain collateral evidence sufficient to judge whether the test result fits the longitudinal clinical picture.
- Keep the estimation of intellectual functioning distinct from adaptive-behaviour measurement and from severity grading.

5.2 What an IQ score is, and what it is not

An IQ is a standardised comparison of an individual's performance with that of the reference group used by the test. Its clinical value lies in making the comparison explicit and reproducible. It is not a direct measure of worth, potential, character, family effort or entitlement to care. Nor does an overall score reveal the route by which a person reached it. People with a similar overall result may have quite different cognitive strengths, educational experiences, communication demands and support needs.

Older teaching may describe **ratio IQ** using the expression IQ = (mental age / chronological age) x 100. This formulation is historically useful because it explains the term "IQ", but it should not be casually substituted for the standard-score framework used in contemporary interpretation. A clinician should state which metric the report provides rather than sliding between different meanings of the same abbreviation.

For the test metric assumed when deriving the diagnostic convention, the mean is 100 and the standard deviation is 15. The phrase “standard deviation” expresses the spread of scores around the mean in the reference population. It allows a clinician to describe the distance of a result from the population average without converting that distance into a moral or deterministic statement about the person.

100	15	≥2 SD below population mean	65-75
ASSUMED TEST MEAN	ASSUMED STANDARD DEVIATION	CLINICAL CONVENTION	SCORE INTERVAL REQUIRING INTERPRETATION

The intellectual-functioning convention lies **approximately 2 standard deviations or more below the population mean**. This is a convention for interpreting a result in relation to a population distribution, not permission to diagnose from arithmetic alone. The clinician still asks whether the obtained result is valid, whether the reference group is appropriate, and whether the person’s presentation is consistent with the assessment as a whole.

A report should name the metric before interpreting it. This is especially important because a composite score and a subtest score are not interchangeable. The overall number is a summary; it should not erase clinically meaningful variation within the assessment. Equally, a striking isolated weakness is not automatically evidence of a broad intellectual impairment. Interpretation requires pattern recognition, corroboration and restraint.

5.3 Measurement error and the discipline of reporting a range

An obtained score is an estimate. Every psychometric result contains uncertainty arising from the instrument, the testing occasion and the person’s performance on that occasion. The relevant question is therefore not “what is the person’s true IQ?” but “what range of intellectual performance is reasonably supported by this assessment, given its quality and context?” This language is more accurate and prevents a falsely precise clinical conclusion.

The generally applicable margin around an obtained full-scale score is **plus or minus 5 IQ points**. Thus, once measurement error is considered, the score interval associated with the intellectual-functioning criterion is **65-75**. The range must be read as an interval requiring clinical interpretation, not as a new single cut-off and not as a device for negotiating a preferred label.

GOOD TO REMEMBER

A result near the relevant convention calls for particularly careful attention to the quality of administration, the person’s engagement, collateral evidence and adaptive-behaviour assessment. An isolated point score does not settle a complex clinical question: explain how the assessment was conducted and what may have affected it.

TRAP **Examination trap:** treating an IQ result as exact. A technically sound answer states the obtained score, acknowledges measurement error, interprets the interval, and then integrates the wider assessment.

INTERPRETIVE QUESTION	WHAT THE ASSESSMENT CAN CONTRIBUTE	WHAT STILL REQUIRES CLINICAL JUDGMENT
Is the score low?	It locates performance relative to relevant norms.	Whether the administration and norms make that comparison valid.
Is the result stable enough to use?	It provides an estimate with a stated margin of uncertainty.	Whether state factors or testing conditions make the estimate misleading.
Is there a broad difficulty?	It summarises performance across sampled tasks.	Whether the pattern fits history and everyday functioning.
Does a single total tell the whole story?	It offers a convenient summary score.	Whether uneven abilities make the profile more informative than the summary.
What support is needed?	It may identify cognitive demands likely to be difficult.	Adaptive-behaviour assessment and the real-world support context.

5.4 Selecting and conducting a valid assessment

Choice of assessment follows the person, not the convenience of the clinic. Select an individually administered, appropriately standardised measure that can address the referral question and can be administered meaningfully to the person in front of you. Language, communication access, sensory and motor capacity, familiarity with formal testing, educational opportunity and current state all affect the evidentiary weight that can be placed on an obtained result.

Norming for sociocultural background and native language is a requirement, not a courtesy. A score derived from unsuitable norms risks measuring distance from the test's expected experience rather than the intellectual ability the clinician intends to estimate. This is particularly important when the assessment language is not the person's strongest language or when the testing situation makes culturally specific knowledge carry disproportionate weight. The report should make any limitation transparent rather than concealing it behind a number.

Standardisation means that the conditions of administration, scoring and comparison are controlled as far as possible. In clinical practice, that does not mean rigidly ignoring a person's needs. It means recording adaptations, interruptions and barriers, then judging whether they preserve or weaken the interpretability of the score. Rapport, a quiet setting, comprehensible instructions and adequate opportunity to respond are not cosmetic features. They are part of the validity of the encounter.

Observe the process as carefully as the final result. Does the person understand the task? Do they persist, become overwhelmed, guess rapidly, seek repeated reassurance, lose the instruction, or show a discrepancy between verbal explanation and non-verbal problem solving? Such

observations can clarify a profile, expose a threat to validity or indicate the need to defer a conclusion. They should be documented in clinically intelligible language, not reduced to a vague statement that the person was "cooperative".

Assessment should be paced to protect effort without coaching the answer. The examiner may clarify the task only within standardised rules, attend to physical comfort, and recognise when fatigue or distress has made further performance unrepresentative. If a session is not interpretable, the responsible clinical conclusion may be that the assessment has not answered the question yet. A forced total score is not more scientific than an honest limitation.

- Explain the purpose of the assessment in language the person and family can understand.
- Record the assessment language, relevant accommodations and any barriers to participation.
- Describe behavioural observations that bear on validity, rather than using generic reassurance.
- State clearly when a result should be treated as provisional or when re-assessment is needed.

5.5 Composite scores, subtests and cognitive profiles

Do not confuse the ruler with its markings. A composite or index score uses a different metric from an individual subtest scaled score. **Wechsler composite and index scores: mean 100, SD 15**; **Wechsler subtest scaled scores: mean 10, SD 3**. Mixing these metrics is a basic interpretive error: a subtest value should not be read as though it were an IQ, and a composite should not be treated as though it describes a single task.

A **cognitive profile** is the pattern of relative strengths and weaknesses across the assessment, interpreted cautiously in context. It directs attention to how the person approaches demands rather than merely where the overall score falls. Profiles are most useful when they explain an observed clinical discrepancy, guide further assessment or suggest a practical way to communicate and support learning. They are least useful when they are turned into speculative labels unsupported by history or observation.

The preference for individual cognitive profiles over a single IQ is reflected in **cross-battery intellectual assessment**, which is more useful for understanding intellectual abilities than a single overall score alone. This does not mean that every patient needs an elaborate battery. It means that the breadth of assessment should match the clinical ambiguity. Where the total score and the observed person tell the same coherent story, a focused assessment may suffice. Where they diverge, the divergence is data, not an inconvenience to be averaged away.

Large differences across tasks require interpretation before the full-scale result is treated as a stable global estimate. Highly discrepant individual subtest scores may make an overall IQ score invalid. The key word is "may": discrepancy prompts clinical scrutiny rather than an automatic rejection of the entire assessment. Consider whether the pattern is understandable from language demands, attention, motivation, sensory or motor factors, emotional state, educational experience or a specific cognitive weakness. The conclusion should describe what is known and what remains uncertain.

This distinction prevents a common diagnostic shortcut. Specific learning disorder may show a verbal-performance discrepancy, usually with performance IQ higher, and an ACID pattern of relative weakness in Arithmetic, Coding, Information and Digit-span. It may also show weaknesses in verbal comprehension, working memory and processing speed. These findings concern an uneven learning profile; they must not be converted mechanically into a severity statement about intellectual disability. The broader formulation belongs with the educational and communication assessment described in the *ADHD and specific learning/communication disorders notes*.

5.6 Threats to validity and misleadingly high or low results

Flynn effect refers to the risk of spuriously high IQ scores when test norms are out of date. It is a norms problem, not a personal change in ability. **Practice effects** are different: repeated testing can yield spuriously high scores because the person has learned from prior exposure. The distinction matters because the remedy is different. In one situation the concern is the currency of the reference norms; in the other it is the interpretive impact of repeated familiarity with the material.

Before accepting an unexpectedly high or low score, ask whether the test was suitable and whether the score was generated in a valid way. A brief intelligence screening test or group test may produce an invalid IQ score. Such tools can be useful for triage or as one source of information, but they do not automatically carry the evidentiary weight of a comprehensive individual assessment. The report should identify the method used and resist upgrading a screen into a definitive estimate simply because it has yielded a number.

Other validity threats are often visible in the room. A person who cannot hear or understand the instruction, whose motor difficulty prevents the intended response, or whose distress and fatigue collapse effort may obtain a result that reflects the barrier more than the target ability. The same caution applies when language mismatch or unfamiliarity with the testing format changes what is being sampled. The clinical response is not to “correct” the score by intuition. It is to document the limitation, seek appropriate assessment conditions and integrate other evidence.

Validity is not an all-or-none property. Some results provide useful minimum estimates, some are broadly representative but qualified, and some should not be interpreted as an IQ estimate at all. This graded approach is more honest than either rejecting every imperfect assessment or accepting every printed total. It also makes the report usable to colleagues: they need to know how much confidence to place in the result and why.

> An IQ score is clinically meaningful only when the test, norms, administration and interpretation are all appropriate for the individual.

5.7 Integrating and communicating the result

The final account should read like a clinical formulation, not a laboratory slip. Start with the reason for referral and identify the assessment method and conditions. State the obtained result as an estimate, include its uncertainty where relevant, describe important profile findings, and

explain the factors that strengthen or limit confidence. Then integrate this with the history, observation, collateral information and adaptive-behaviour assessment. This sequence allows another clinician to see how the conclusion was reached and where its boundaries lie.

Families commonly hear an IQ result as a verdict. The psychiatrist's language can either amplify that harm or restore accuracy. Explain that the assessment describes performance on a defined set of tasks compared with relevant norms. Discuss strengths as well as difficulties, avoid presenting a score as a ceiling on development, and translate findings into practical implications. Do not use technical uncertainty as evasiveness: it can be explained plainly as the ordinary fact that a single testing occasion estimates rather than perfectly captures ability.

When a low score appears inconsistent with everyday functioning, do not decide in advance which source is wrong. Re-examine the referral question, test validity, language and norming, profile variability, collateral quality and the possibility that a different condition is influencing performance. When everyday difficulties seem greater than the score would predict, the same discipline applies. Intelligence testing and adaptive-behaviour assessment answer related but non-identical questions; neither should be used as a shortcut for the other.

Finally, communicate the scope of the opinion. A psychometric assessment may inform diagnosis and care planning, but it does not replace medical evaluation, aetiological work-up or adaptive-behaviour measurement. The laboratory investigation of possible causes is addressed in the etiological work-up section of this chapter, and formal adaptive-behaviour instruments are addressed in the adaptive behaviour assessment section. Keeping these strands separate while integrating their conclusions is the hallmark of a defensible intellectual-disability assessment.

5.8 Choosing intelligence instruments in Indian practice

IN INDIA

Selection is a clinical discrimination: fit the instrument to age, language and schooling, not departmental familiarity. A verbal age scale may be technically well administered yet answer the wrong question when the child is being tested outside the language of its norms. Conversely, a performance measure is not a lesser substitute for a "real" intelligence test when language is the obstacle: it samples ability through a different route. Before choosing a battery, decide whether the task requires a broad verbal and performance profile, a brief performance estimate, or a nonverbal reasoning measure, and be explicit about what the chosen instrument actually reports.

RULE **Choose the instrument by construct, age and language before choosing it by familiarity.** The score is interpretable only when the test's norms and task demands fit the child in front of you.

INSTRUMENT	AGE BAND AND FORM	WHAT IT YIELDS	WHEN IT IS THE BETTER FIT - AND ITS LIMIT
Binet-Kamat Test of Intelligence	**3 years to adulthood**; a **predominantly verbal age scale**.	**Age-scale estimate**, not a language-free performance measure.	Use only when the verbal demands and language are appropriate. Its verbal items are unavailable in vernacular languages other than **Kannada and Marathi**.
Stanford-Binet, Kulshreshta Indian adaptation	**3 years to adulthood**; **predominantly verbal age scale**.	**Age-scale assessment**.	It is more suitable for a **Hindi-speaking population**; its norms did not include people with low intelligence.
Malin's Intelligence Scale for Indian Children	**6-16 years**; **Indian adaptation of the original Wechsler child scale**.	**Performance IQ, Verbal IQ and Full-scale IQ**.	Useful when a Wechsler-style profile is needed, but some verbal scales depend on **formal education**.
Bhatia's Battery of Intelligence	**standardised on Indian boys aged 11 to 16**, though the Indian guideline prints the range as 11 years and above; **indigenous performance battery**.	**Performance-based intelligence assessment**.	Choose it when verbal load is the principal barrier. It measures IQ only **above 70**, so it is unsuitable for establishing lower-range intellectual functioning.
Seguin Form Board	Reliable at **3-11 years**; valid across age groups of people with ID.	**Performance test and quick measure of general intelligence**.	A valuable low-verbal-demand option. Above **11 years**, it increasingly reflects visuo-motor speed rather than global intelligence.
Raven's Standard Progressive Matrices	**11 years to adulthood**; **nonverbal matrices**.	**Does not yield an IQ score**.	Assesses general ability through form comparison and analytical reasoning. It is not suitable for **illiterates or people at the lower end of the ID spectrum**.
Raven's Coloured Progressive Matrices	**5-11 years**.	**Percentile ranks**, not an IQ.	Use when a developmentally younger child needs a matrix task, while reporting the actual metric rather than relabelling it as IQ.

INSTRUMENT	AGE BAND AND FORM	WHAT IT YIELDS	WHEN IT IS THE BETTER FIT - AND ITS LIMIT
WISC-IV India	**6 years to 16 years 11 months**; a full child battery.	A full-scale score with an index profile, reported in this edition as **verbal comprehension, working memory and processing speed** alongside perceptual reasoning.	Use where age and language permit a full child battery. Do not describe this edition's output as a verbal/performance split: that is the older Wechsler structure, and some Indian teaching sources still print it. Do not wrongly transfer Bhatia's stated **above-70 floor** to WISC-IV India.

The **Binet-Kamat** and the **Kulshreshta Stanford-Binet adaptation** are the classic age-scale choices, but both are predominantly verbal. That is their clinical cost. If the child cannot comprehend the assessment language with the same ease as the norm group, a low result may partly index linguistic distance, schooling or test familiarity rather than the intended construct. Binet-Kamat's language restriction makes this concern concrete. The Hindi orientation of the Kulshreshta adaptation is similarly a selection rule, not a minor reporting footnote. Neither should be used merely because it is available in the department.

MISIC is often the most useful familiar-scale option for a school-age child because it derives from the Wechsler child scale and separates performance, verbal and full-scale results. The separation matters clinically: an uneven verbal-performance pattern can signal that the overall total conceals how the child is managing the tasks. It does not remove the education dependence of some verbal scales. A child with limited formal schooling should not be made to look globally less able because a verbally loaded test has been treated as culture-free.

The performance instruments address this problem directly. **Bhatia's Battery** includes many indigenous subscales and is the performance route for an older child, within its norms. Two limits govern its use and both are examinable. Its stated floor prevents it from being the decisive measure when lower-range intellectual functioning is suspected. And although the Indian guideline prints its range as 11 years and above, the battery was standardised on Indian boys aged 11 to 16 in the 1950s, so reading an adult against it applies sixteen-year-old norms to a grown patient. That is an extrapolation beyond the instrument, not normative adult use, and the open-ended phrasing of the guideline is exactly what invites it. The **Seguin Form Board** is a quick performance measure, useful where verbal demands obstruct assessment, but its changing emphasis after the relevant age means that fast visuo-motor work must not be misread as a global estimate of intelligence.

TRAP **"Raven's gives an IQ" is wrong. Standard Progressive Matrices does not output an IQ.** Do not convert a percentile or a matrix result into an IQ merely because the test is familiar.

Raven's Standard Progressive Matrices has Indian norms and is culture-fair to a large extent, but that does not make it universally suitable or make it an intelligence quotient. It evaluates form comparison and analytical reasoning, and its limits in illiteracy and more severe intellectual impairment are clinically important. **Coloured Progressive Matrices** supplies percentile ranks for younger children. State the result in the metric provided. Finally, **WISC-IV India** offers a full child battery across its specified age range, reporting a full-scale score with an index profile rather than the verbal/performance split of the older Wechsler editions. It may be the appropriate comprehensive choice only after language, schooling and tolerance of formal testing have been considered. Selection is a defensible match between construct and patient.

6 · Adaptive behaviour assessment (Vineland, DST, VSMS, Gesell)

The question is not merely what the person can do in a testing room. Adaptive-behaviour assessment asks how a person manages the ordinary demands of life in the setting in which they live. It therefore supplies information that a performance score cannot supply: whether an observed difficulty is translated into dependence in self-care, communication, social participation, safety, or practical daily routines. For intellectual disability, this is clinically decisive because support planning must follow real-world functioning, not the elegance of a single test result.

The instruments often grouped together in examination answers do not measure one thing. A developmental screen estimates developmental progress; a social-maturity scale yields a social quotient; an adaptive-behaviour scale documents functioning through an informant; and a drawing screen provides only a screening impression. The candidate who first identifies the construct, the respondent, and the output is much less likely to call a developmental quotient an IQ score or to use a social-maturity result as though it were a statutory disability percentage.

0-15	0-15	0-90
DST AGE SPAN, YEARS	VSMS AGE SPAN, YEARS	VINELAND-3 AGE SPAN, YEARS

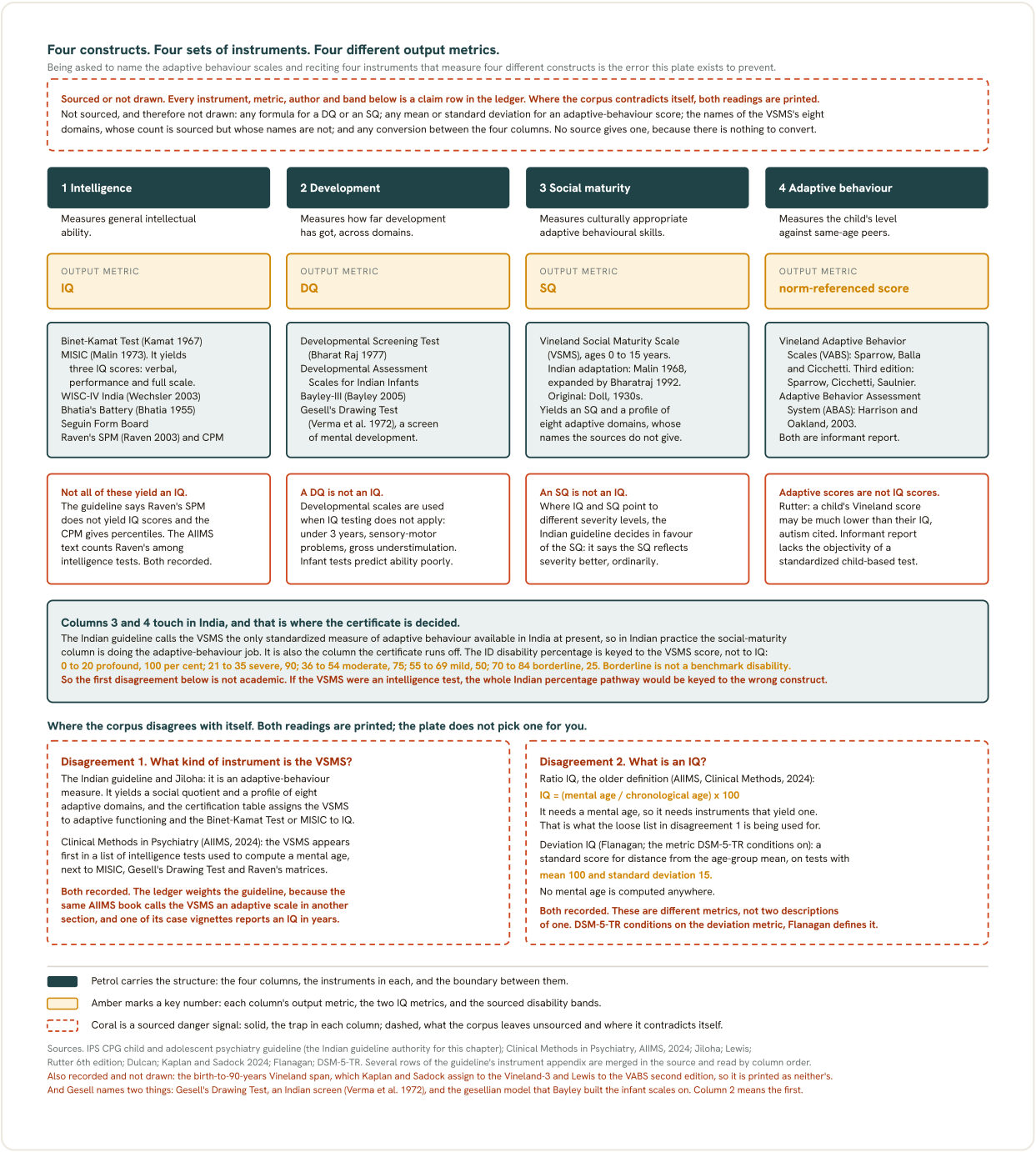


Fig 21.4 **Four constructs, four instruments.** Intelligence, development, social maturity and adaptive behaviour are four separate constructs with four separate instrument sets and four different output metrics, and no score in one column converts into a score in another. The plate also prints the two places the corpus contradicts itself, rather than resolving them: whether the VSMS is an intelligence test or the adaptive-behaviour instrument, and whether an IQ is a ratio of mental to chronological age or a deviation score with a mean of 100 and a standard deviation of 15. Choosing an ability instrument by age and language is the previous plate's job; this one is only about what each column measures and what it reports.

6.1 Begin with the construct, not the familiar name

Adaptive-behaviour measurement is a structured account of habitual functioning. It is not an abbreviated intelligence test. The best information comes from a respondent who has seen the person across ordinary situations and can distinguish what is done independently, what is done with prompting, and what is not yet part of the person's routine. The assessor then integrates this account with direct observation, history, opportunities for learning, physical or sensory

limitations, and the quality of the informant's knowledge. A score may be useful, but the clinical formulation is stronger when it retains the profile behind the score.

This distinction matters especially when opportunity and capacity have been confused. A person may fail to perform a task because it has never been taught, because the setting does not permit it, because the task is inaccessible, or because of a primary developmental limitation. Conversely, an apparently competent answer from a family member may describe what the family does for the person rather than what the person can actually do. Adaptive assessment should therefore examine typical performance, not an idealised report and not a one-off best performance in clinic.

RULE **Rule:** treat an adaptive score as an organised summary of everyday functioning, then return to the underlying profile whenever the score and the clinical picture do not fit.

The terminology itself provides a useful safeguard. **Developmental quotient** is the output of developmental testing and is interpreted in the same metric logic as an IQ score, but it describes developmental assessment rather than a conventional intelligence battery. **Social quotient** is the output of the Indian social-maturity scale. Neither label should be casually substituted for the other. A developmental result, a social-maturity result, and a formal ability result answer overlapping but non-identical clinical questions.

CLINICAL QUESTION	APPROPRIATE CONSTRUCT	ILLUSTRATIVE OUTPUT	WHAT IT MUST NOT BE CALLED
Has the child's overall development progressed as expected?	**Developmental screening**	DQ	An IQ result
How does the person manage socially and practically in their milieu?	Social maturity and adaptive profile	SQ with domain profile	A developmental quotient
What is the person's functioning across adaptive domains by informant report?	Adaptive behaviour	Adaptive profile	A direct performance battery
Is a rapid developmental screen needed before fuller assessment?	Performance screening	Screening impression	A definitive diagnostic quotient

6.2 The Indian social-maturity scale in practice

The **Vineland Social Maturity Scale**, in its Indian adaptation, is the central practical instrument for this purpose. The adaptation is attributed to Malin and was later expanded by Bharatraj. It assesses culturally appropriate adaptive behavioural skills across **eight** important domains and yields an SQ. The domain profile is often more clinically useful than a compressed global statement because it shows where the person is independent, where cueing is enough, and where sustained assistance is needed.

IN INDIA

In the cited guideline, the VSMS has a unique place among standardised adaptive-behaviour measures in India. This gives it an established role in Indian assessment, without implying that every clinical population, language group, or contemporary setting is equally represented. Document the sociocultural context of responses: functioning is enacted in a household, school, neighbourhood, and set of expectations.

GOOD TO REMEMBER

Administration is not a mechanical interview. Identify who knows the person's usual functioning best, and ask for concrete examples. Caregiver disagreement may reveal different opportunities, supervision, or settings, or an informant who has not observed the task. Test implausibly high or low responses with observation and collateral information rather than forcing premature consensus.

- Ask what the person does on an ordinary occasion, not what they have done once under exceptional help.
- Clarify whether initiation, sequencing, supervision, and completion are independent.
- Separate lack of opportunity from inability, while recording both as clinically relevant.
- Interpret a scattered profile as a starting point for formulation and support planning, not as a nuisance to be averaged away.

The VSMS is an adaptive-behaviour instrument: it yields a social quotient plus a profile of 8 domains of adaptive behaviour, never an IQ. Naming it, the DST or Gesell among intelligence tests is the single commonest error in this topic, and where a social quotient and an IQ disagree the severity decision is made on adaptive grounds, as the severity discussion sets out.

This is a measurement rule, not a licence to ignore contradictory evidence. Discordance should prompt a review of language demands, test engagement, motor or sensory barriers, educational opportunity, and the reliability of the informant account. It should also prompt a return to the person's actual support needs. Grading intellectual disability is dealt with separately; the point here is narrower but essential: everyday adaptation has priority when the two numerical summaries pull in different directions.

If the scale cannot be administered, the recommended fallback is to ask socioculturally relevant questions and use DSM-5 severity specifiers as an interim clinical reference, pending ICD-11 guidance. This fallback must be documented as a clinical assessment rather than presented as a scored scale. It is particularly important to make explicit who supplied the history, which settings were represented, and which ordinary activities could not be meaningfully assessed.

6.3 Vineland scales: distinguish the scale family from the Indian VSMS

The **Vineland Adaptive Behavior Scales** are distinct from the Indian VSMS despite their similar names. The scale family is associated with Sparrow, Balla and Cicchetti; later editions are separately attributed in the source. The earlier social-maturity tradition is associated with Edgar Doll. In a viva, naming "Vineland" without specifying which instrument invites avoidable ambiguity about the form, the respondent, the norms, and the result being discussed.

The Vineland scales assess adaptive capacities for self-sufficiency across domains that include **communication, daily living skills, socialization, and motor skills**. They can be administered as a survey interview, expanded interview, informant-administered rating form, or teacher rating form. This range is clinically useful because the person's everyday capacities are often expressed differently at home and in an educational setting. It also creates an interpretive obligation: a discrepancy between parent and teacher reports should be explored as a context effect, not dismissed as an error.

Vineland-3 provides an age-normed framework for adaptive functioning and has self-report, parent-report, and teacher-rated youth forms. Its reported age span runs from **birth to age 90**. The strikingly broad span is a reminder that adaptive behaviour is not a childhood-only construct. Nevertheless, a score gains meaning only when the clinician considers the person's communication style, culture, opportunities, environmental supports, and the informant's frame of comparison.

Informant scales can compare a child with peers of the same age, but their parent-report basis lacks the objectivity of standardised child-based assessment. This is not a reason to discard informant data. It is a reason to use it intelligently. Direct testing samples what the person can show under examination conditions; an informant scale samples what is habitually enacted across time. These evidence streams should be reconciled, not forced into a false competition.

TRAP **Examination trap:** do not use VSMS and VABS as interchangeable labels. They are distinct instruments with different stated adaptations, formats, and assessment frameworks.

6.4 Developmental assessment: where the DST and DQ belong

The **Developmental Screening Test** was developed by Bharat Raj. It uses developmental tasks to assess global development across the stated childhood age range. The result is developmental screening, not an IQ test. This distinction is not semantic fussiness. A developmental screen is appropriate when the clinical question is whether development is globally delayed and whether fuller assessment is needed; it should not be made to carry a conclusion that its construct does not support.

Developmental scales are used when formal IQ tests are inapplicable, including very young age, sensory-motor issues, and gross understimulation. The Indian instruments named for this role are the Developmental Screening Test and the Developmental Assessment Scales for Indian Infants. Such selection is clinical reasoning, not a lesser substitute for formal testing. If a child cannot access the demands of an ability test, the clinician should choose a measure that captures the available developmental evidence and record why the conventional approach was not suitable.

The DST is highly loaded with speech and language items. It therefore requires cautious interpretation in cerebral palsy, autism, speech impairment, and hearing impairment. A low score in these circumstances may reflect the route through which the test asks for competence rather than the global developmental capacity one is trying to understand. The report should make the limitation visible and integrate nonverbal observation, history, and relevant specialist assessment. It should not quietly convert a language-loaded result into a global intellectual conclusion.

SITUATION	ASSESSMENT IMPLICATION	CLINICAL SAFEGUARD
Very young child, sensory-motor difficulty, or gross understimulation	Developmental scales can replace inaccessible IQ testing.	State why a conventional ability test could not answer the question.
Marked speech or language limitation	DST results need guarded interpretation.	Do not equate verbal loading with global developmental capacity.
Need for an adaptive account in an Indian setting	VSMS supplies the adaptive component.	Keep its SQ and profile separate from an IQ result.
Motor deficit affects an ability-test performance	Binet-Kamat profile analysis can separate a motor contribution.	Refer detailed IQ-instrument selection to the clinical assessment and IQ testing discussion.

6.5 Gesell: developmental model, drawing screen, and infancy

The **Gesellian model** starts with the newborn and tracks normal developmental trends across distinct but interrelated domains, rather than reducing infancy to a single factor of intellectual ability. This is a conceptual advance worth retaining in practice. Infancy is a period in which posture, movement, communication, social engagement, regulation, and emerging problem solving may develop unevenly. A developmental account should preserve that pattern rather than overstate the precision of an infant intelligence label.

The older infant-IQ model did not successfully extend conventional IQ assessment into infancy and has largely fallen out of use. The practical lesson is modest: report developmental findings with appropriate caution about what they predict, and explain the functional significance of the observed pattern. Prediction from infant intelligence testing is limited, whereas preschool testing is generally more informative. This is why longitudinal clinical review and repeated observation often matter more than attaching undue certainty to an early numerical label.

The **Gesell's Drawing Test** is an Indian screening instrument adapted by Verma and colleagues and later revalidated by Venkatesan. It is a performance screen for mental development in the stated age span. It obtains a rough estimate of intelligence from the child's ability to draw age-expected shapes. That word "rough" is the safeguard: the task can orient an assessment, but it cannot carry the authority of a comprehensive diagnostic conclusion.

Drawing performance is inseparable from opportunity and motor access. The test is not valid for children with no school attendance, no experience with a pencil, or specific finger-dexterity problems. A poor drawing response in such circumstances should be treated as a limitation of the

method, not as evidence of global inability. The same discipline applies to all brief screens: a screen should direct the next clinical question, never close it.

In the broader infant-assessment lineage, Nancy Bayley drew heavily on Gesell's developmental tasks and techniques. The Bayley-III assesses **five** domains: cognitive, language, motor, adaptive behaviour, and social-emotional functioning. It is reported for children from **1 to 42 months**. This breadth explains why infant assessment cannot be compressed into the question, "What is the IQ?" The clinician must communicate which developmental dimensions were sampled and which require follow-up.

6.6 Selecting and reporting measures when access is uneven

Test choice should begin with access to the task. Vision, hearing, motor control, expressive language, familiarity with materials, schooling, and cooperation can all change what a score represents. For screening in intellectual disability alone or with visual impairment, the stated pairing is DST with VSMS. With hearing impairment, the guideline pairs Gesell's Drawing Test and Seguin Form Board with VSMS; with cerebral palsy or locomotor difficulty, it pairs DST and Gesell's Drawing Test with VSMS. These are selection aids, not substitutes for clinical judgement about whether the child could genuinely engage with each task.

The Binet-Kamat Test may be unsuitable where its verbal and language demands dominate the child's route to a response. Detailed selection among IQ instruments belongs with formal clinical and IQ assessment; the important point here is to resist an instrument's familiar name when its route of administration is mismatched to the child. A defensible report says what was selected, why it was selected, what barriers remained, and how the barriers temper the conclusion.

A good report makes uncertainty useful. State the construct assessed, the source of information, the settings represented, any access barriers, and the practical implications of the profile. Avoid converting a screening estimate into a diagnostic certainty, an informant account into an objective observation, or an adaptive score into a legal percentage. The final formulation should identify present abilities, difficulties, supports, and the next assessment step. It should also state which conclusions are well supported, which remain provisional, and what observation or collateral history would reduce the uncertainty. That is more clinically valuable than a crowded list of scales.

TRAP **Examination trap:** DST, VSMS, and Gesell's Drawing Test are not interchangeable IQ tests. They respectively address developmental screening, social maturity and adaptive profile, and developmental screening.

The causes of intellectual disability

7 · Etiology of intellectual disability (genetic, prenatal, perinatal, postnatal)

Aetiology is clinical work, not a label-collecting exercise. Intellectual disability (ID) is the final common developmental outcome of heterogeneous influences on the developing brain. The

useful question is not merely “what diagnosis does this person have?”, but whether the history, examination and targeted investigations can identify a cause that changes care, anticipatory guidance, recurrence discussion, or surveillance. A causal formulation should preserve timing, mechanism and certainty. It should also distinguish a demonstrated cause from an associated finding and from an unexplained presentation.

For the examination candidate, a timing-based account gives a dependable spine: prenatal influences, events around birth, and postnatal acquired influences. This is not a claim that every presentation fits a clean compartment. Developmental vulnerability may begin before birth and become clinically apparent later; genetic susceptibility and environmental exposure can coexist. The classification is useful because it structures the history, directs investigation, and keeps preventable acquired causes visible without reducing a complex developmental history to a single event.

50-70% ESTIMATED PRENATAL CONTRIBUTION	**10-20%** ESTIMATED PERINATAL CONTRIBUTION	**5-10%** ESTIMATED POSTNATAL CONTRIBUTION	**25-50%** GENETIC CAUSES OVERALL

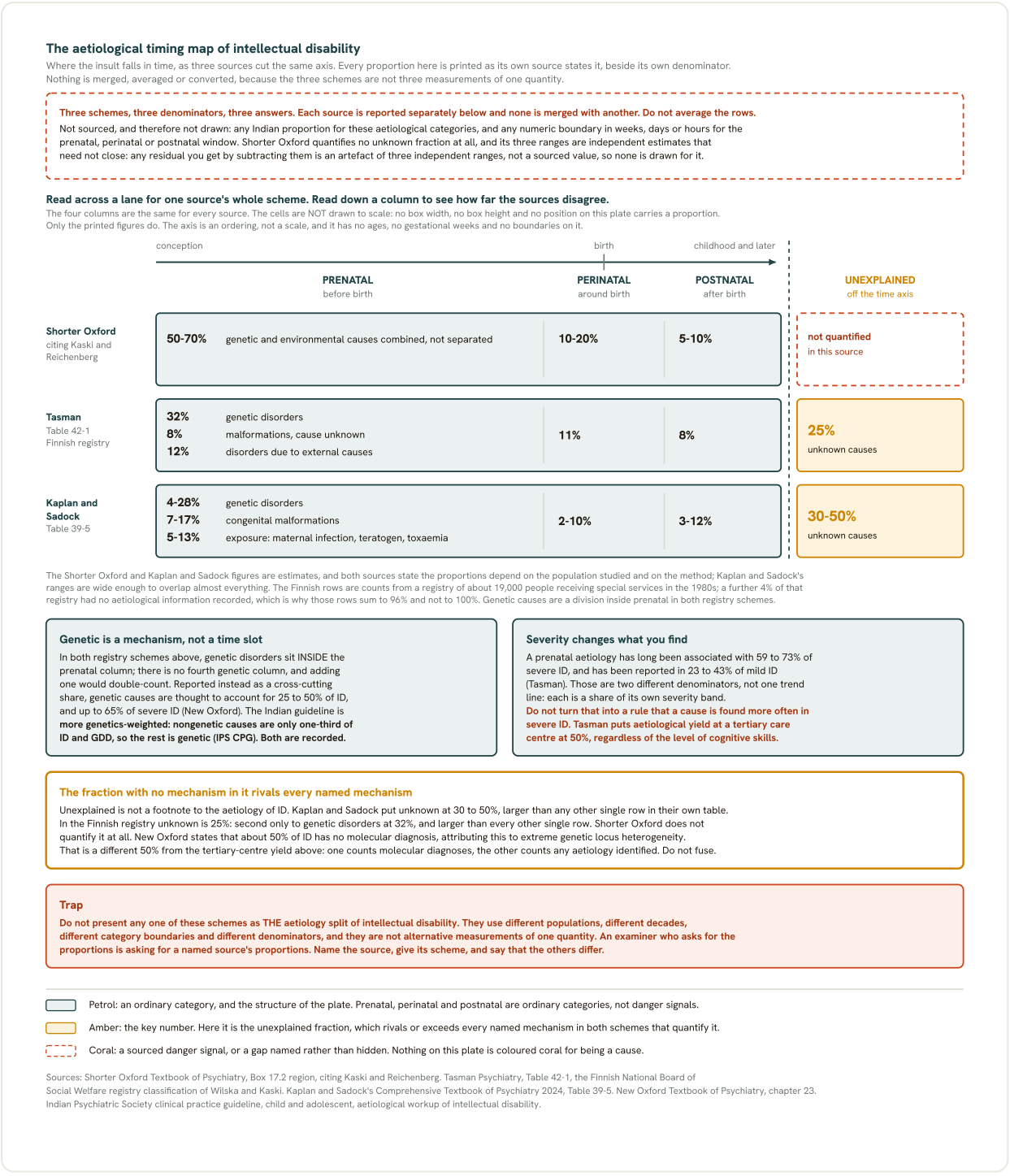


Fig 21.5 The aetiological timing map of intellectual disability. The causes of ID sorted by when the insult falls, and the reason a single answer does not exist: three sources cut the same axis with different category boundaries, different populations and different denominators, so their proportions are reported lane by lane and never merged. The plate teaches three things at once. Genetic is a mechanism that sits inside the prenatal column, not a fourth time slot. Severity shifts the prenatal and genetic share upward without licensing the conclusion that a cause is found more often. And the fraction with no mechanism in it rivals or exceeds every named mechanism in both schemes that quantify it. Shorter Oxford quantifies no unknown fraction, and none is inferred for it here.

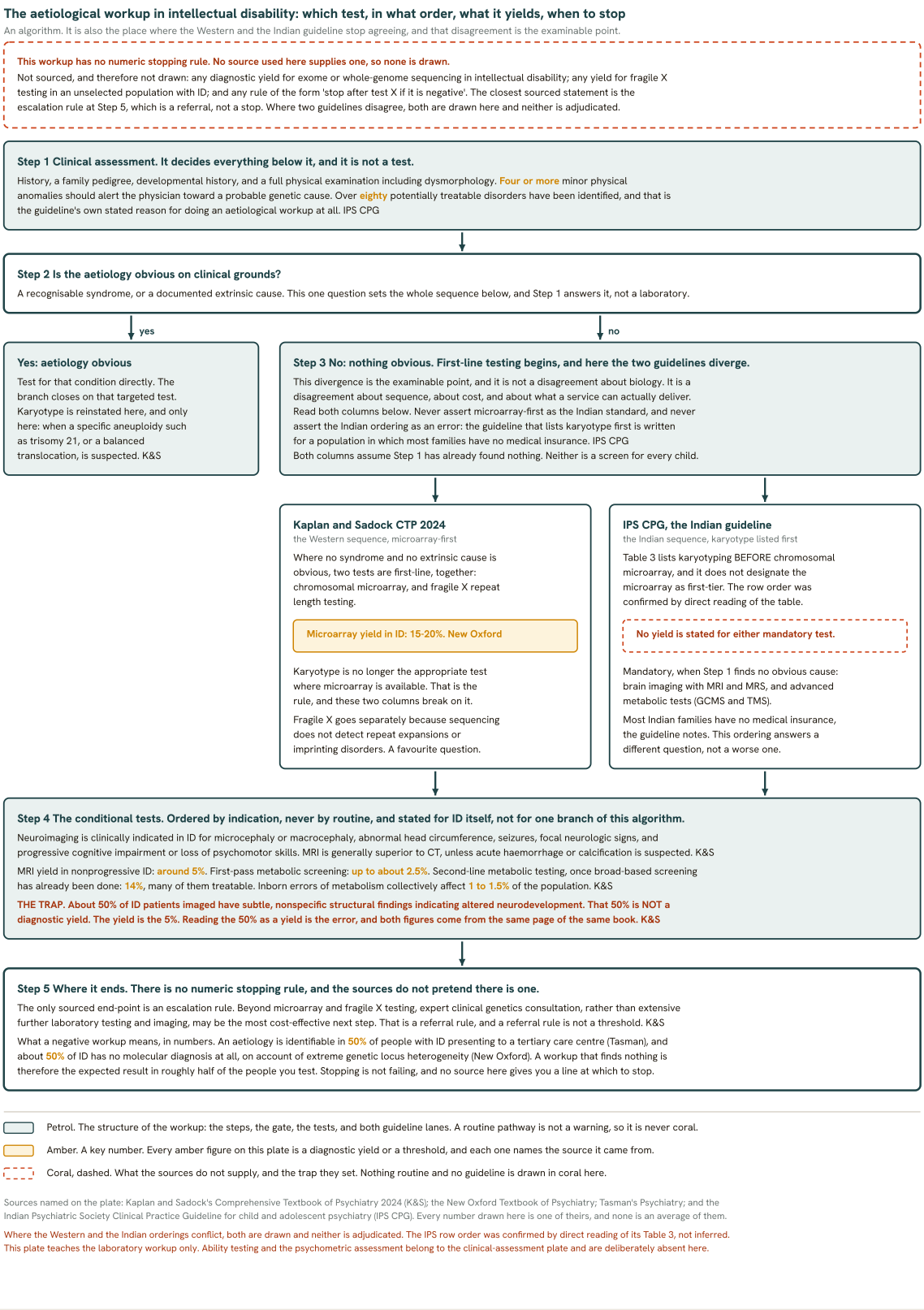


Fig 21.6 The aetiological workup in intellectual disability. The sequence a clinician actually runs: clinical assessment first, then a single gate that asks whether the aetiology is already obvious, then first-line testing, where the Western and the Indian guideline diverge and both orderings are drawn without either being called correct. Each test carries its own sourced diagnostic yield, and the yields are deliberately shown to be small. The workup ends not at a threshold but at a referral, because no source used here supplies a numeric stopping rule, and because an aetiology is found in only about half of the people tested.

7.1 A timing-based causal framework

Prenatal aetiology includes genetic causes, congenital malformations and adverse intrauterine exposures. Across all severities, prenatal factors are estimated to account for 50-70% of cases. The figure is best read as an orientation, not as a prediction for an individual patient. It combines genetic and environmental pathways, and its apparent size will vary with ascertainment, availability of neonatal care, diagnostic methods and the population being studied.

Perinatal aetiology refers to influences operating around birth. It is estimated to contribute 10-20% of ID overall. The central clinical discipline is temporal: obtain the obstetric and neonatal narrative carefully, but do not convert a difficult delivery into a causal conclusion without corroborating developmental, neurological and investigation findings. A birth complication may be relevant, incidental, or one part of a broader developmental disorder.

Postnatal aetiology comprises acquired influences after birth that affect brain development. It is estimated to account for 5-10% of cases. In practice, the history asks whether there was a previously acquired skill that was lost, a period of severe illness or injury, or evidence that development changed trajectory. Such questions protect against prematurely calling a presentation genetic or unexplained.

Causal language should match the evidence. A documented exposure or event can be part of the history without being sufficient to explain the developmental phenotype. Concordance matters: the proposed timing, neurological findings, developmental course and investigation results should tell a coherent story. Where they do not, retain alternative explanations and revisit the formulation as new records or findings become available. This avoids both diagnostic anchoring and the equally unhelpful practice of listing every historical event as a cause.

RULE **Timing is a map for enquiry, not proof of causation.** The history proposes a mechanism; examination and appropriately chosen tests decide how securely it can be attributed.

The categories also overlap in a meaningful way. A congenital brain malformation is prenatal in origin even if recognised only after delivery. A genetic condition may make a child more vulnerable to an intercurrent insult. Conversely, an exposure can cause a developmental phenotype without yielding a pathognomonic laboratory result. Good formulation therefore states the strongest supported explanation and records residual uncertainty honestly.

7.2 Reading frequencies without misreading the patient

Published distributions are not interchangeable. In a broad classification, prenatal genetic disorders were estimated at **4-28%**, congenital malformations at 7-17%, and prenatal exposure at 5-13%. The wide ranges are themselves the lesson: aetiological proportions are shaped by what a service can recognise and whom it sees. The same classification estimated perinatal causes at 2-10%, postnatal causes at 3-12%, and unknown causes at 30-50%.

Severity also changes the diagnostic landscape. Prenatal aetiology has been reported in **59-73%** of severe ID and in 23-43% of mild ID. This does not make mild ID “non-biological”, nor does it authorise an assumption that a severe presentation must be genetic. It means the threshold for

pursuing a careful aetiological workup should remain high across presentations, while the pattern of findings and the probable yield may differ.

CLINICAL QUESTION	WHAT IT PROTECTS AGAINST	HOW TO USE THE ANSWER
When did concern first become apparent?	Assigning timing from a single memorable event	Place the concern beside pregnancy, birth and developmental history.
Was development static, delayed, or did skills diminish?	Missing an acquired or progressive process	Use the trajectory to decide how urgently neurological and metabolic causes need consideration.
Are there family, physical or neurological clues?	Calling a pattern idiopathic before examining it	Integrate inheritance, morphology and examination into the testing plan.
Is a cause demonstrated, probable, or still unknown?	Overstating causality to families	Document the degree of certainty and keep a route for review.

Unknown cause is not synonymous with inadequate care. It can reflect extreme genetic heterogeneity, limitations of the available test, incomplete historical information, or an interaction of influences that cannot be disentangled. In tertiary care, an aetiology is identifiable in 50% of people presenting with ID, yet about 50% of cases may lack a molecular diagnosis. These statements refer to different denominators and endpoints. One is a clinically identifiable cause; the other is a molecular answer. They should never be treated as competing estimates of the same thing.

7.3 Genetic architecture: why no single test tells the whole story

Genetic does not mean simple inheritance. Genetic causes are thought to account for **25-50%** of ID overall, and family and twin studies suggest heritability between 30-90%, depending on diagnostic category and study design. These broad estimates should not be turned into an individual recurrence estimate. They establish that inherited and newly occurring genomic changes deserve active consideration, particularly when the phenotype, family history or examination points in that direction.

Copy number variation refers to a deletion or duplication of genomic material. Cumulatively, CNVs are described as the commonest genetic abnormality class causing ID. Array-based CNV analysis identifies microdeletions or duplications in approximately **15-20%** of tested patients with ID. This is why a genomic test that can detect dosage imbalance belongs early in the assessment rather than being reserved only for those with striking dysmorphism.

Single-gene heterogeneity is equally important. Mutations in more than 700 genes are estimated to be capable of causing ID. In severe sporadic non-syndromic ID, trio-based exome sequencing has identified de novo heterozygous variants or small indels in 13-55%. The range should prevent opposite errors: neither assume that an apparently isolated case lacks a genetic explanation, nor promise that sequencing will always deliver a definitive answer.

X-linked intellectual disability provides a reason to elicit family history with attention to affected males and the maternal lineage. Mutations in known X-linked genes account for **10-12%** of males with ID, and yet no single one of those X-linked genes explains more than **0.1%** of intellectual disability. This is locus heterogeneity in practical form: a recognisable inheritance pattern does not identify the molecular diagnosis by itself. The syndrome-specific genetics and recurrence counselling belong with the relevant syndrome discussions, not with a generic workup.

Autosomal recessive intellectual disability deserves particular attention when the pedigree suggests shared ancestry or recurrence among siblings. In outbred populations it may account for 10-20% of ID; autosomal recessive disorders are reported to be up to threefold more frequent among inbred than non-inbred cases. The history must be asked respectfully and documented neutrally. Its purpose is to guide diagnosis and counselling, never to assign blame.

7.4 From phenotype to an ordered genetic evaluation

The diagnostic interview begins before the requisition form. Obtain a pregnancy and birth history, a developmental trajectory, a pedigree, previous records and the parents' account of strengths as well as difficulties. On examination, look for growth pattern, head size, minor anomalies, skin findings and focal neurological signs. The presence of at least four minor physical anomalies should alert the clinician to a probable genetic cause. The value of this threshold is not that it diagnoses a syndrome; it is that it changes the prior probability and supports a more deliberate genetic examination.

In a person without an obvious syndrome or clear extrinsic cause, **first-line genetic testing** comprises chromosomal microarray and fragile X repeat-length testing.

> **GOOD TO REMEMBER**
>
> These tests answer different questions and are complementary: microarray looks for chromosomal copy-number abnormalities, whereas a separate fragile X assay addresses a repeat-length abnormality. Do not assume that one result substitutes for the other merely because both are called genetic tests.

Karyotyping is no longer the appropriate routine test when chromosomal microarray is available. It remains indicated when a specific aneuploidy or a balanced translocation is suspected. This is a test-selection distinction, not a hierarchy of "old" and "new" medicine: the suspected abnormality determines which technique can answer the question.

TRAP **Examination trap:** a normal sequencing result does not exclude every genetic mechanism. Currently available sequencing tests do not detect **repeat expansions and imprinting disorders**.

After initial testing, a negative result should trigger re-phenotyping and **expert clinical genetics consultation**, rather than indiscriminate laboratory panels and imaging. A genetics assessment can refine the phenotype, determine whether a different genomic method is appropriate, revisit

the pedigree and frame the residual uncertainty for the family. This is also the right setting for generic counselling: explain what is known, what remains unresolved, and why recurrence discussion must be based on the established or suspected mechanism rather than on a generic population figure.

Test ordering should remain hypothesis-led. A phenotype can make a particular mechanism more plausible, but it should not be used as a gate that excludes broad genetic evaluation solely because dysmorphism is subtle or family history is unrevealing. Conversely, a long list of investigations is not a substitute for a better history and examination. Reassessment after each result is often more informative than simply adding tests: ask whether the new finding explains the developmental picture, whether it is concordant with the family history, and whether it creates an actionable surveillance or counselling question. This is how the workup stays both clinically rigorous and proportionate.

> A negative initial genetic screen ends neither the clinical search nor the duty to explain uncertainty clearly.

7.5 Metabolic and neuroimaging pathways

Inborn errors of metabolism are individually uncommon but collectively affect 1-1.5% of the general population. Their importance in ID is disproportionate to their frequency because some are potentially modifiable and because clinical clues may be subtle. First-pass metabolic screening accounts for up to approximately 2.5% of screened patients with ID. A low yield is not an argument for abandoning clinical reasoning; it is an argument against indiscriminate testing in the absence of a phenotype-driven question.

When broad screening has already been undertaken, second-line metabolic testing identified an IEM in **14%** of those tested, many of which were treatable. Progressive change, loss of psychomotor skills, episodic deterioration, unusual neurological features or a family pattern should therefore prompt reconsideration of metabolic disease. The detailed disorders and their treatment are addressed in the section on inborn errors of metabolism; here, the point is to keep an aetiology that may alter outcome within the diagnostic field of view.

Neuroimaging is clinically indicated with microcephaly or macrocephaly, abnormal head circumference, seizures, focal neurological signs, progressive cognitive impairment, or loss of psychomotor skills. MRI is generally superior to CT, except when acute haemorrhage or calcification is suspected. Imaging must answer a clinical question. Subtle nonspecific findings may support altered neurodevelopment without establishing a diagnosis, whereas a structural abnormality may reorganise the entire workup.

Guidelines can differ by setting and resource pathway. The candidate should state the local recommendation accurately while retaining the principle that testing is most useful when integrated with phenotype and clinical trajectory.

IN INDIA

Where obvious clinical aetiologies are not found, the Indian Psychiatric Society guideline designates **brain MRI with MRS and advanced metabolic tests using GCMS and TMS** as mandatory investigations.

7.6 Prenatal, perinatal and acquired causes in the clinical narrative

After the genetic and structural possibilities have been considered, return to the developmental history and ask what may have acted before birth, around birth or after birth. Prenatal exposure categories include maternal infection, teratogen exposure and toxaemia. The purpose of asking is to establish timing and biological plausibility, not to retrospectively place moral responsibility on a mother or family. The same non-judgemental stance applies to histories of substance exposure, obstetric difficulty, neonatal illness and later acquired insults.

Perinatal history should be reconstructed from records wherever possible. The more remote the event and the less specific the account, the more cautious the attribution should be. The examiner's answer becomes stronger when it separates a reported event from evidence of its neurological consequence. Likewise, a postnatal history should seek changes in development, neurological symptoms and the sequence between illness and altered functioning. Aetiological formulation can then state whether the acquired explanation is established, plausible or insufficiently supported.

- Start with timing, then ask what mechanism could link the event to altered development.
- Seek corroboration from records, examination and the developmental trajectory before naming causation.
- Keep genetic and acquired explanations open together when the evidence supports neither as exclusive.

This approach matters especially when families arrive with a single explanatory story, such as a difficult delivery or a relative with developmental problems. Both may be important. Neither should stop the clinician from completing the rest of the formulation. The respectful task is to test the story, preserve what is supported, and correct what is overconfident without making the family feel dismissed.

7.7 Aetiological formulation and communication

A useful written formulation has a layered structure: developmental phenotype, best-supported timing category, demonstrated mechanism where available, associated comorbid findings, and a statement of certainty. It is better to write "ID with a probable prenatal genetic aetiology under evaluation" than to attach a speculative syndrome name. This language guides follow-up and is safer for families, schools and other clinicians who may read the record later.

Explain the purpose of testing before ordering it. Families should understand that testing may identify a diagnosis, clarify future health needs, inform recurrence discussion, or remain non-diagnostic. They should also be prepared for findings whose significance is uncertain. Counselling is a process of interpretation and decision support, not simply the handing over of a laboratory

report. When a family pattern is suspected, referral allows the family to discuss who else may benefit from assessment without converting a probability into a certainty.

The clinical payoff is precision with humility. Aetiology helps prevent a treatable contributor from being missed, prevents unnecessary investigations from accumulating, and makes recurrence counselling more honest. Yet the person's current needs do not wait for molecular closure. Diagnostic workup should proceed alongside support planning and management, with each service informed by the level of certainty rather than delayed by it.

8 · Down syndrome and other chromosomal causes

Why this cause deserves its own clinical frame. Down syndrome is the chromosomal condition most likely to turn a general discussion of intellectual disability into a real consultation: the clinician has to recognise a developmental profile, separate appearance from diagnosis, look for treatable contributors to disability or behavioural change, and explain why the cytogenetic result matters to a family. It is therefore not enough to say that the person has an extra chromosome. The mechanism determines the meaning of the result for relatives and future pregnancies.

For the psychiatry candidate, the useful mental model is a sequence: establish the syndrome and its chromosome mechanism; understand the pattern of associated health needs; then interpret new symptoms against the person's usual functioning. Intellectual disability is part of the presentation, but its definition, assessment and severity grading belong in their respective sections. Equally, a behavioural change should not be dismissed as an inevitable consequence of the syndrome. It may be the presentation of an associated medical disorder, sensory loss, or a co-occurring psychiatric condition.

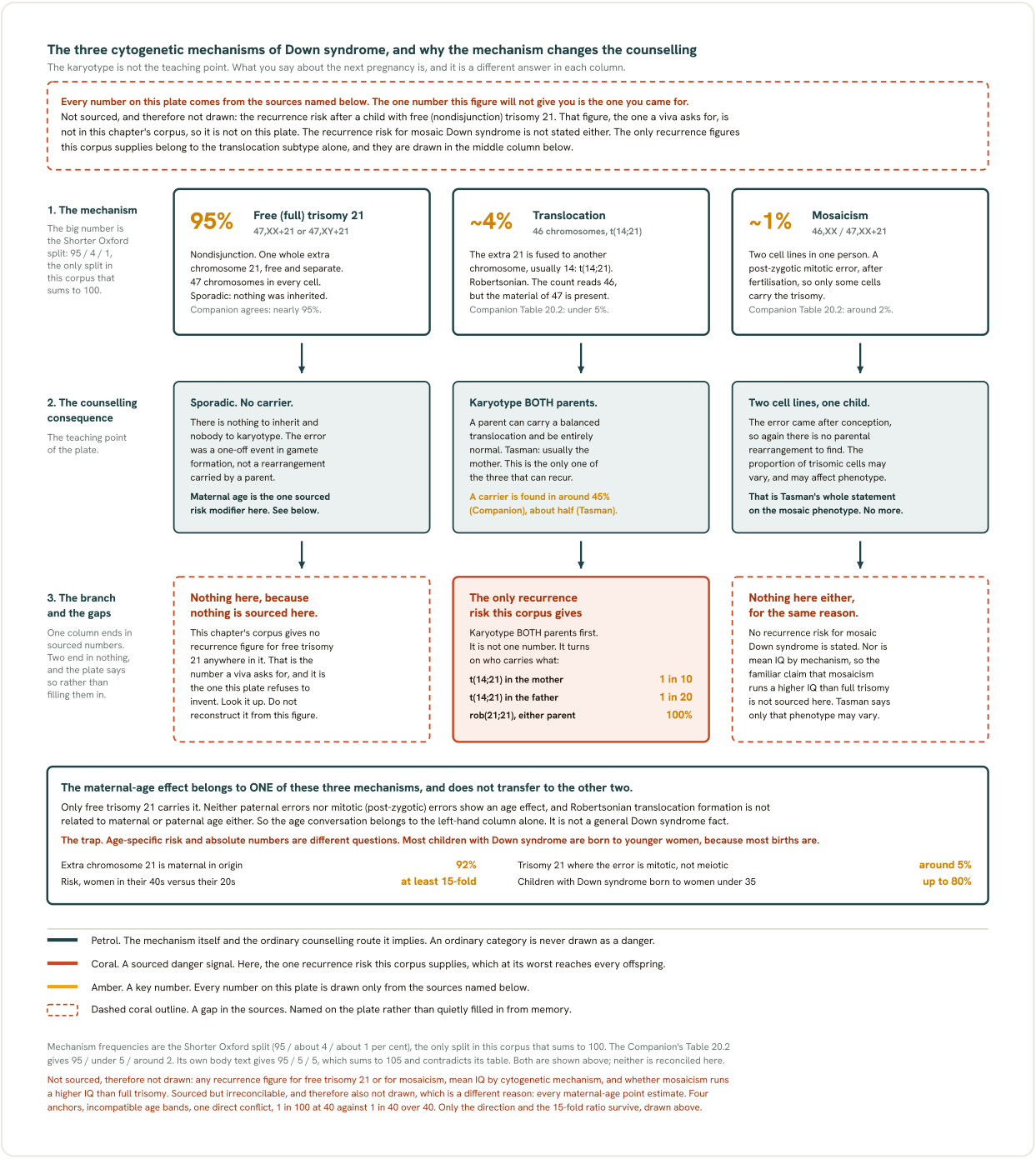


Fig 21.7 The three cytogenetic mechanisms of Down syndrome. Free trisomy, translocation and mosaicism read across from the karyotype to the only thing that changes management, which is what you tell the parents about the next pregnancy. The plate is built so the three columns diverge exactly where the counselling diverges: only the translocation column earns a parental karyotype, and only that column ends in recurrence figures the sources actually supply. The other two columns end in a named gap, because this chapter's corpus states no recurrence risk for free trisomy 21 and none for mosaicism, and the plate declines to supply from memory the figure a viva asks for. The maternal-age effect is shown last and bound to one mechanism only.

8.1 Epidemiological position and clinical significance

Down syndrome is a leading chromosomal cause of intellectual disability and is clinically important precisely because it is common enough to be encountered across child, adult and liaison settings. Textbook birth estimates differ: Shorter Oxford gives about **1 in 650** live births, whereas Tasman gives **1 in 800** live births. These are not competing bedside tests. They illustrate that a quoted frequency depends on the source population and the outcome counted. A figure

expressed per pregnancies must not be treated as interchangeable with a live-birth figure: a cited source estimates trisomy 21 at 1.5 in 1000 pregnancies.

Within the population of people with intellectual disability, Down syndrome accounts for between **6-8%**. That burden explains why psychiatrists need fluency in its medical context, not only recognition of its phenotype. The consultation often occurs at a transition point - concern about development, a new behavioural presentation, family anxiety about recurrence, or declining function in an adult - when an apparently familiar label can obscure the actual clinical question.

95%	about 4%	about 1%
FREE TRISOMY 21	TRANSLOCATION	MOSAICISM

8.2 Cytogenetic architecture: the mechanism is the counselling clue

RULE **State the chromosome mechanism before discussing recurrence: the phenotype may be similar, but the family implication is not.**

Down syndrome results when there is extra chromosome 21 material. In **full trisomy 21**, the additional chromosome is present as a separate chromosome. This is the usual arrangement and, in the source cited here, applies to nearly 95% of people with Down syndrome. The extra material arises through a chromosome-separation error, conventionally termed **nondisjunction**. The molecular event makes it essential to distinguish this arrangement from a structural rearrangement identified in a parent.

In **translocation Down syndrome**, the extra chromosome 21 material is attached to another chromosome, usually chromosome 14, designated t(14;21). The total count can still be 46 chromosomes, but chromosomal material from 47 is present. This is the exam discriminator: counting chromosomes alone does not tell the whole genetic story. The relevant question is whether the chromosome complement is balanced in a parent or unbalanced in the child. The answer changes the recurrence discussion and is why a cytogenetic report must be read rather than merely filed.

Mosaicism means that different cell lines are present. Its presence should prompt precision in description rather than casual prediction from appearance or a presumed developmental outcome. The safe clinical teaching point is not to manufacture a hierarchy of functioning from a mechanism alone. Mechanism informs counselling; it does not replace individual assessment of development, communication, adaptive functioning and health.

CHROMOSOME ARRANGEMENT	WHAT THE REPORT MEANS CLINICALLY	WHY PSYCHIATRY SHOULD CARE
Full trisomy	An extra chromosome 21 is present as a separate chromosome.	It frames the syndrome but does not predict an individual's support needs.
Unbalanced translocation	Extra chromosome material is attached to another chromosome.	Parental chromosome studies become central to the family discussion.
Mosaicism	Different cell lines are present.	Do not infer clinical ability from the label alone; assess the person.

The accompanying figure is useful as a memory aid because it puts these arrangements beside their counselling consequence. It should prevent a common error: treating every Down syndrome result as if it carried the same familial implication.

Read the report in a clinically disciplined way. **Cytogenetic interpretation** starts by identifying whether it describes a separate extra chromosome, attached extra material, or differing cell lines. Next ask whether the wording establishes a structural rearrangement that could also be present in a parent. Finally, distinguish what the report establishes from what still needs direct clinical assessment. A chromosome result explains the syndrome's mechanism; it does not by itself describe communication, capacity, family resources, psychiatric symptoms or the cause of a new behavioural change. This prevents genetic language from becoming a shortcut around formulation.

The same distinction helps when communicating with other clinicians. “Down syndrome confirmed” may be adequate as a diagnostic headline but is incomplete as a handover when the mechanism has not been stated. Record the arrangement in words, note whether parental studies are relevant, and separate known information from questions that remain open. This makes later counselling more coherent and stops a vague family-history statement from acquiring unwarranted certainty as it passes between services.

8.3 Origin of the extra material and maternal-age reasoning

Most molecularly studied trisomy 21 cases have an extra chromosome of maternal origin, reported as 92%. Maternal meiotic error is therefore the dominant framework, but a candidate should avoid turning that into an indiscriminate statement about every pathway. Around 75% of nondisjunction errors are reported at the first maternal meiotic division, although the cited source does not make the denominator sufficiently explicit to claim more than that. In contrast, 60% of paternal meiotic errors occur at the second meiotic division, and mitotic error is estimated in around 5% of trisomy 21 cases.

The maternal-age association belongs mainly to the maternal meiotic pathway. **Paternal and mitotic errors do not show an age effect, and Robertsonian formation is not related to parental age.** This distinction matters because a risk factor should be linked to the biological pathway it plausibly modifies, not applied as a universal explanation after the event.

Age-specific risk rises substantially. Shorter Oxford reports about 1 in 2000 live births among mothers aged 20-25, compared with 1 in 30 among those aged 45 years or over. Another table gives 1 in 2000 at maternal age 30 and 1 in 100 at age 40. The direction is the high-yield point, not the temptation to merge differing source anchors into a fabricated universal rate. Women in their forties have at least a 15-fold increase in risk compared with women in their twenties.

TRAP **Do not confuse high age-specific risk with where most affected children are born.**

Up to 80% of children with Down syndrome are born to mothers under 35. This is not a paradox. Population impact depends on both individual risk and the number of births in each age group. It is the cleanest way to explain why counselling cannot be reduced to stereotypes about maternal age.

8.4 Phenotype, development and associated health needs

The physical phenotype is a recognition aid, not a diagnostic algorithm. Individual signs vary, and no individual sign is universal. The most consistent physical feature, upslanting palpebral fissures, is still restricted to 80% of people with Down syndrome. A transverse palmar crease occurs in 53%, while a protruding tongue is present in fewer than 50%. The practical implication is simple: absence of a familiar sign should not overrule a chromosome result, and presence of an isolated sign should not be treated as proof of the syndrome.

Intellectual ability varies considerably. Tasman reports a mean IQ of approximately 50, but the number is not a prognostic verdict for an individual and must not be used to grade intellectual disability. The clinician should describe present skills, communication, daily functioning and the support environment. The sections on clinical assessment, IQ testing and adaptive behaviour explain how to do that without mistaking a test score for the person.

ASSOCIATED FINDING	REPORTED FREQUENCY	CLINICAL CONSEQUENCE IN A PSYCHIATRIC ASSESSMENT
Congenital heart disease	40-50%	Medical history and exertional symptoms shape both assessment and treatment planning.
Gastrointestinal atresias	12%	Past surgical and nutritional history can clarify the developmental context.
Hearing loss	75%	Often conductive and related to otitis media; it can mimic inattention, withdrawal or a language problem.
Ocular findings	approximately 50%	Visual difficulty may alter engagement, learning and apparent behaviour.
Gluten intolerance	4-20%	Gastrointestinal symptoms should not be reduced to a behavioural complaint.
Autoimmune thyroid dysfunction	up to 20%	In adolescents and adults, consider it when behaviour, mood or cognition changes.

GOOD TO REMEMBER

These are not decorative associations. Hearing and visual difficulty alter how a person is read in the clinic. Pain, discomfort or endocrine disturbance may appear as irritability, reduced participation, slowing, sleep change or loss of previously reliable skills. In particular, thyroid dysfunction is a crucial differential when behavioural disturbance, depression or an apparent dementia presentation emerges. Start with a change from baseline and ask what has changed in the body, the sensory environment and the person's routines before assigning a psychiatric explanation.

Some children with Down syndrome also meet criteria for autism spectrum disorder, estimated at approximately 7%. Autism should be assessed using its own diagnostic framework. The diagnostic criteria, clinical features and differential diagnosis of autism are dealt with in the autism sections of this chapter. Here the point is narrower: a known chromosomal diagnosis does not make an additional neurodevelopmental diagnosis impossible, nor does it make assessment dispensable.

8.5 A psychiatry assessment that does not miss the treatable problem

RULE **A new symptom is a clinical question, not an automatic feature of Down syndrome.**

Begin by identifying the change in observable terms: what is different from the person's own baseline, where it occurs, and what has altered in eating, sleep, activity, communication, comfort or social participation. A **baseline comparison** prevents the interview from confusing a lifelong trait with a fresh symptom. Establish the time course from people who know the person well, but

do not let collateral description substitute for direct engagement. A quieter person may be depressed, unwell, hearing less well, overwhelmed, or responding to a changed environment. These possibilities require clinical discrimination.

- Review hearing and vision when apparent inattention, language difficulty or withdrawal is new or worsening.
- Ask about physical discomfort and gastrointestinal symptoms when behaviour becomes irritable or avoidant.
- Consider thyroid dysfunction when there is behavioural disturbance, depressive symptomatology or cognitive decline.
- Keep autism assessment criterion-based rather than treating the chromosomal diagnosis as either proof or exclusion.

This approach guards against diagnostic overshadowing, discussed fully in the dual-diagnosis section. The syndrome-specific safeguard is especially important here because associated health conditions can change behaviour without announcing themselves in conventional psychiatric language. The clinical record should preserve the observed baseline, the reported change, the relevant physical and sensory questions, and the working explanation. Doing so makes it possible for the next clinician to see whether the proposed explanation accounted for the change or merely accompanied it.

MNEMONIC

CHROME

: **C**hromosome mechanism; **H**earing and vision; **R**eview change from baseline; **O**rgan-system contributors; **M**ood and mental-state assessment; **E**xplain the recurrence implication. Use it as an interview sequence, not as a substitute for examination.

When behaviour changes in Down syndrome, look for a change in health, sensory input or environment before attributing it to the syndrome.

8.6 Family implications of translocation Down syndrome

The family conversation becomes most specific when a translocation is identified. In about half of translocation Down syndrome cases, a parent has a balanced translocation; a related source estimates that around 45% of Robertsonian translocation Down syndrome cases are inherited from a balanced carrier parent. The practical message is not that every parent is a carrier, but that parental chromosome status is material to that particular family's recurrence discussion.

For a balanced **t(14;21) translocation**, the cited recurrence risk for a subsequent child with Down syndrome is **1 in 10** when the mother carries the translocation and **1 in 20** when the father carries it. These are mechanism-specific figures. They must not be offered as the risk after ordinary full trisomy, nor copied into a discussion before the parental studies have established the relevant arrangement.

A familial balanced **rob(21;21) fusion** has a qualitatively different implication: all offspring will have Down syndrome (100%). This is exactly why wording matters. “Down syndrome runs in the family” is too imprecise to guide care. The chromosome report, the parent who carries any balanced rearrangement, and the specific rearrangement together determine what can responsibly be said.

Refer families for genetic counselling and interpret recurrence as a syndrome-specific issue, not as a generic estimate. The general approach to aetiological investigation and counselling referral is covered in the etiology of intellectual disability section. The psychiatrist's contribution is to recognise when the mechanism makes that referral urgent, to communicate uncertainty honestly, and to ensure that distress does not prevent the family from receiving a clear explanation.

8.7 Examination synthesis: how to write the answer safely

A strong answer begins with Down syndrome as a chromosomal cause of intellectual disability, then moves from cytogenetics to clinical care. Describe full trisomy, translocation and mosaicism as different chromosome arrangements; explain that translocation has particular implications for parental studies and recurrence; then show why sensory, endocrine, gastrointestinal and cardiac associations matter to psychiatric formulation. Finish with the warning against diagnostic overshadowing. This structure demonstrates reasoning rather than reciting a facial-feature list.

TRAP **The classic examination mistake is to give a single recurrence figure for every form of Down syndrome.**

That answer is unsafe because it erases the distinction that the cytogenetic result is meant to reveal. A further mistake is to treat the physical phenotype as diagnostic while ignoring the associated health review. The better answer states that phenotype prompts recognition, cytogenetics establishes mechanism, and longitudinal assessment identifies the problem requiring action now.

Other chromosomal causes of intellectual disability need syndrome-specific evidence on their phenotype, mechanism and recurrence patterns. This fragment deliberately does not generalise Down syndrome data to them. Where a person has a chromosomal finding without the characteristic Down syndrome arrangement, the clinical task is to read the individual report and pursue the aetiological pathway rather than forcing it into a familiar template.

9 · Fragile X syndrome

Why this syndrome deserves a deliberate search. Fragile X syndrome is a recognisable cause of intellectual disability, but it cannot safely be reduced to a facial gestalt or a single childhood phenotype. It is the most common inherited cause of intellectual disability, while among all specific causes it is described as second to Down syndrome. Its importance extends beyond the proband: the same locus can produce a developmental disorder in one family member and late-life neurological or reproductive disorders in relatives who carry a premutation.

The useful clinical stance is therefore syndromic and family-based. Recognise the developmental and behavioural pattern, examine without over-relying on dysmorphology, confirm the molecular finding with an appropriate repeat-expansion assay, and explain that molecular category and parent of origin determine the implications. The accompanying figure can be used to hold the relationship between repeat expansion, gene silencing and the distinct premutation disorders in mind.

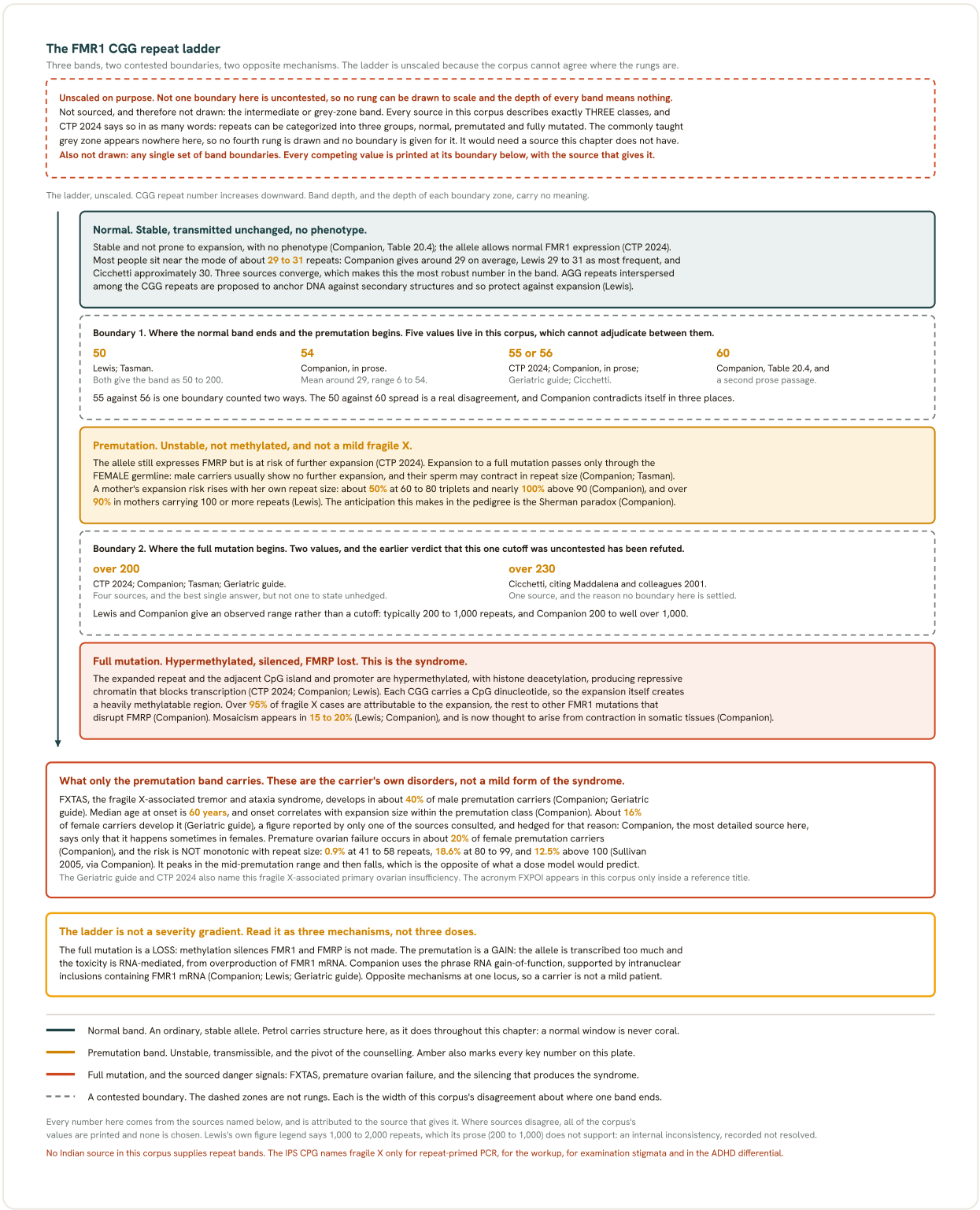


Fig 21.8 The FMR1 CGG repeat ladder. The three repeat classes this corpus supports, what each one carries, and the fact that neither boundary between them can be stated as a single number.

9.1 Molecular lesion and its consequences

Fragile X syndrome results from loss of function of **FMR1 (fragile X mental retardation gene 1)**, located at **Xq27.3**. The unstable sequence is a **CGG (cytosine-guanine-guanine)** repeat in the promoter-adjacent 5' untranslated region. It is worth retaining the anatomical wording rather than insisting on a single exon label, because the sources differ at that finer level while agreeing that the repeat is non-coding and close to the promoter.

Repeat expansion accounts for **more than 95%** of affected cases. In the full-mutation state, the expanded sequence and neighbouring regulatory region become hypermethylated; histone deacetylation and repressive chromatin then prevent transcription. Thus the mutation is not simply a long piece of DNA. It converts a transcriptionally active locus into one that is functionally silent. The extent of methylation is directly related to the loss of functional protein, which helps explain why repeat size alone is not a complete clinical predictor.

The protein product, **FMRP, an RNA-binding protein**, normally binds polyribosomes in dendrites and synaptic spines and largely restrains local translation of synaptic proteins. Loss of that restraint provides a coherent bridge from molecular change to altered synaptic maturation, learning, attention and behaviour. It also prevents a simplistic conclusion that every clinical feature is fixed at birth: the disorder concerns regulation of developing neural systems and its expression changes with development.

RULE **Full mutation means loss of FMRP through epigenetic silencing; premutation disorders arise through excess FMR1 messenger RNA, not a mild form of the same process.**

This distinction is the organising principle of the chapter. In a premutation, the locus is not silenced in the same way. Instead, **RNA-mediated toxicity from overproduction of FMR1 mRNA** is implicated. Calling a premutation carrier “mildly affected with fragile X syndrome” is therefore biologically misleading and clinically unhelpful. One state is principally a protein-loss developmental disorder; the other is an RNA gain-of-function state with its own age-related and reproductive associations.

9.2 Repeat categories, instability and anticipation

Repeat-length bands are useful in laboratory interpretation, but their lower boundaries vary modestly across reference texts. A normal stable allele is reported in ranges such as 5-55 CGG repeats, while a commonly used premutation band is **55-200 repeats**. The clinically robust distinction is that a full mutation is generally defined as more than 200 repeats, although a cited source uses a higher threshold. A report must be interpreted using the laboratory’s stated classification rather than by treating a disputed boundary as a universal biological cliff.

Normal alleles tend to be stable. **AGG interruptions** interspersed within the repeat are proposed to stabilise the sequence by reducing secondary structures during replication. Long uninterrupted stretches are more prone to expansion. This is why molecular counselling cannot be reduced to a family member’s label alone: the repeat architecture and its transmission context matter.

The inheritance is **X-linked**, but it does not behave like a simple static X-linked mutation. Expansion from a premutation to a full mutation occurs through the female germline; male carriers usually transmit without further expansion and may show contraction in sperm. This parent-of-origin effect explains the apparently paradoxical pedigree in which an unaffected transmitting man has daughters who are carriers, yet the more severely affected descendants emerge through subsequent maternal transmission.

TRAP **Do not infer full-mutation risk from "X-linked" alone.** The relevant question is whether the premutation is being transmitted through a woman, because that is the route in which expansion to a full mutation occurs.

The historical pattern of worsening expression in successive generations is called the **Sherman paradox**. It was an observation before it had a molecular explanation. The repeat-expansion mechanism now turns that observation into a counselling problem: a pedigree should be drawn across generations, noting cognitive and developmental histories as well as tremor, ataxia and early ovarian failure in relatives. Recurrence counselling should be syndrome-specific and undertaken with clinical genetics support; the generic framework belongs in the etiological assessment discussion.

- Ask which side of the family carries the molecular result and whether transmission has been maternal or paternal.
- Obtain an extended pedigree history rather than stopping at developmental delay in the child.
- Explain that a premutation and a full mutation have different clinical implications.
- Offer cascade evaluation to relatives through an appropriately supported genetics service.

9.3 Population burden and the diagnostic place of fragile X

Published prevalence estimates differ because ascertainment and molecular methods have changed. A sensible examination answer is to state the estimate with its denominator: fragile X syndrome occurs in about 1 in 4,000 males and around 1 in 8,000 to 1 in 9,000 women. Female prevalence is described as approximately one-half of the male rate. These figures refer to the syndrome, not to premutation carriage, which is much more frequent and has a different sex distribution.

1 in 4,000	1 in 8,000-9,000	~40%	~20%
MALES WITH SYNDROME	WOMEN WITH SYNDROME	MALE PREMUTATION CARRIERS WITH FXTAS	FEMALE PREMUTATION CARRIERS WITH POF

The distinction between affected syndrome and carrier state changes the conversation with families. Premutation carriage is reported at 1 in 150-200 females and 1 in 400-450 males in the general population. It should not be quoted as the prevalence of fragile X syndrome, nor should syndrome prevalence be used to dismiss the relevance of a premutation in an extended family.

Fragile X is also clinically relevant in autism assessment. It is described as the most commonly known single genetic cause of autism spectrum disorder. The direction of the statistic is the trap:

the proportion of people with fragile X who meet autism criteria is not the same as the proportion of autism attributable to fragile X. Reported rates of autism-spectrum diagnosis within fragile X vary substantially with the diagnostic construct and method, so they should be read as evidence of overlap rather than as a single invariant percentage. The medical and genetic workup of autism is addressed in the autism workup section.

9.4 Recognising the phenotype without overcalling it

The physical phenotype is developmental and variable. In adults, the familiar pattern includes an elongated face, prominent or cupped ears, a high arched palate and connective-tissue features. **Large anteverted ears, smooth velvet-like skin, hyperextensible finger joints, flat feet and mitral valve prolapse** are the useful connective-tissue cluster. Macro-orchidism becomes relevant after pubertal development and should be asked about and examined for sensitively, with appropriate consent and privacy.

ADULT PHYSICAL FEATURE	APPROXIMATE FREQUENCY AMONG ADULT CASES	CLINICAL USE
Macro-orchidism	~90%	Most evident after puberty
Elongated face	~80%	Supports, but never establishes, suspicion
Prominent or cupped ears	~65%	Look for it in the broader phenotype
High arched palate	~60%	Examine as part of a general dysmorphology review
Flat feet	~60%	Connective-tissue clue rather than a diagnostic sign

GOOD TO REMEMBER

These are group frequencies, not a checklist diagnosis. Up to 20% of affected children may have no obvious physical features, while individual facial or connective-tissue traits occur in many people without the syndrome. Developmental history, family history and a coherent behavioural phenotype raise suspicion; molecular testing establishes the diagnosis.

Associated physical morbidity deserves ordinary clinical attention. Seizure disorder occurs in 20% of individuals with fragile X syndrome. Visual and hearing assessment, cardiac review when indicated, and review of neurological symptoms should be integrated into care rather than deferred because the presenting concern is developmental. The specific diagnostic approach to epilepsy in autism belongs to the autism medical-workup section, but a seizure history in fragile X is directly relevant to this syndrome review.

9.5 Cognitive, language and behavioural profile

In males with a full mutation, intellectual disability is often moderate, with an average IQ of **about 40**; individual functioning is nevertheless broad and must not be assumed from a molecular result. In females with a full mutation, random X inactivation offers partial protection,

and cognitive impact is correspondingly variable. The assessment of IQ, adaptive behaviour and intellectual-disability severity is covered elsewhere in this chapter; here, the key point is that the fragile X profile is uneven, not merely globally delayed.

Short-term memory, visuomotor coordination, arithmetic and sequential processing are relative weaknesses, while simultaneous processing and long-term storage or retrieval may be relative strengths. This matters clinically. A child may do better when information is presented as a whole visual structure than when it is delivered as a long serial verbal sequence. It also matters in the consultation: apparently inconsistent performance can be a profile effect rather than evidence of poor effort or oppositionality.

The behavioural phenotype has a distinctive social-anxious quality. **Gaze aversion or reduced eye contact, shyness and social anxiety, hand-flapping and repetitive mannerisms, hyperactivity with short attention span, and poor peer relations** can co-occur. These features invite diagnostic shadowing in both directions: gaze avoidance and repetitive behaviour can be assumed to establish autism, while an established genetic diagnosis can lead the clinician to overlook a co-occurring autism-spectrum condition. Evaluate the person's actual social communication, restricted behaviour and functional needs rather than using the syndrome label as a shortcut.

Disruptive behaviour is reported to peak in preschool years and to decline progressively with age, although attentional difficulties may remain. This course is useful when setting family expectations: a difficult early behavioural phase does not predict a static trajectory. ADHD itself and its formal assessment are dealt with in the ADHD and specific learning/communication disorders notes. In fragile X, practical supports should take account of attention, anxiety, sensory responses, communication and the relative cognitive profile rather than treating "behaviour" as a single symptom.

MNEMONIC

FRAME the consultation

Family transmission, **R**epeat category, **A**ppearance across development, **M**ind and behavioural profile, **E**xtended-family premutation effects. This is a clinical sequence, not a diagnostic score.

9.6 Females, mosaicism and the premutation family phenotype

Female presentation requires particular care because it is often quieter and more variable. **Random X inactivation (lyonization)** protects many full-mutation females from full clinical expression. About half of females with the full mutation show cognitive deficits. A clinician should therefore neither assume that all female relatives are unaffected nor equate absence of intellectual disability with absence of a meaningful molecular result.

Mosaicism is described in 15-20% of individuals with FMR1 mutations. Different cell lines can carry different molecular profiles, which is one reason phenotype, repeat length and methylation pattern may not align in a simple linear fashion. The clinical response to an apparently

discordant result is not to reject it, but to interpret it with the reporting laboratory and genetics team in the context of the person's functioning and family history.

Premutation-associated disorders are not peripheral footnotes. **FXTAS (fragile X-associated tremor/ataxia syndrome)** is a late-onset neurodegenerative syndrome in premutation carriers, characterised by intention tremor and cerebellar ataxia, with later neuropathy, autonomic symptoms and cognitive or behavioural change in some individuals. Its median onset is 60 years. When an adult relative of a child with fragile X reports these symptoms, the family history has become a neurological referral issue, not merely background information.

Female premutation carriers may also develop premature ovarian failure, also termed fragile X-associated primary ovarian insufficiency. Around **20%** are reported to develop premature ovarian failure. The risk does not rise in a simple straight line with repeat size, so counselling should avoid a crude "more repeats always means more ovarian risk" message. Reproductive, neurological and developmental implications should be held together in family counselling while respecting the different needs of each relative.

9.7 Confirmation, interpretation and management

Confirmation is molecular, not morphological. The diagnostic method of choice is **DNA-based fragile X repeat-length testing, using PCR and/or Southern blot methods including repeat-primed PCR**. Cytogenetic fragile-site methods are historical: culturing lymphocytes without sufficient folic acid made an apparent X-chromosome breakpoint visible, but that laboratory phenomenon is not a treatment rationale. In particular, folic acid should not be prescribed as therapy for the primary disorder on the basis of the old cytogenetic technique.

Testing is especially appropriate when intellectual disability is unexplained, when the developmental and behavioural pattern is suggestive, or when the pedigree contains compatible developmental, neurological or reproductive histories. The broader etiological workup for intellectual disability is covered in the causes-and-workup discussion; this section's responsibility is to ensure that the fragile X assay is a dedicated repeat-expansion test. Routine sequencing alone may not detect repeat expansions, so the test request must match the suspected mechanism.

The written report should be read as a molecular category, not merely as a positive or negative result. A full mutation directs attention to the developmental phenotype and its supports; a premutation redirects the family conversation toward transmission, reproductive implications and later neurological symptoms. Mosaic findings require correlation rather than dismissal. This interpretive step protects against two opposite errors: making the molecular result the whole explanation for a person's needs, or treating a clinically relevant finding as incidental because the current phenotype is subtle.

TRAP **A normal-looking child does not exclude fragile X, and a standard sequencing result is not automatically a fragile X test.** Suspect from the phenotype and pedigree; confirm with the repeat-expansion assay.

There is no established treatment for the primary disorder; management is of associated conditions. This is not therapeutic nihilism. It means that care is organised around developmental assessment, communication support, educational adaptation, treatment of target symptoms and associated medical conditions, and genetic counselling for the family. Developmental interventions and educational planning belong to the management section of this chapter. A syndrome diagnosis should improve the precision of that plan, not replace person-centred assessment.

Must remember: Fragile X is an X-linked repeat-expansion disorder in which maternal transmission can convert a premutation into a full mutation; full mutation silences FMR1 and reduces FMRP, whereas premutation disorders are mediated by toxic excess messenger RNA.

10 · Inborn errors of metabolism causing ID (PKU and others)

Why this group earns disproportionate attention. Inborn errors of metabolism are individually uncommon causes of intellectual disability, but they matter because a metabolic diagnosis may change the trajectory of a child and the advice offered to the family. The useful clinical question is not whether every person with developmental difficulty has a metabolic disorder. It is whether the pattern contains enough biological coherence to make a potentially modifiable cause worth pursuing. For the psychiatrist, these disorders are also a reminder that intellectual disability can be the long-term expression of a disturbance in a biochemical pathway rather than a fixed label detached from mechanism.

Inborn errors of metabolism are best approached as disorders in which a missing or ineffective metabolic step has consequences for the developing brain and, often, for other organs. Their psychiatric relevance lies in the combination of developmental phenotype, neurological signs, behaviour, systemic clues and the possibility of pathway-directed treatment. A good answer therefore does more than list syndromes: it links substrate, enzyme, clinical pattern and the therapeutic implication.

1 in 10,000 to 1 in 20,000	first 3 months	below 20%
PKU BIRTH INCIDENCE	LATEST WINDOW TO BEGIN PKU DIETARY RESTRICTION	ADULTS WITH PKU PERHAPS MAINTAINING DIETARY CONTROL

10.1 Clinical frame: think pathway, phenotype and treatability

Metabolic disorders should not be reduced to an examination catalogue. They are a way of thinking about causation. A child may have global developmental difficulty or established intellectual disability as one part of a wider disorder, while another person may present later to psychiatry because attentional, behavioural or affective symptoms have become more conspicuous than the developmental history. The history and examination should therefore remain alert to discordance: a developmental presentation that sits alongside altered growth, pigment, eyes, skeleton, movement, episodic illness or a distinctive form of self-injury deserves a broader explanatory frame.

The pattern matters more than any isolated sign. A physical clue is not merely a dysmorphology spotter's flourish; it can identify the organ system in which the metabolic disturbance is declaring itself. Similarly, an apparently psychiatric symptom may be a direct consequence of altered neurochemistry, a response to neurological disability, or both. This is why a formulation should preserve the medical diagnosis, the developmental phenotype and the current mental-state problem as separate but connected layers.

RULE **Define the problem by the disrupted pathway, then ask what can still be modified.** The same developmental endpoint can arise from very different biochemical mechanisms, and treatment must follow the mechanism rather than the behavioural surface.

There is an important boundary here. The detailed sequence of aetiological tests and the general genetic-counselling process belong to the wider intellectual-disability assessment. This section instead explains the recognised metabolic conditions and the clinical reasoning that makes their recognition consequential. When a metabolic disorder is known or suspected, psychiatric care works alongside paediatrics and metabolic medicine, rather than attempting to translate a complex behavioural presentation into a purely psychiatric diagnosis.

10.2 Phenylketonuria: the prototype of preventable metabolic neurodevelopmental injury

Phenylketonuria (PKU) is an autosomal recessive disorder and remains the classic example of a biochemical defect in which early recognition changes neurodevelopmental outcome. It is described as the commonest inborn error of metabolism. Textbook estimates give an incidence between **1 in 10,000 and 1 in 20,000 births**; a further estimate is 1 in 10,000 live births. These figures are useful as scale, but the clinical lesson is larger: rarity at the level of an individual clinic does not make a reversible diagnosis unimportant.

The primary defect is deficiency of **phenylalanine hydroxylase**, a hepatic enzyme. In ordinary biochemical terms, this enzyme mediates conversion of phenylalanine to tyrosine. The relevant gene is the PAH gene, located on chromosome 12, with a reported locus at 12q22-24.1. This is not genetic detail for its own sake. It explains why the disorder is familial in pattern and why a diagnosis has implications beyond the presenting child.

When this metabolic conversion fails, the brain is affected through more than one route. Low tyrosine contributes to depletion of central monoamines, including dopamine and serotonin. Reduced availability of large neutral amino acids also compromises synaptogenesis and myelination. That dual mechanism makes sense of a phenotype that crosses developmental cognition, attention, executive control and behaviour. It also prevents a simplistic conclusion that a child who survives the early developmental period without profound disability has necessarily escaped neurocognitive consequences.

MNEMONIC

PKU clinical spine: pathway, prevention, persistence

Start with the failed phenylalanine-to-tyrosine step; connect it to early dietary treatment; then remember that later executive, attentional and affective symptoms still deserve active review.

10.3 PKU phenotype: untreated disease and the treated survivor are different clinical questions

Untreated PKU is associated with intellectual disability and developmental delay. The recognisable untreated phenotype may include reduced pigment with fair or blond hair and blue eyes, microcephaly, eczema, epilepsy, growth retardation and hyperactivity. These features should be understood as clinical clues within a syndrome, not as a diagnostic shortcut. No one sign establishes the diagnosis, and a psychiatric assessment should not turn a physical sign into a substitute for biochemical confirmation.

The name itself reflects the secondary pathway. **Phenylketones**, produced through phenylpyruvic acid in the secondary pathway, are excreted in urine. This is a compact but useful examination link between the disease name and its biochemistry. It also illustrates a broader metabolic principle: once the usual route is blocked, diversion products can become clinically informative.

The early-treated adult or adolescent requires a different formulation from the untreated child described in older accounts. Even with early treatment, there can be subtle reductions in IQ with executive and attentional impairment. The psychiatrist should therefore ask concrete functional questions about planning, sustained attention, error monitoring, academic or occupational organisation, adherence to a demanding diet and the emotional burden of living with a lifelong disorder. This avoids two equal errors: attributing every difficulty to PKU, or dismissing cognitive and emotional concerns because gross intellectual disability has been prevented.

TRAP **Examination trap:** do not present early-treated PKU as synonymous with absence of neuropsychiatric morbidity. Early treatment changes the expected phenotype; it does not make follow-up unnecessary.

There is evidence of elevated depression, anxiety disorders, particularly agoraphobia, and attention-deficit disorder in early-treated adolescents with PKU compared with matched healthy individuals. In practice, this means that a symptom review needs ordinary psychiatric discipline. Establish the symptom's time course and impairment, consider medical and psychosocial contributors, and do not write off distress as an inevitable accompaniment of a metabolic label.

10.4 PKU screening and treatment: timing is part of the treatment

PKU illustrates why a metabolic diagnosis must be made early enough for the intervention to matter. Neonatal screening uses blood or urine. For prevention of severe, irreversible cognitive deficit, dietary phenylalanine restriction must begin within the **first 3 months of life**. The phrasing is deliberately temporal: a correct diet begun too late cannot be assumed to restore an already altered developmental pathway.

A low-phenylalanine diet maintained for at least the **first decade of life** is associated with an essentially normal cognitive outcome. The diet is not a casual instruction to avoid a food group. It is a sustained medical treatment whose implementation, monitoring and acceptability need specialist support.

IN INDIA

In the Indian Psychiatric Society clinical practice guidance, tetrahydrobiopterin with a low-phenylalanine diet is listed as a small-molecule intervention for PKU.

Longitudinal care needs to acknowledge that adherence can become difficult when parents no longer organise every meal and social life broadens. Perhaps **below 20%** of adults with PKU maintain dietary control throughout adulthood. This is not a reason to moralise about non-adherence. It is a reason to explore practical constraints, autonomy, cognition, family support, mood and the person's own understanding of risk. The psychiatrist's role includes making these barriers discussable while ensuring that metabolic management remains with the specialist team.

The broader lesson is that screening, diagnosis and treatment cannot be separated into unrelated facts. In PKU, the specimen identifies the condition, the biochemical pathway explains the diet, and the time window explains why developmental prevention is possible. That integrated chain is what candidates should reproduce rather than a bare list of enzyme and symptoms.

10.5 Homocystinuria: systemic clues should change the psychiatric formulation

Homocystinuria is an important contrast to PKU because the developmental presentation sits alongside striking extra-central-nervous-system findings. These include ocular lens dislocation, pectus excavatum, and a tall, thin, long-limbed Marfan-like stature. It is also associated with strokes in young and middle-aged adults.

GOOD TO REMEMBER

In a psychiatric setting, this combination should prevent diagnostic compartmentalisation: intellectual disability, compulsive symptoms or altered personality should not obscure systemic vascular and physical risk.

Intellectual disability occurs in almost all cases, and the behavioural phenotype can include obsessive-compulsive symptoms and personality disorders. These associations do not permit a psychiatrist to infer homocystinuria from an obsessive-compulsive presentation. They do, however, show why apparently ordinary phenomenology becomes diagnostically different when it appears with developmental and somatic clues.

Treatment may include pyridoxine, or vitamin B6, which increases homocysteine metabolism and reduces methionine and homocysteine. The Indian guidance also lists pyridoxine with vitamin B12 and folate among small-molecule approaches. Pyridoxine reduces these biochemical levels in approximately **50%** of patients. This is a treatment-response fact, not a licence for a psychiatrist to manage metabolic therapy independently.

A particularly useful negative finding is that folate plus vitamin B6 does not reduce stroke risk in adults with elevated homocysteine. The distinction matters because biochemical improvement and clinical vascular protection cannot simply be treated as interchangeable outcomes. It is the kind of nuance that strengthens an answer and, more importantly, prevents overconfident counselling.

10.6 Other named metabolic disorders: recognise the syndromic pattern

DISORDER	METABOLIC BASIS	CLUES THAT SHOULD REDIRECT THE FORMULATION	SPECIFIC THERAPEUTIC IMPLICATION
Galactosaemia	Autosomal recessive deficiency of one of three enzymes in galactose metabolism, including GALT	After milk introduction: failure to thrive, hepatosplenomegaly and cataracts	Galactose-free diet; if treated, intellectual disability is usually mild.
Lesch-Nyhan syndrome	X-linked mutation of the HGPRT gene causing a purine-metabolism defect with excessive uric acid production and excretion	Severe self-injurious behaviour with compulsive biting of fingers and lips, with spastic cerebral palsy and choreoathetoid movements	Behavioural risk management must be integrated with neurological and metabolic care.
Hurler syndrome (MPS I)	Autosomal recessive alpha-L-iduronidase deficiency	Storage disorder phenotype requires a multisystem formulation rather than an ID-only label.	Enzyme replacement for MPS and bone marrow transplantation for MPS I are listed interventions.

Galactosaemia teaches the importance of a feeding-linked systemic presentation. The triad of failure to thrive, hepatosplenomegaly and cataracts after milk introduction should redirect attention from an isolated developmental formulation toward galactose metabolism. The therapeutic link is equally direct: removing galactose is an intervention aimed at the pathway, not merely at a developmental consequence.

Lesch-Nyhan syndrome is especially memorable because the self-injury is severe and compulsive, rather than a generic behaviour problem. The child may be normal at birth before developing low-to-mild intellectual disability. The associated movement disorder and self-biting should prompt a syndrome-level formulation. In the consultation room, the practical priority is safety without losing explanatory precision: pain, communication difficulty, environmental response and the underlying neurobiological disorder can all shape the observed behaviour.

For the mucopolysaccharidoses, **Hunter syndrome (MPS II)** and Sanfilippo syndrome (MPS III) are both described as milder than Hurler syndrome (MPS I). The examination value is to recognise that MPS is a family of disorders rather than an eponym to be memorised in isolation. The clinical value is to retain the possibility that a developmental and behavioural presentation belongs to a multisystem storage disorder.

10.7 Treatability across metabolic disorders: use a mechanism-led therapeutic map

The key contrast across these conditions is not simply treated versus untreated. It is the kind of intervention that fits the disrupted biology. The Indian Psychiatric Society guidance identifies special or modified diets for many organic acidurias and aminoacidopathies, including PKU, glutaric aciduria type 1 and maple syrup urine disease. This should be read as an illustrative list, not as a claim that every metabolic disorder responds to dietary restriction.

- Replacement of deficient molecules includes thyroxine for hypothyroidism, enzyme replacement for mucopolysaccharidoses and copper histidine for Menkes disease.
- Small-molecule therapy includes creatine monohydrate for cerebral creatine deficiency syndromes and pyridoxine, vitamin B12 and folate for homocystinuria.
- Bone marrow transplantation is listed for alpha-mannosidosis and MPS I.
- Pharmacotherapy with vigabatrin and chelation of excess metals for Wilson disease and manganese transporter deficiency are further listed approaches.

These modalities are conceptually useful because they prevent therapeutic nihilism while also preventing indiscriminate treatment. A diet is appropriate when the disease mechanism supports dietary modification; replacement is appropriate when a missing molecule can be supplied; chelation addresses a harmful excess. The psychiatrist needs this map to understand why treatment is being offered and to recognise how psychiatric symptoms, adherence and family burden may affect it. Selecting the regimen, monitoring response and managing disease-specific risk remain specialist tasks.

> When intellectual disability is linked to a metabolic pathway, the decisive question is whether recognition permits pathway-directed treatment before or alongside irreversible developmental consequences.

10.8 Psychiatric practice: formulate, do not overshadow

Metabolic diagnosis should sharpen psychiatric assessment, not end it. A person may require assessment for depression, anxiety, attentional difficulty, obsessive-compulsive symptoms, personality change, severe self-injury or behavioural distress. The diagnosis helps the clinician ask better questions about timing, cognitive baseline, neurological change, diet, treatment burden and family interpretation. It must not become a blanket explanation that erases a treatable psychiatric syndrome or a preventable safeguarding problem.

Conversely, a familiar psychiatric label should not absorb clues that point beyond psychiatry. In PKU, executive and attentional impairment can coexist with mood and anxiety disorders. In homocystinuria, a psychiatric presentation sits beside ocular, skeletal and vascular clues. In Lesch-Nyhan syndrome, self-injury needs immediate compassionate management, yet its form and accompanying neurological signs carry diagnostic information. The senior clinical move is to hold both truths at once: symptoms deserve direct care, and their biological context may alter risk, referral and prognosis.

For examination writing, organise the answer around the prototype, then demonstrate range. Begin with PKU - inheritance, enzyme, biochemical block, phenotype, newborn screening and dietary treatment. Use homocystinuria to show why systemic findings and psychiatric symptoms must be integrated. Add galactosaemia, Lesch-Nyhan syndrome and mucopolysaccharidoses as concise pattern-recognition examples, and finish with the principle of mechanism-matched intervention. That organisation conveys clinical understanding rather than memorised accumulation.

11 · Fetal alcohol spectrum disorder

Why this diagnosis earns its place in an intellectual-disability chapter. Fetal alcohol spectrum disorder (FASD) is the clinical umbrella for developmental consequences attributed to prenatal alcohol exposure. It matters because the developmental phenotype is often broader than the visible phenotype: a child may have substantial difficulty with learning, regulation and everyday functioning without displaying the full facial pattern of fetal alcohol syndrome (FAS). Conversely, the recognisable physical pattern should prompt a careful account of neurodevelopment, rather than being treated as a diagnosis made from appearance alone. FAS is **probably the most common nongenetic cause of intellectual disability**, but FASD must not be reduced to intellectual disability alone.

The useful clinical stance is therefore developmental and longitudinal. Ask whether prenatal exposure is documented, look deliberately for the phenotype, establish whether growth or brain-development indicators are present, and describe the functional profile in its own right. The label should bring order to a scattered presentation; it should not become a shortcut that explains every behaviour, nor should an absent facial phenotype be used to dismiss a credible exposure-related neurodevelopmental disorder.

11.1 Scope, language and diagnostic lineage

FASD is an umbrella, not a synonym for FAS. Its subdiagnoses include FAS, partial FAS (PFAS), alcohol-related neurodevelopmental disorder (ARND), alcohol-related birth defects (ARBD), and neurobehavioural disorder associated with prenatal alcohol exposure (ND-PAE). This terminology is clinically useful because it separates a full syndromic presentation from partial physical phenotypes and from neurodevelopmental presentations in which physical signs are not demonstrable. ARBD is primarily a physical diagnosis and is not the focus of this neurodevelopmental discussion.

The original FAS criteria were set out by **Jones and Smith in 1973**. Contemporary clinical criteria are commonly described as the **Hoyme criteria**, updated from earlier Institute of Medicine criteria. For examination answers, the important point is not to recite a lineage of labels. It is to show that FASD diagnosis is organised around a coherent set of diagnostic domains: prenatal alcohol exposure, facial anomalies, growth deficiency, deficient brain growth or abnormal morphogenesis, and neurobehavioural impairment. These domains provide the framework for deciding which diagnosis best fits the available evidence.

RULE **Diagnostic principle.** In FASD, exposure history, physical findings and neurodevelopmental findings are interpreted together; none should be allowed to stand in for the others.

This framework also explains an apparently counterintuitive feature of the full syndrome. A diagnosis of **FAS** rests on the characteristic physical and neurobehavioural domain combination, so documentation of confirmed prenatal alcohol exposure is not required for that diagnosis. That is not permission to treat exposure history as unimportant. It reflects the specificity attributed to the combination when it is present, while recognising that reliable retrospective histories can be unavailable.

MNEMONIC

Use the spine, not a checklist blur

Think *exposure, face, growth, brain, behaviour*. It keeps the assessment ordered and makes it less likely that a clinician will mistake a single facial observation or a behavioural complaint for the whole diagnosis.

11.2 Biology and the syndromic phenotype

Alcohol is a developmental teratogen, so the clinical phenotype should be understood as the outcome of disruption during development rather than as a static cosmetic syndrome. A proposed mechanism is inhibition of **NMDA receptors** that mediate postsynaptic excitatory effects of glutamate, with effects on cell proliferation. This provides a biologically intelligible bridge between structural findings and later developmental difficulties. It does not, however, permit one to infer a particular cognitive profile from mechanism alone. Developmental outcomes are heterogeneous, and clinical formulation remains more informative than mechanistic shorthand.

Recognising the face requires discipline. The classic sentinel pattern is a **triad of short palpebral fissures, a smooth philtrum and a thin upper lip**. These are midfacial findings, not a general impression that a child "looks dysmorphic". The examiner should observe and document each component carefully, because casual facial judgement is vulnerable to expectation, ancestry-related variation and poor-quality photographs. In the facial-anomaly domain, **at least two** characteristic features are required. A palpebral fissure length at or below the **10th centile** qualifies as a facial feature; a thin vermilion border or smooth philtrum is assessed on a racially normed lip-philtrum guide at **rank 4 or 5**.

TRAP **Examination trap.** Do not diagnose FASD from a nonspecific statement such as "typical facies". State the individual facial findings, then relate them to the wider diagnostic domains.

Growth and brain-development indicators add clinical weight when present. Growth deficiency is represented by height or weight at or below the 10th centile. Deficient brain growth or abnormal morphogenesis may be reflected by head circumference at or below the 10th centile, structural brain abnormalities, or recurrent nonfebrile seizures. These are diagnostic indicators, not interchangeable measures of the same process. A growth measurement answers a different

question from head growth, and neither replaces a careful account of neurobehavioural functioning.

DIAGNOSTIC DOMAIN	WHAT THE CLINICIAN ESTABLISHES	WHY IT CHANGES REASONING
Prenatal alcohol exposure	A reliable history when it is available.	It is essential for some FASD diagnoses and strengthens causal attribution.
Facial anomalies	The sentinel midfacial pattern, measured rather than guessed.	It distinguishes a recognisable physical phenotype from a purely behavioural presentation.
Growth deficiency	Appropriate anthropometric evidence.	It contributes a separate developmental domain.
Brain growth or morphogenesis	Head-growth, structural or seizure indicators.	It links the physical assessment to neurodevelopmental risk.
Neurobehavioural impairment	Impairment on a global estimate or a relevant neurobehavioural or behavioural domain.	It prevents the diagnosis becoming a morphology exercise.

The threshold for neurobehavioural impairment in this clinical framework is performance at least 1.5 SD below the mean on a global cognitive estimate or on a single relevant neurobehavioural or behavioural domain. The key clinical inference is modest but important: a child can require a detailed functional evaluation even when the physical findings are incomplete. The physical phenotype and the developmental phenotype overlap, but they are not identical.

11.3 Distinguishing FAS, partial FAS and ARND

The diagnostic categories are best used as descriptions of the configuration before you, not as a hierarchy of "real" and "less real" disorders. FAS is the full syndromic pattern described above. PFAS retains facial anomalies and neurobehavioural impairment in the presence of confirmed exposure. When exposure is not confirmed, PFAS requires those findings together with impairment in either growth or brain growth. This additional requirement is logically necessary: without a documented exposure history, the clinician needs a more specific configuration before making an exposure-attributed diagnosis.

Alcohol-related neurodevelopmental disorder is made in the absence of the physical indicators of face and growth, but requires confirmed prenatal alcohol exposure and neurobehavioural impairment. This category is especially important because it captures children whose most disabling difficulties are cognitive, behavioural and functional rather than dysmorphic. Its existence protects against a common diagnostic error: assuming that the absence of the FAS face excludes alcohol-related neurodevelopmental harm.

- For FAS, look for the characteristic syndromic combination and do not make documented exposure a false prerequisite.
- For PFAS, decide first whether prenatal exposure is confirmed, because that changes the required domain configuration.

- For ARND, do not demand facial or growth indicators when a documented exposure history and neurobehavioural impairment are present.

These distinctions should improve the quality of documentation. Record what was observed, what was measured, which part of the exposure history is corroborated and which diagnostic domain is not established. That language is more clinically defensible than forcing a label from an incomplete record. It also keeps diagnostic uncertainty visible, which is preferable to either overcalling a syndrome or making a false reassurance from an absent physical feature.

11.4 ND-PAE: the proposed neurodevelopmental diagnosis

Neurobehavioural disorder associated with prenatal alcohol exposure is placed in the DSM appendix under Conditions for Further Study, with proposed rather than full diagnostic criteria. This status matters in a written answer: ND-PAE is a structured clinical construct, but it is not a full DSM diagnosis. It should therefore be described accurately rather than presented as though it had identical nosological standing to every routine DSM diagnosis.

Its exposure criterion specifies more-than-minimal gestational exposure as more than 13 drinks per month during pregnancy, or more than 2 drinks on any one occasion. This threshold is a criterion for the proposed diagnosis, not a declaration that exposure below it is safe. There is **no established safe threshold** or safe period of gestational alcohol exposure. The effects of exposure remain difficult to reduce to a single number because developmental stage and maternal and fetal factors modify risk.

The ND-PAE structure makes the clinical interview more precise. It requires impairment in at least one neurocognitive domain, at least one self-regulation domain, and at least two adaptive-functioning domains, one being communication or social communication and interaction. The neurocognitive domains include global intellectual performance, executive functioning, learning, memory and visual-spatial reasoning. The self-regulation domains include mood or behavioural regulation, attention and impulse control. Adaptive difficulties can involve communication, social communication and interaction, daily living skills or motor skills.

Global intellectual performance may be demonstrated by an IQ or comprehensive developmental standard score at or below 70. That is one route within the neurocognitive criterion, not the definition of the whole condition. A sound assessment asks which abilities fail in ordinary demands and whether the pattern coheres with the exposure history. Neurocognitive deficits, regulation problems and adaptive limitations are clinically separable, though they often compound one another in a child's life.

Diagnosis should be deferred at or below 3 years, because reliable neurocognitive assessment is limited at that stage. Deferral is not abandonment. It means recording exposure and developmental concerns, following the child's profile over time, and avoiding a prematurely fixed label. ND-PAE can be diagnosed in alcohol-exposed children with or without FAS, including those without facial dysmorphology. That independence is precisely why the clinician must inspect the neurodevelopmental phenotype rather than use the face as a gatekeeper.

11.5 Cognitive and behavioural clinical correlation

FASD creates a frequent conceptual trap in intellectual-disability teaching. The presence of FASD does not mean that every affected child has intellectual disability, and a normal-looking child should not be assumed to have intact higher functions. The majority of children with FASD do not have IQ below 70; observed ability spans from severe intellectual impairment to average performance. In children with FAS, the average full-scale IQ is around 72, compared with around 80 in children with heavy prenatal alcohol exposure without FAS. Intellectual disability in FAS is usually mild to moderate, though it may be severe.

Interpretation of intelligence testing itself belongs in the intellectual-disability assessment material.

GOOD TO REMEMBER

Do not let a single global score decide whether FASD is clinically important. A child with broadly higher measured ability may still struggle with executive demands, new learning, memory, visual-spatial reasoning, attention, impulse control or social communication. Translate these findings into difficulties at school, with home routines and in judgement; do not misread them as wilful noncompliance.

Psychiatric comorbidity adds a second layer of complexity. **Attention-deficit/hyperactivity disorder** is the most common co-occurring psychiatric diagnosis, with reported prevalence among children with FASD between 44% and 96%. Mental health problems have been identified in more than 90% of people with histories of significant prenatal alcohol exposure. These figures should not invite indiscriminate diagnostic labelling. They should prompt the clinician to ask whether attention, impulsivity and emotional dysregulation are being described carefully, rather than folded uncritically into a single explanatory story.

The absence of the classic face does not exclude an alcohol-related neurodevelopmental disorder; assess exposure history and functional phenotype together.

11.6 Differential diagnosis and disciplined attribution

Attribution is a clinical conclusion, not a moral judgement. Prenatal alcohol exposure may be difficult to establish and the behavioural presentation is not pathognomonic. The diagnostic task is therefore one of pattern recognition followed by active exclusion of alternatives. A credible history is valuable, but history alone cannot establish that every developmental difficulty is caused by alcohol. Equally, uncertainty in history should not lead to a careless assumption that a distinctive physical and neurobehavioural pattern is irrelevant.

The named differential includes genetic conditions such as Williams syndrome, Down syndrome and Cornelia de Lange syndrome, and other teratogenic conditions such as fetal hydantoin syndrome and maternal phenylketonuria. These conditions can share physical and behavioural characteristics with ND-PAE. The practical lesson is to seek positive evidence for the proposed

diagnosis and contradictory evidence for alternatives. “Alcohol exposure reported” is not a substitute for a differential diagnosis; “facial features absent” is not a substitute for one either.

TRAP **Examination trap.** A documented exposure history does not remove the need for differential diagnosis, and an undocumented history does not automatically rule out FAS when the required syndromic configuration is present.

- Separate observations from inferences: document anthropometry, facial examination, neurodevelopmental findings and exposure evidence distinctly.
- Ask whether each finding belongs to the FASD configuration or whether it suggests an alternative developmental or teratogenic condition.
- State the limits of the available evidence rather than converting an uncertain history into false certainty.

For a broader developmental formulation, use the relevant sections of the *Child psychiatry: assessment, treatment, services and protection notes*. Assessment of adaptive behaviour, intellectual testing and the multidisciplinary management of intellectual disability are deliberately treated elsewhere in these notes. Here, the point is narrower: recognise when alcohol-related developmental injury is a plausible organising diagnosis and describe why.

11.7 Epidemiology, prevention boundary and clinical significance

Reported prevalence varies greatly with population, case definition and completeness of assessment, so a single headline figure can mislead. The mean global general-population prevalence of FASD reported for 2012 was 7.7 per 1,000. The corresponding mean for the Americas was 8.8 per 1,000. In the United States, FASD has been estimated at between 1.1% and 5.0%, whereas the reported estimate for FAS itself is between 2 and 7 per 1,000 children. These are not competing answers. They measure differently framed populations and, crucially, the spectrum is wider than the full syndrome.

7.7 per 1,000	8.8 per 1,000	1.1% to 5.0%	251.5 per 1,000
FASD: GLOBAL MEAN	FASD: AMERICAS MEAN	FASD: UNITED STATES ESTIMATE	ND-PAE: CHILDREN IN CARE SETTINGS

Ascertainment is not a technical footnote. ND-PAE estimates in fully evaluated children have ranged between 31.1 and 98.5 per 1,000, and a meta-analytic estimate among children in care settings was 251.5 per 1,000. The latter should never be generalised to all children. It does, however, underline why a high-risk care context should lower the threshold for a thoughtful developmental history and a careful search for unmet needs.

The clinical preventive message is uncomplicated: there is no established safe threshold or safe period for alcohol exposure in pregnancy. The public-health framework for prevention is addressed in the *Prevention of intellectual disability* material. In this section, the practical diagnostic corollary is that clinicians should discuss exposure without blame. Shame drives

concealment, while accurate histories and respectful care improve recognition of children who need a fuller developmental evaluation.

12 · Tuberous sclerosis, neurofibromatosis and neurocutaneous syndromes in ID

Why these disorders matter in intellectual disability. Neurocutaneous syndromes make the aetiological assessment of intellectual disability clinically visible. The examination of skin, eyes and nervous system can connect a developmental presentation to a multisystem disorder, while the psychiatric formulation must remain alert to seizures, cognitive change, behaviour, family burden and the practical implications of a long-term condition. They reward disciplined observation: a lesion on the skin is not a diagnosis by itself, but it may be the clue that makes the whole developmental history intelligible.

These syndromes are especially useful for teaching this approach. **Tuberous sclerosis complex** brings developmental, epileptic and systemic manifestations into a formulation. **Neurofibromatosis type 1** likewise has cutaneous signs that may point to neurodevelopmental and neurological needs. The task for the psychiatrist is neither to reduce the patient to a syndrome nor to overlook the syndrome while treating only behaviour.

1 in 7000 TSC POPULATION PREVALENCE	**about 70%** TSC DE NOVO MUTATIONS	**above 90%** TSC SEIZURES AT PRESENTATION	**50%** NF1 NEW CASES IN UNAFFECTED FAMILIES

12.1 Neurocutaneous syndromes as a clinical frame

A neurocutaneous syndrome should be approached as a pattern across organ systems rather than as a dermatological label. In a person with intellectual disability, the clinician asks whether physical stigmata, epilepsy, cognition and change in functioning can be explained by a shared condition. This is useful because the consequences of an established syndrome are not merely diagnostic. It changes the threshold for collateral history, medical liaison and surveillance for new symptoms.

For tuberous sclerosis, older texts may use **epiloia** or tuberose sclerosis. The classical teaching spine comprises **three** linked manifestations: epilepsy, intellectual disability and facial angiofibromas on the malar surface. It remains a memorable clinical entry point, but it is not a substitute for examining the person or for recognising that features evolve across development. In particular, facial angiofibromas may first become apparent in adolescence; their absence earlier in life should not falsely reassure the clinician.

MNEMONIC

E-I-F for the classical TSC spine

epilepsy, intellectual disability and facial angiofibromas. Use it to prompt a whole-person review, not as a rule-out checklist.

RULE **When intellectual disability coexists with characteristic skin or neurological signs, formulate the syndrome as a multisystem condition and reassess for changing neurological function.**

Physical signs are most useful when they sharpen the examination rather than when they create diagnostic tunnel vision. In tuberous sclerosis, look deliberately for facial angiofibromas, hypomelanotic macules including ash-leaf spots, shagreen patches over the lower trunk or buttocks, ungual fibromas and retinal phakoma. These findings support a coherent clinical hypothesis, yet the psychiatrist should also document what they mean functionally: distress about appearance, stigma, pain, caregiving burden and barriers to education or work can all influence the presenting complaint.

12.2 Tuberous sclerosis complex: genetics and mechanism

Tuberous sclerosis is inherited in an **autosomal dominant** pattern, although many presentations are sporadic. What varies between affected people, and even within one family, is the **expressivity** of the disorder rather than its penetrance: current genetic sources put penetrance at essentially complete, so a relative who carries the pathogenic variant is affected, but the severity and organ pattern of that involvement differ widely. Older texts, including some in common examination use, describe this as variable penetrance. Say expressivity, and know why the distinction matters: it means a mildly affected parent is affected, not a non-penetrant carrier, and their apparently trivial skin sign is a reason to examine them properly. This matters when taking a family history: the history may be apparently negative without making a genetic disorder implausible. In conventional testing, an alteration in TSC1 or TSC2 is identified in 80% to 90% of people with the condition. The choice and sequence of tests belong to the ID aetiological work-up; the syndrome-specific result is interpreted alongside phenotype and family context rather than in isolation.

TSC1, located at 9q34, encodes hamartin. **TSC2**, located at 16p13, encodes tuberin. Their products act as tumour suppressors that restrain Rheb-mediated mTOR kinase activity. This molecular account is clinically valuable because it explains why mTOR-directed treatment has a rational place in selected manifestations; it does not imply that every cognitive or behavioural difficulty is directly reversible by treating a lesion.

The broad developmental lesson is that a molecular diagnosis describes risk and mechanism, not destiny. Cognitive ability in tuberous sclerosis has been reported as bimodally distributed, with a group having normal IQ and another in the profoundly affected range. Such heterogeneity makes an individual developmental and adaptive assessment indispensable. It also prevents the opposite errors of assuming that every person with the syndrome has intellectual disability, or overlooking substantial support needs in someone whose physical signs dominate the consultation.

GENETIC OR BIOLOGICAL LEVEL	CLINICAL MEANING	PSYCHIATRIC IMPLICATION
TSC1 and TSC2 encode hamartin and tuberin	The disorder has a defined tumour-suppressor pathway	Family history and physical findings should be integrated with the syndrome phenotype
The complex restrains mTOR activity	Selected structural manifestations have a pathway-based treatment rationale	Do not equate lesion-directed treatment with a complete explanation or cure for developmental needs
Variable penetrance and sporadic presentation	A negative family history does not settle the question	Give counselling within the wider genetic-work-up framework and avoid premature certainty

12.3 TSC phenotype: seeing the syndrome without missing the person

The skin examination is often the most accessible part of recognition. **Facial angiofibromas** are classically malar, while hypomelanotic macules, shagreen patches and ungual fibromas provide other cutaneous clues. Describe what is actually seen rather than using a label as shorthand. A careful record helps later clinicians distinguish longstanding stigmata from new change and allows the family to understand why a psychiatric consultation involves a full physical review.

Central nervous system lesions account for much of the developmental and neurological burden. Subependymal tubers may line the ventricles. Their reported size is **1 to 3 cm**, but the clinical question is not their size alone: one must ask whether a growing lesion could be producing seizures, headache, altered alertness, loss of skills or progressive cognitive impairment. **Subependymal giant cell astrocytomas** may compress and irritate adjacent cortex, and obstruction at the foramen of Monro can cause obstructive hydrocephalus. New deterioration should therefore trigger medical rather than purely behavioural attribution.

TRAP **Do not call a new behavioural change "part of the intellectual disability" before considering seizure activity, lesion-related change or hydrocephalus in a person with TSC.**

Systemic review is equally important. Tuberous sclerosis can involve renal cysts and angiomyolipomas, and cardiac rhabdomyomas that may cause dysrhythmias. These are not incidental facts for the psychiatrist. Pain, sleep disruption, repeated admissions, frightening cardiac symptoms and treatment demands can present indirectly as irritability, avoidance, low mood or family conflict. A liaison formulation distinguishes a psychiatric syndrome from a psychological response to a medical burden, while allowing both to be present.

Liaison also requires intellectual humility. Each specialty may see a valid but partial account of the person: the lesion, the seizure, the renal issue, the visible skin finding, the behaviour, or the family's exhaustion. The psychiatric assessment can integrate these accounts around daily functioning and distress. It should make uncertainty explicit, identify the next clinically answerable question and avoid implying that a behavioural description can settle a neurological concern.

For candidates, the practical inference is simple: findings in brain, skin, heart, eye and kidney make the syndrome multisystem. The specialist medical team directs organ surveillance. Psychiatry contributes longitudinal developmental description, a baseline of behaviour and cognition, assessment of emotional symptoms at the person's communication level, and support for carers faced with uncertainty.

12.4 Epilepsy, intellectual disability and behaviour in TSC

Epilepsy is central to the neuropsychiatric phenotype of tuberous sclerosis. Partial motor and complex partial seizures are particularly characteristic. Other seizure forms may occur, with classic absence seizures a probable exception; severe presentations may begin with infantile spasms. This seizure history should be taken with attention to onset, semiology, treatment response, injuries, sleep, postictal behaviour and apparent developmental plateau or loss. A family's account of "spells" may be much more informative than a single label in an old record.

Vigabatrin is particularly effective for infantile spasms in tuberous sclerosis. A randomised, single-masked trial in young children found the subgroup with TSC-related spasms particularly responsive. This is a responsiveness observation, not a licence to treat without specialist epilepsy oversight.

IN INDIA

The Indian psychiatric guideline also lists tuberous sclerosis among its relevant uses.

The limitation is clinically serious. Vigabatrin can cause progressive, permanent bilateral vision loss, including concentric visual field constriction. It has also been associated with behavioural disturbance or ADHD symptoms, depression and psychosis, with no beneficial psychotropic effect reported.

GOOD TO REMEMBER

When behaviour worsens after an antiseizure treatment change, the formulation must consider medication effect as well as seizure control, environment and the person's baseline vulnerabilities.

- Obtain a careful seizure and medication chronology before attributing agitation or decline to a primary psychiatric disorder.
- Ask carers for concrete examples of change in sleep, attention, mood, behaviour and daily functioning.
- Escalate new focal neurological symptoms or progressive cognitive change to the relevant medical team.

There are useful risk associations, but they must not become deterministic predictions. TSC2 mutations are associated with more severe epilepsy and cognitive impairment; moderate to severe intellectual disability is more common in people with infantile spasms. These associations support vigilance and early support planning; they do not replace an assessment of the individual's communication, strengths, behaviour or family resources.

TSC also has a limited reverse relationship with autism at population level: **less than 4%** of all autism is attributable to TSC. The point is diagnostic humility. TSC may be relevant to autism in an individual with the syndrome, but it does not explain autism generally, and autism assessment should still be conducted on its own clinical evidence.

12.5 TSC management: targeted treatment and longitudinal formulation

The psychiatric role in a multisystem syndrome is coordinative and longitudinal. First, identify the current problem in concrete terms: seizure-related risk, a change in cognition, behavioural disturbance, emotional symptoms, school or vocational difficulty, caregiver strain, or difficulty navigating multiple specialties. Next, establish the baseline against which change can be judged. Finally, communicate concerns in a form useful to neurology, paediatrics, dermatology, ophthalmology, cardiology and renal services as appropriate.

For subependymal giant cell astrocytomas, the mTOR inhibitors everolimus and sirolimus can shrink the lesion. Topically applied mTOR inhibitors can reduce facial angiofibromas. These examples clarify an important distinction: a medicine can be directed at a structural or skin manifestation, while the psychiatrist still addresses developmental support, distress, adherence, family understanding and the lived effects of a chronic disorder.

In TSC, a change in behaviour or cognition is a clinical event to explain, not a baseline feature to dismiss.

Generic genetic counselling principles and the sequence of laboratory investigation are addressed in the ID aetiological work-up. In this syndrome-specific setting, the clinician should explain that inheritance may be variable and that sporadic presentation is common, then ensure that the family receives appropriately coordinated counselling. Avoid giving a falsely precise recurrence discussion from a psychiatric interview alone, especially when the molecular result and the family phenotype have not yet been clarified.

12.6 Neurofibromatosis type 1: recognition and diagnostic reasoning

NF1 is associated with the tumour-suppressor gene product **neurofibromin**, at 17q11.2. Neurofibromin is involved in regulation of cell division and tumour suppression. As in TSC, the psychiatric significance lies in recognising a medical context for developmental and behavioural needs while retaining ordinary clinical discipline: a person can have NF1 and an independent mood, anxiety, psychotic or behavioural disorder.

Diagnosis usually requires **at least two** diagnostic features from the established list. The recognised features span skin lesions, peripheral tumours, regional freckling, ocular findings, optic-nerve involvement, bony lesions and an affected first-degree relative. The list is best used as a structured prompt for referral and documentation, not as an invitation for the psychiatrist to make a final diagnosis without appropriate examination.

NF1 FINDING DOMAIN	FEATURES TO LOOK FOR OR ELICIT	WHY IT MATTERS IN AN ID ASSESSMENT
Skin	**Café-au-lait macules**, axillary or inguinal freckling	May supply the first clue to a syndromic formulation
Peripheral tumours	Neurofibromas, including **plexiform neurofibroma**	Symptoms and visible difference may affect distress, self-image and help-seeking
Eye, bone and family history	Lisch nodules, optic nerve glioma, bony lesions, affected first-degree relative	Signals the need for appropriately coordinated specialist assessment

The café-au-lait threshold is a common recall point but should be applied with precision. The cited criterion specifies **six or more** lesions, larger than 5 mm before puberty or 15 mm after puberty. An alternative source frames the sign as more than six lesions, each larger than 5 mm in children and 1.5 cm in adults, and notes that individual café-au-lait spots occur in normal individuals. Therefore, a single lesion should not be over-interpreted; the pattern, associated signs and clinical context matter.

TRAP **Do not diagnose NF1 from an isolated café-au-lait spot, and do not mistake a referral criterion for a psychiatric explanation.**

12.7 A psychiatrist's practical synthesis

When a neurocutaneous syndrome is known or suspected in a person with intellectual disability, begin by separating the immediate clinical question from the aetiological label. Is the consultation about new seizures, progressive cognitive difficulty, aggression, emotional distress, medication adverse effects, family conflict or a support failure? That distinction prevents a syndrome name from becoming a catch-all explanation and makes the response clinically useful.

A good formulation has parallel layers. The biological layer records the established or suspected syndrome, current neurological concerns and treatment exposure. The developmental layer records the person's baseline abilities, communication and pattern of support. The psychological layer asks how the person understands symptoms, procedures, visible differences and restrictions. The social layer identifies who observes change, who carries care and what makes access to coordinated treatment difficult. Writing these layers separately helps the team avoid both diagnostic overshadowing and fragmented, organ-by-organ care.

Communication with families deserves the same precision as communication with specialists. Explain why a new symptom is being taken seriously, what the immediate safety concern is, and which clinician will address it. Invite carers to retain their observations rather than dismissing them as vague. In people with limited spoken language, an account of altered routine, sleep, facial expression, mobility, appetite, engagement or self-care may be the earliest available evidence of a medical or emotional change. The psychiatrist's value is often to translate this observation into an actionable question for the wider team.

- Examine and document relevant skin signs, while asking what they mean to the patient and family.
- Seek a longitudinal account of development, seizures, cognition, behaviour and treatment exposure.
- Compare the present state with the person's own baseline rather than with an abstract stereotype of the syndrome.

These disorders are a reminder that psychiatry in intellectual disability is whole-person medicine. The syndrome gives a framework for anticipation; it does not erase the individual's personality, preferences, communication style, social context or capacity for psychological distress. The best answer in the examination, and the best practice in the clinic, links a recognisable phenotype to a thoughtful developmental and psychiatric formulation without claiming more certainty than the evidence supports.

13 · Behavioural phenotypes of genetic syndromes

The useful question is not "what behaviour does this syndrome cause?" A behavioural phenotype is a clinically recognisable pattern that can sharpen observation, guide anticipatory enquiry and prevent diagnostic overshadowing. It is not a substitute for seeing the person in front of us. In intellectual disability, knowing the aetiology may provide **clues to psychiatric comorbidity**, because some syndromes are repeatedly associated with particular behavioural patterns. The practical gain is disciplined curiosity: ask about the pattern that is plausible, then establish whether it is actually present, distressing and functionally important.

The term **behavioural phenotype** was introduced by Nyhan. Earlier clinical observers such as Penrose had noted a distinctive social presentation in Down syndrome, but the modern concept is broader and more cautious. Its working definition is associated with Dykens: a syndrome is associated with a raised likelihood of a behavioural or developmental outcome when compared with people without that syndrome. That is a **probabilistic association**, not a diagnostic label attached to every individual.

13.1 What the concept does - and does not - mean

A behavioural phenotype is best understood as a pattern of vulnerabilities, strengths and recurrent clinical problems that occurs more often in a defined condition than outside it. This distinguishes a useful phenotype from an attractive anecdote. The clinician is looking for the **combination of features and its association with a condition**, rather than claiming that an isolated common symptom identifies a syndrome. Attentional difficulty, social anxiety, sleep disruption, stereotypy and self-injury all occur across diagnostic groups. Their meaning comes from their form, timing, associated features and clinical setting.

RULE **A behavioural phenotype changes prior probability; it does not determine the individual's behaviour.**

The concept explicitly rejects genetic determinism. Behavioural expression is shaped by developmental level, communication, physical illness, sensory experience, family responses, opportunity and the person's own temperament. A syndrome may create a particular vulnerability while the environment influences how often it is triggered, how it is understood and how much disability follows. This is why a behavioural phenotype should lead to more careful assessment, not to premature explanation.

Equally, no behavioural phenotype is invariant. The absence of a familiar feature should not be used to dismiss a molecular or clinical diagnosis; its presence should not be used to make one. This is especially important in adults, in people with co-occurring sensory or neurological problems, and where developmental history is incomplete. The phenotype is a clinical map, not a gatekeeping criterion.

Specificity lies on a spectrum. A broad feature may be shared across several syndromes, whereas a particular form of that behaviour may be unusually informative. For example, a particular form of stereotyped hand wringing is almost invariably seen in Rett syndrome, even though hand stereotypies occur in many clinical contexts. The distinction protects against the common error of treating the category "stereotypy" as though it carried the specificity of its most characteristic form.

TRAP **Do not turn a phenotype into a personality stereotype.** A recurring association may help formulate a question, but it never excuses failure to assess pain, communication, mood, safeguarding, environmental demands or co-occurring psychiatric illness.

13.2 How to use a phenotype in assessment

Use the syndrome to improve the interview, not to abbreviate it. Start with a baseline account of communication, adaptive functioning and the person's usual temperament. Then ask about the behaviours linked to the syndrome in a neutral, concrete way: their form, onset, antecedents, consequences and effect on family life, education, work and health. A behavioural signature has most value when it prompts a question that might otherwise not have been asked.

When a behaviour is established, the next task is formulation. Food-seeking, self-injury, withdrawal, agitation or sleep disruption can be part of a phenotype, but can also signal an untreated medical problem, a mismatch between demands and ability, a communication failure or a psychiatric disorder. The syndrome changes the differential weighting; it does not make competing explanations disappear. The assessment of psychiatric comorbidity in intellectual disability is addressed in the chapter's discussion of dual diagnosis, and the general assessment framework belongs in the Child psychiatry: assessment, treatment, services and protection notes.

- Describe the behaviour precisely before naming it as syndrome-related.
- Establish what has changed from the person's own baseline and what has changed around them.
- Look for the linked behavioural cluster, not an isolated headline symptom.
- Use the formulation to anticipate risk and support needs without attributing blame.

This approach matters to families. A **strong behavioural phenotype** describes a vulnerability that may be evident even in a supportive environment. In such situations, parents and teachers who have worked hard should not be invited to conclude that the behaviour proves a failure of care. The right message is more exact: environment still matters greatly for safety, distress and functioning, but it may not fully remove a syndrome-linked vulnerability.

The historical literature illustrates why this caution is necessary. Reviews in the 1970s discussed only **five** chromosomal syndromes in this context. The concept has since become clinically richer, but a larger catalogue does not make its inferences categorical. The contemporary task is to be specific enough to notice a pattern and humble enough not to overread it.

13.3 Cross-syndrome signatures: a working comparison

The table is a pattern-recognition aid, not a diagnostic algorithm. It deliberately pairs the most discriminating feature with the clinical caution that prevents overclaiming. Several entries contain symptoms that are common in many people with intellectual disability; their value comes from the surrounding phenotype.

SYNDROME	USEFUL BEHAVIOURAL OR COGNITIVE SIGNATURE	CLINICAL READING
Down syndrome	Relative strength in social communication, with a wide variety of behaviour.	Do not replace assessment with the old "very sociable" personality stereotype.
Fragile X syndrome	Gaze aversion, shyness, social and performance anxiety, social withdrawal and hyperarousal, often with attention and activity difficulties.	Social avoidance can coexist with interest in others and must not be equated automatically with autism.
Prader-Willi syndrome	Hyperphagia with food seeking, foraging or hoarding, and lifelong food preoccupation.	Food access and supervision are central environmental issues, while the wider formulation must also consider distress and routine.
Angelman syndrome	Episodic paroxysmal or inappropriate laughter with absence of speech.	The apparently happy expression should not conceal major communication needs.
Williams syndrome	High, sometimes indiscriminate sociability with reduced social flexibility.	Warm approach behaviour can coexist with poor peer relationships, anxiety and vulnerability in unfamiliar social settings.
Smith-Magenis syndrome	Severe self-injury, disruptive or aggressive outbursts, tantrums and hyperactivity.	Ask specifically about sleep, because disturbed sleep can amplify daytime behavioural difficulty.
Cri-du-chat syndrome	High-pitched cat-like cry in infancy.	This is an early developmental clue; later assessment should address language, attention and activity rather than treating the cry as a complete phenotype.

SYNDROME	USEFUL BEHAVIOURAL OR COGNITIVE SIGNATURE	CLINICAL READING
Lesch-Nyhan syndrome	Severe self-injury, typically compulsive biting of fingers and lips.	Its form and apparent compulsion make this a high-priority safety phenotype.
22q deletion syndrome	Childhood ADHD, autistic features and mood swings, with an important later psychosis vulnerability.	Developmental and psychiatric surveillance must extend across developmental transitions.
Rett syndrome	Loss of purposeful hand skills, replaced by stereotyped hand wringing and clapping.	The course and the exact hand phenomenology matter more than the label "stereotypy".
Cornelia de Lange syndrome	Self-injury, autistic features and pleasurable responses to vestibular stimulation.	Communication may be disproportionately impaired relative to adaptive function, which should shape the formulation.

Attentional problems demonstrate the point about shared symptoms: they are frequent in fragile X, Williams and cri-du-chat syndromes. They are therefore clinically important but not syndrome-specific. By contrast, relatively characteristic forms include the unusual hand behaviour in Rett syndrome and extreme hyperphagia in Prader-Willi syndrome. An examination answer is stronger when it says whether a feature is shared, relatively specific or simply one component of a wider pattern.

13.4 The phenotype is often richer than its headline feature

The classic "Down personality" stereotype is difficult to verify. A safer clinical account is that Down syndrome has a relative social-communication strength and a broad behavioural range. It is also associated with less frequent and less severe psychopathology than in other groups with intellectual disability, while still requiring ordinary clinical vigilance. In later life, the phenotype includes high rates of dementia or Alzheimer disease in middle-aged to late adulthood. The teaching point is not that people with Down syndrome are protected from psychiatric problems; it is that group-level tendencies should never flatten individual presentation.

Fragile X syndrome is often recognised through its anxious, avoidant social style. In full-mutation males, the tabled phenotype also includes anxiety, perseverative language, an ADHD-like presentation, aggression, stereotypies and ASD-like behaviours. Yet men with fragile X syndrome are usually affectionate and lack the aloof quality typical of autism, despite social impairment. This is a useful discriminator, not a rule that excludes autism. It directs the clinician to observe the quality of social engagement rather than inferring it from gaze aversion alone.

The food phenotype of Prader-Willi syndrome extends beyond hunger. Skin-picking, hoarding, concerns with symmetry, exactness, ordering, cleanliness and sameness, tantrums, aggression, stubbornness, underactivity and emotional lability may form part of the behavioural picture. Sleep also deserves active enquiry: excessive daytime sleepiness with sleep-architecture abnormalities can contribute to apparent inattention, irritability or reduced participation. The

assessment should separate an environmental response to restricted food access from a mood, anxiety or behavioural disorder, while recognising that these may coexist.

In Angelman syndrome, laughter is only the opening clue. Other observed features include overactivity with short attention span, enjoyment of social and physical contact, mouthing objects and fascination with water. The older label "happy puppet" arose from a particular combination of nonverbal presentation, ataxia, frequent laughter and flexed arms. It may help recall the history, but it is not respectful shorthand for the person's needs. The clinically relevant point is severe to profound intellectual disability with absence of speech, requiring an active search for nonverbal communication and distress signals.

Williams syndrome is another corrective to superficial impressions. It may show loquaciousness with "cocktail party manners" and a relative strength in language, auditory processing and music alongside poor visuospatial performance. However, friendliness may coexist with anxiety, including specific phobia and generalized anxiety. Treating verbal fluency as evidence of social comprehension is the trap. The person may initiate readily yet struggle with reciprocal flexibility, boundaries or sustained friendships.

13.5 Self-injury, stereotypy and sleep: read the mechanism before the behaviour

Self-injury is a description, not an explanation. Its diagnostic and safety significance depends on form, function and context. Lesch-Nyhan syndrome illustrates a highly characteristic pattern: stereotypic dystonic movements and self-mutilation of fingers, lip biting and other self-injury occur unless the individual is restrained. This is not interchangeable with all self-injury in neurodevelopmental conditions. It should prompt urgent attention to tissue protection, pain, oral injury, family burden and specialist coordination.

In Rett and Cornelia de Lange syndromes, self-injury may arise secondarily from hand-to-mouth stereotypies. The mechanism matters because it changes the clinical questions: is the injury a direct repetitive motor act, a response to distress, a communication of pain, or an outcome of a hand-to-mouth movement? The same visible lesion may therefore require very different prevention plans.

Contrasting source lists make a useful methodological warning. The DSM-5-TR names Lesch-Nyhan, Rett, fragile X, Cornelia de Lange and Smith-Magenis syndromes as examples of neurogenetic disorders with stereotypies. Another source instead names Cornelia de Lange, fragile X, Smith-Magenis, cri-du-chat and Rett syndromes in relation to stereotypic movements and self-injurious behaviour. Neither list should be treated as an exclusion list. They illustrate overlapping clinical territory, while the detailed phenotype supplies the discriminating information.

Smith-Magenis syndrome is especially instructive because its phenotype links daytime behaviour and sleep. Alongside self-injury and outbursts, a **self-hug** can be a useful signature gesture, and the sleep problem may include absence of REM sleep. The reported disruption of melatonergic control of the sleep cycle provides a plausible bridge between sleep management and behavioural

or cognitive functioning. This is not an invitation to prescribe from a syndrome name; it is a reminder that sleep assessment may be central rather than incidental.

MNEMONIC

PATTERN before label

Precise form, **A**ntecedents, **T**rajectory, **T**riggering pain or sleep problem, **E**ffect on function, **R**elated syndrome features, **N**eeds and safety.

13.6 Developmental trajectories and later psychiatric risk

Some phenotypes are defined as much by change over time as by a single behaviour. In Rett syndrome, loss of hand skills occurs with language regression, communication deficits, social withdrawal, hyperventilation or breath-holding, bruxism, sleep irregularities and some autistic symptoms. The early withdrawal should not be read as a fixed autism phenotype: socialization may later improve, with intense eye gaze in many patients. Rett syndrome itself is considered in the regression discussion elsewhere in this chapter; here, its course demonstrates why a behavioural phenotype must be longitudinal.

For 22q deletion syndrome, the crucial temporal pattern is a childhood developmental and behavioural presentation followed by risk of psychosis in adolescence or adulthood. About **25%** develop schizophreniform psychosis. At the same time, no other single risk factor is so strongly associated with psychosis. These findings belong together: striking association does not imply universal penetrance.

nearly 100%	**~25%**	**up to 30%**
OBESITY RISK IN PRADER-WILLI SYNDROME WITHOUT AGGRESSIVE DIETARY INTERVENTION	SCHIZOPHRENIFORM PSYCHOSIS IN 22Q DELETION SYNDROME	MAJOR PSYCHIATRIC ILLNESS IN VELOCARDIOFACIAL SYNDROME

The final statistic measures a broader outcome. In velocardiofacial syndrome, up to **30%** develop major psychiatric illness, a category that includes schizophrenia and bipolar disorder. It must not be substituted for the figure for schizophreniform psychosis. This is a classic exam error: the number may be familiar, yet the construct and denominator are wrong. Anticipatory follow-up should therefore ask about emerging psychotic and affective symptoms without assuming that developmental or autistic features are their explanation.

13.7 Communicating the formulation without blame or fatalism

A syndromic formulation can be relieving when it gives a family language for a pattern that had previously been interpreted as wilfulness, poor parenting or inevitable deterioration. It should be communicated as a vulnerability profile: “this condition makes this cluster more likely; we will look carefully at it and address what is modifiable.” That preserves realism and agency. It also makes room for strengths, relationships and the person’s own preferences, which a list of syndrome behaviours can easily erase.

GOOD TO REMEMBER

Use the phenotype to organise monitoring, environmental support and safety planning. For a food-related phenotype, explore access, supervision and conflict around food. For self-injury, define the topography, injuries, pain and immediate safeguarding needs. For a sleep-related phenotype, obtain a sleep history before interpreting daytime behaviour as purely psychiatric. For social disinhibition or withdrawal, assess vulnerability, consent, peer context and anxiety. These are clinical applications of the phenotype, not a generic management programme.

Be precise when discussing uncertainty. “Characteristic” does not mean inevitable; “shared” does not mean trivial; and “not specific” does not mean unimportant. The most defensible clinical note describes observed behaviour, names the syndrome-linked hypothesis, records alternative explanations and states what will be monitored. This reasoning protects the person from diagnostic overshadowing as well as from ignoring a syndrome-specific safety signal.

Must remember: A behavioural phenotype is a raised probability of a characteristic developmental or behavioural pattern, not genetic predestination. Read the whole cluster, its course and its context - then use it to anticipate needs without stereotyping the individual.

Living with intellectual disability

14 · Comorbid psychiatric disorders in ID (dual diagnosis)

Dual diagnosis is ordinary psychiatry, adapted rather than abandoned. In intellectual disability (ID), a psychiatric disorder may coexist with the developmental condition and must be sought as an additional clinical problem. In this setting, **dual diagnosis** means the coexistence of ID with an identifiable mental illness or personality disorder. The phrase originated in learning-disability psychiatry, although it has subsequently acquired a different substance-use meaning; state the context whenever using it.

The examination task is not to label every difficult behaviour as illness, nor to excuse illness as inevitable in ID. It is to establish whether there has been a meaningful change from the person's usual functioning, determine the function and context of that change, exclude physical and environmental explanations, and then decide whether a psychiatric syndrome is present. This is clinically consequential because communication differences can alter both the account available to the clinician and the outward form of distress.

3-5-fold	40%	9-18%
RELATIVE RISK OF ANY PSYCHIATRIC DISORDER	AROUND CUMULATIVE PREVALENCE	ESTIMATED ADHD IN ID

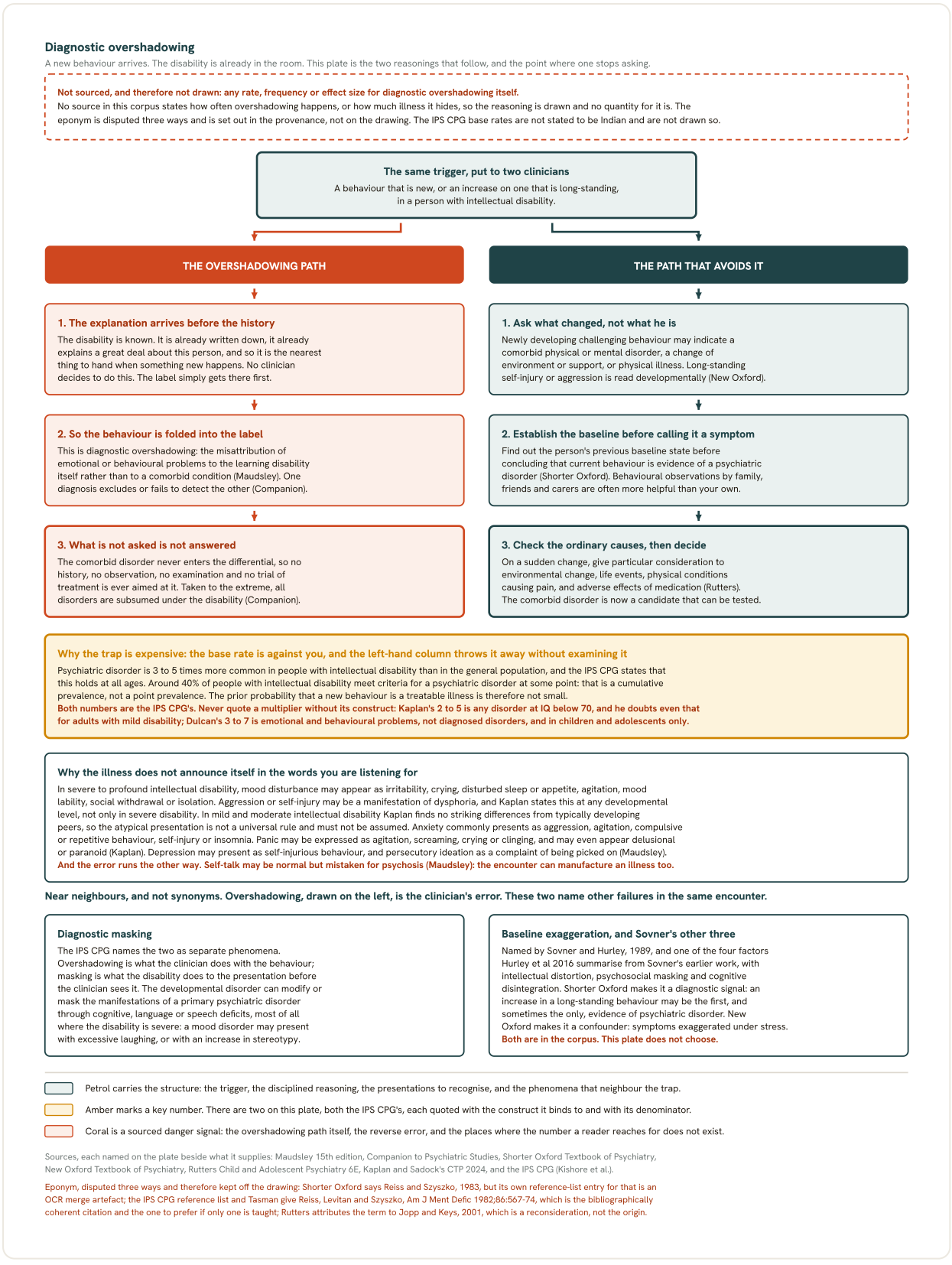


Fig 21.9 Diagnostic overshadowing, drawn as the reasoning it actually is. One trigger, a behaviour that is new or an increase on a long-standing one in a person with intellectual disability, is put to two clinicians, and the plate follows each of them through three steps to a different place. The amber strip is why the error is expensive: on the IPS CPG's figures psychiatric disorder is 3 to 5 times more common in intellectual disability and around 40% meet criteria at some point, a prior probability the left column discards. No rate for overshadowing itself is drawn, because no source in this corpus gives one.

14.1 Burden and the meaning of excess risk

Psychiatric comorbidity is not an occasional complication of ID. Across the lifespan, the relative risk of any psychiatric disorder is **3-5**-fold that in the general population. In children and adolescents, studies similarly report a 3-5-fold excess compared with typically developing peers. These figures should change the clinician's threshold for asking an additional psychiatric question, but not the threshold for making a diagnosis without evidence.

Prevalence estimates differ substantially between studies, with reported values ranging from **10-39%**. The variation is informative: samples differ in age, severity of disability, setting, informants, case definitions, and whether the measure captures a current disorder, symptoms, or cumulative morbidity. A rate is therefore a prompt to assess, not a shortcut to diagnosis. Historical comparisons also remind us that ascertainment matters. In an Isle of Wight comparison, psychiatric disorder was identified in **50%** of children with intellectual disability and in 7% of children with normal IQ.

Risk is unevenly distributed. The severity gradient reported across epidemiological studies means that psychiatric disorder is more often identified with greater intellectual impairment. By contrast, the evidence is less clear for excess prevalence in adults with mild ID. Child and family risk markers include more substantial intellectual impairment, adaptive-behaviour and language difficulties, poor socialisation, and adverse family social circumstances. These are not diagnostic criteria and not explanations that make a person's symptoms less worthy of assessment. A co-occurring autistic spectrum disorder is itself an important risk context for anxiety and depression, bipolar-spectrum presentations, severe obsessional behaviour, anger disorders, and psychosis-like episodes that do not meet schizophrenia criteria.

RULE **Rule:** treat a changed mental state or behaviour in ID as a clinical question with several possible answers, not as proof of either psychiatric illness or developmental disability.

14.2 Diagnostic overshadowing, masking and the baseline

The central diagnostic hazard is **diagnostic overshadowing**: emotional or behavioural problems are attributed to ID itself instead of being considered as a coexisting disorder. At its extreme, the developmental diagnosis becomes so dominant that a second condition is never detected. Overshadowing is a clinician error of attribution. It can lead to a missed depressive episode, psychosis, anxiety disorder, delirium, pain state, or an environmental crisis because the referral is prematurely translated into “part of the disability”.

Diagnostic masking is different. Here, a developmental disorder modifies or obscures the manifestations of a primary psychiatric illness through cognitive, language, or speech limitations, particularly when disability is more severe. The illness has not been dismissed as ID; rather, it is harder to recognise because its usual verbal and phenomenological markers are inaccessible or altered. A mood disturbance, for example, may appear as excessive laughter or an increase in stereotyped behaviours rather than as a verbally reported low mood.

A third concept is **baseline exaggeration**. An increase in a long-standing behaviour may be the first, and occasionally the only, evidence of psychiatric disorder. Thus an established repetitive

act, irritability, aggression, or self-injury should not be dismissed merely because it is familiar. The question is whether its frequency, intensity, setting, consequences, or controllability has changed. The baseline is not background detail. It is the reference point that converts a behaviour into a possible symptom.

TRAP **Examination trap:** "Self-injury in a person with ID" is a presenting problem, not a diagnosis. Do not infer a psychiatric syndrome from the behaviour alone, and do not use the developmental diagnosis to end the enquiry.

PROBLEM IN REASONING	WHAT GOES WRONG	CORRECTIVE CLINICAL MOVE
Diagnostic overshadowing	A new psychiatric or physical problem is assigned to ID.	Ask what is new, then actively consider coexisting illness.
Diagnostic masking	The primary disorder has an altered, less verbal presentation.	Use behaviour, chronology, and informants alongside the interview.
Baseline exaggeration	A familiar behaviour is treated as clinically meaningless.	Compare current frequency, intensity, context, and impact with baseline.
Intellectual distortion	Answers to mental-state questions are overinterpreted.	Adjust language and seek corroboration rather than forcing adult phenomenology.
Psychosocial masking	Developmental symptom expression is mistaken for mental illness.	Interpret the observation in developmental context before diagnosing.
Cognitive disintegration	Acute disorganisation is equated immediately with chronic psychosis.	Assess for an evolving illness, stressor, physical cause, and change in supports.

14.3 Why the mental-state examination needs translation

Increasing severity of ID and associated communication impairment make diagnosis more complex. This does not imply that psychiatric diagnosis is impossible. It means that the clinician must translate the clinical question from a reliance on subjective report to a synthesis of observable behaviour, longitudinal change, collateral history, environmental context, and the person's own communication at their best level.

One useful framework describes **four** sources of diagnostic uncertainty: baseline exaggeration, **intellectual distortion**, **psychosocial masking**, and **cognitive disintegration**. Intellectual distortion concerns how the person understands a question about mental state and whether its implication can be appreciated. Psychosocial masking, in this formulation, describes developmental symptomatology that is misconstrued as mental illness. Cognitive disintegration describes disproportionate disorganisation during an emerging mental illness when cognitive reserve is limited, which can make the person look apparently psychotic. These are prompts for better assessment, not alternative diagnoses.

Behavioural equivalents are particularly important when verbal description is limited. In severe to profound ID, mood disturbance may be signalled by irritability, crying, dysregulation of sleep or appetite, agitation, mood lability, withdrawal, or isolation. Aggression or self-injury may express dysphoria at any developmental level, but neither is specific to mood disorder. Anxiety may present with aggression, agitation, compulsive or repetitive behaviour, self-injury, or insomnia. Panic may appear as agitation, screaming, crying, or clinging and may even look paranoid or delusional to an unwary observer.

Conversely, unfamiliar does not mean pathological. Self-talk can be a normal behaviour that is mistaken for psychosis. A complaint of being picked on may be the accessible form of persecutory ideation, but it also needs ordinary contextual testing. The task is phenomenology by approximation: ascertain what the person experiences where possible, establish what others observe, and decide whether the whole pattern forms a syndrome over time. In mild to moderate ID, mood disorders do not show striking differences in expression from those of typically developing peers; do not exoticise a presentation simply because ID is present.

14.4 A disciplined assessment sequence

Begin with change, not with labels. Assessment begins with the individual's baseline and interprets change with collateral information, behavioural observation, and context.

> **GOOD TO REMEMBER**
>
> Before treating current behaviour as psychiatric evidence, establish the person's previous baseline state. Family, friends, and carers often provide observations more useful than the brief assessment encounter. Use the interview alongside a period of behavioural observation. From first contact, consider approach, activity level, attachment to the informant, response to transitions, spontaneous communication, affective range, and reaction to demands against that baseline.

Build the history around the following dimensions rather than around a checklist of adult symptoms:

- onset and chronological evolution of the change;
- intensity and frequency of the behaviour or symptom;
- the context in which it appears and what follows it;
- precipitating factors, including changes in relationships, routine, supports, or expectations;
- relieving factors and the response to ordinary environmental adjustment.

This sequence gives the history a causal structure. Newly developing challenging behaviour can indicate a comorbid physical or mental disorder, an environmental or support change, or physical illness. Long-standing self-injury or aggression is more often understood developmentally or behaviourally. When change is sudden, give particular consideration to environmental changes, life events, painful physical conditions, and medication adverse effects. In the most disabled person with sensory deficits, exclude physical illness and stressful events before considering whether communicative frustration and understimulation are contributing.

Antecedent-Behavior-Consequences analysis is a functional method for clarifying what precedes a behaviour, what the behaviour achieves, and what consequences may maintain it. It is not a substitute for diagnosis. It prevents a false dichotomy: a psychiatric disorder can coexist with behavioural reinforcement, and a behaviour with an understandable function can still be an expression of distress. Prospective simple ABC or sleep charts are particularly useful when the care environment produces discrepant staff reports, since variation may reflect differing tolerance thresholds or attributions.

> The most reliable discriminator is a documented departure from the individual's own baseline, interpreted with collateral observation and context.

14.5 Disorder patterns: recognise without overcalling

Detection of psychiatric syndromes in ID is shaped by communication, developmental level, and context. Schizophrenia should be diagnosed from a coherent longitudinal syndrome rather than from odd behaviour, self-talk, or a single report of suspiciousness. In a community sample of people with learning disability, point prevalence of schizophrenia was **over 4%**. Among people with learning disability and schizophrenia, auditory hallucinations are reported in around 90%; this is helpful when a reliable account can be obtained, but its absence or inaccessibility cannot settle the diagnosis.

Attention-deficit/hyperactivity disorder is also relevant: estimated prevalence among people with ID is 9-18%. The core diagnostic safeguard is developmental proportionality. Symptoms must be excessive for the person's mental age, and the threshold should be higher in severe to profound ID. Inattention, overactivity, impulsivity, poor frustration tolerance, and a chaotic environment can mimic each other. The clinician should seek a pervasive pattern rather than equating high activity in one setting with ADHD.

Anxiety estimates vary widely, from 1-25%, in part because diagnosis is difficult. This is a reminder to search for anxiety when distress is expressed behaviourally, not a basis for diagnosing it indiscriminately. Mood symptoms require the same restraint. Appetite and sleep change, social withdrawal, loss of usual enjoyment, tearfulness, agitation, or altered stereotyped behaviour gain significance when they are new, clustered, sustained, and not better explained by pain, medication, or altered circumstances.

TRAP **Examination trap:** do not diagnose psychosis from self-talk, depression from self-injury, or ADHD from activity level without establishing change, phenomenology, developmental proportionality, and context.

14.6 Instruments and adapted diagnostic systems

Questionnaires and structured interviews designed for children of average ability have a serious norming limitation in ID: almost none are normed for ID populations, and their scores or cut-offs may therefore be invalid. They may inflate symptoms or underestimate them depending on the setting. Use them as aids to gathering information, never as a rule-in or rule-out mechanism. This caution does not mean that all measures are unsuitable. The **Developmental Behavior**

Checklist is comprehensive and provides norms and subscales for the ID population, making it particularly useful where general-population measures fit poorly.

The clinical interview, observations, informant account, and functional data must retain primacy. ID-specific instruments may identify general psychopathology, yet studies do not show clear agreement between such instruments in detecting specific psychiatric disorders. A positive screen should trigger a more focused assessment; a negative screen should not override a persuasive longitudinal history. This is why an apparent numerical score is less important than the quality of the behavioural description from which it arose.

Modified diagnostic systems can support clinical reasoning where conventional criteria rely heavily on subjective experience or developmentally inappropriate thresholds. **DC-LD** was developed to address diagnostic issues in moderate to severe learning disability. **DM-ID** is another adapted diagnostic resource. Such systems do not create a separate psychiatry for ID; they make explicit the adaptations required to apply psychiatric nosology fairly. For eating disorders whose criteria depend substantially on subjective experience, conventional diagnoses such as anorexia nervosa and bulimia are effectively precluded in severe or profound ID. Pica is perhaps the most common eating disorder in this population.

14.7 Formulation and diagnostic accountability

A defensible formulation has parallel tracks. One track describes the person's developmental profile and baseline repertoire. A second describes the current change in behaviour, affect, cognition, biological rhythm, and functioning. A third examines physical illness, pain, sensory impairment, epilepsy, medication effects, interpersonal stress, care environment, and reinforcement contingencies. A fourth asks whether the data support a psychiatric syndrome, and if so what evidence distinguishes it from the alternatives. Writing these tracks separately prevents the developmental diagnosis from swallowing the entire formulation.

Behaviour that challenges has person-specific associations, including male sex, lower adaptive functioning, poorer communication, autism, and painful physical conditions. Unlike oppositional defiant or conduct disorder, it is not associated with family adversity. This distinction guards against importing a formulation from typically developing populations without testing whether it fits. Similarly, a psychiatric presentation may coexist with environmental contingencies; neither cancels the other.

Carefully document the source of every observation, the degree of confidence in the person's self-report, baseline evidence, collateral agreement or disagreement, and the physical and environmental exclusions considered. This makes diagnostic reasoning reviewable and protects against therapeutic drift toward medication for an unclearly defined target. Management of ID itself, including multidisciplinary support and educational planning, is addressed in the management section of this chapter. The full child psychiatric assessment battery, services and protection are covered in the Child psychiatry: assessment, treatment, services and protection notes.

Formulation should also preserve the person's perspective. A carer's account may be essential, but it should not replace direct engagement with the person. Adapt the pace, language, and mode of communication; allow time for response; and test important inferences gently rather than relying on a single answer. The purpose is not to force adult verbal psychopathology into a developmentally mismatched interview. It is to make the available evidence as fair, transparent, and clinically useful as possible. Where uncertainty remains, state it explicitly, define what further observation would discriminate between competing explanations, and avoid converting uncertainty into a confident label.

The practical standard is neither diagnostic minimalism nor diagnostic enthusiasm. It is intellectual humility with enough structure to notice a second disorder. When the pattern, chronology, collateral history, and context converge, people with ID deserve the same diagnostic seriousness afforded to anyone else.

15 · Management of ID (multidisciplinary, early intervention, special education)

Management is the organisation of opportunity. Intellectual disability is not managed by a single clinic, a prescription, or a school placement in isolation. The clinical task is to turn an assessment of abilities, barriers, health and family circumstances into a coherent plan that improves participation and preserves dignity. This makes management a longitudinal exercise: the clinician must help the person acquire useful skills, reduce avoidable barriers, respond to mental or physical illness when present, and keep the family able to continue caring.

Marks are usually earned by showing this architecture rather than by naming a succession of therapies. A good answer begins with coordinated goals, places early intervention and education within the person's everyday settings, explains how behaviour is understood before it is treated, and recognises that medical treatment has a limited, target-led role. Severity should be named only as a descriptor of support need; its classification belongs with the assessment and severity sections of this chapter.

15.1 The multidisciplinary plan

Multidisciplinary care is the appropriate treatment model because intellectual disability has biological, psychological, social and developmental consequences at the same time. It is therefore multimodal: psychoeducation, psychological work and medication, where indicated, are coordinated rather than offered as competing alternatives. The plan also has to restore wellbeing within the family system. In practice, the psychiatrist's distinctive contribution is diagnostic clarification, assessment of mental and physical contributors to change, risk formulation, judicious prescribing, and helping the team keep a shared hierarchy of goals.

The team should not be imagined as a referral list. Its work is to translate a common formulation into actions that can actually happen at home, in education, and in the community. For optimal management, coordination includes at least six disciplines, including psychiatry, psychology, occupational therapy, speech and language therapy, genetic counselling and nursing. This is not an exhaustive job description. It illustrates why a locally available team should be assembled around the person's needs rather than around an idealised service chart.

RULE **Rule:** Start with shared functional goals, then assign one accountable person for coordination; referrals without a joint plan merely move fragmentation from one service to another.

SERVICE FUNCTION	CLINICAL PURPOSE	WHAT COORDINATION PREVENTS
Prevention and prompt recognition	Identify needs before they become fixed barriers to development or participation.	Late referral after family strain or school failure has already escalated.
Strengthening family capacity	Make everyday care, communication and goal-setting possible.	Advice that is technically sound but impossible to sustain.
Learning and productive roles	Build participation and future roles around meaningful skills.	Care that remains custodial when the person can learn more.
Health and mental-health care	Address health needs and emotional or behavioural presentations without diagnostic overshadowing.	Attributing a new problem automatically to the disability.
Living support and self-care	Match support to real-world functioning and safety.	Discharge plans that fail because ordinary living demands were ignored.

The level of specialist involvement should follow complexity. Mainstream mental-health services may prescribe for mild or borderline intellectual impairment, while assessment and treatment of behavioural or emotional disorders in more marked or atypical cognitive impairment should be undertaken with specialist clinical input. This is a service-triage principle, not a claim that a mainstream clinician should work alone when communication barriers or atypical presentation make assessment uncertain.

Family work is clinical work. Families need an explanation that can be revisited, not a single overwhelming disclosure. Parents may require repeated discussion before the implications can be absorbed; the clinician should return to prognosis, available help and the family's active role as understanding changes. Family support must be individualised to the caregiving burden and its meaning for that particular household. Parents and primary caregivers should also be routinely screened for stress-related disorders. This is not a courtesy add-on: depleted carers have less capacity to implement a plan, and their distress deserves treatment in its own right.

15.2 Early intervention as a developmental service

Early intervention begins before a child has entered an educational system. The Indian guideline permits a starting point from the prenatal period, with recognition of high-risk pregnancies, appropriate health care and attention to psychosocial adversity. After birth, the plan is not simply stimulation exercises. It begins with accurate identification of developmental and associated needs, then creates opportunities for sensory-motor, communication and socioemotional development in the child's ordinary relationships and routines.

The central relational mechanism is healthy bonding and attachment between the child and mother. The practical implication is important: early stimulation can also be delivered by any

stable caregiver. Programmes should stabilise current developmental achievements while opening the next achievable task. Play-based work and culturally rooted early-child-care practices are often more usable than imported materials; when materials are inaccessible or culturally mismatched, parents can become overwhelmed and the programme loses its place in daily life.

Evidence supports a measured, not triumphalist, account. **Family-centred intervention** can be effective, but uncertainty remains about the active components and how help is best delivered. This is why the clinician should set observable functional goals and revise them with the family, rather than promise a uniform developmental result. One intensive compensatory-education experience illustrates both the possibility and the limits of inference: teaching began at 3 months; at a later preschool assessment the intervention group had a mean advantage of 27 IQ points over controls. The finding does not prove a durable individual IQ gain, and its design limitations matter. Its defensible teaching point is that sustained, skilled effort with socially disadvantaged families can make a meaningful difference.

3 years	3-6	6-18
INITIAL LEARNING PHASE	SCHOOL-READINESS PHASE	CONSOLIDATION PHASE

A life-span skill plan has three **developmental stages**. In the initial years, work addresses sensory-motor ability, socio-communication, basic self-help and concepts. The subsequent school-readiness period emphasises readiness skills and culturally appropriate adaptive behaviour. Later childhood and adolescence consolidate academic and independent personal skills in preparation for vocational training, employment and adult living. These are organising priorities, not rigid age-based ceilings. The immediate task is always to find the skill that will increase autonomy in the child's own context.

IN INDIA

For formal early intervention in India, referral can be made to the District Early Intervention Centers of the Rashtriya Bal Swasthya Karyakram and the Anganwadi Centers of the Integrated Child Development Services. The referral should carry a concise developmental formulation and the family's priorities, so that the service encounter becomes part of a continuing plan rather than a one-time certification exercise.

The clinical details of normal milestones and attachment are covered in the Normal child development and milestones notes.

15.3 Education, participation and placement

Normalization is the guiding principle of arranging a pattern of life as close to ordinary life as possible, supported by specialist help that allows the person to reach potential. It directs attention away from segregation as a default and toward family life, community participation, education, work and self-advocacy. It does not mean pretending that support needs do not exist. Normalisation becomes clinically useful only when the necessary teaching, communication support, supervision and environmental adjustments are actually provided.

Placement is a clinical and ethical judgement. Ordinary-school education may offer more normal social surroundings, social integration, expectation of progress and learning for peers that participation is ordinary. It may also lack the teaching skills, equipment and methods required by some learners, particularly where specific language and communication teaching is needed. The evidence cited here does not settle which children benefit from ordinary schooling. Therefore, do not turn inclusion into a slogan or special education into a failure. Consider the child's learning needs, social experience, available supports and family preference, then review the placement when the fit changes.

TRAP **Exam trap:** "Mainstream" and "beneficial" are not synonyms. Educational inclusion without the support needed to learn or remain safe is not meaningful participation.

The ethical tension is real. In secondary schools, children with special needs were reported to be bullied **three times more often** than other children. This should prompt active safeguarding and a discussion of the individual child's present interests, not a simplistic conclusion that community education is always unsafe. At a service level, a systematic review found better outcomes with community-based services than with institutional care. The practical inference is to design community support well, monitor the child's experience, and address exclusion rather than treating it as inevitable.

Education should be functional. Literacy, numeracy, communication, self-care and social learning have value insofar as they enlarge the person's choices in daily life. At the end of school, reassessment and vocational guidance are necessary before a young person is expected to make an adult transition. Opportunities should be ambitious but realistic, and should not be narrowed merely because others hold limited expectations. The fuller approach to disability certification, benefits and statutory entitlements belongs in the disability certification and benefits section of this chapter.

15.4 Behavioural formulation before behavioural control

Behaviour that challenges is a question, not a diagnosis. Its onset or worsening should lead first to a search for communication difficulty, environmental demand, reinforcement, health problems and mental disorder. A **comprehensive personalised functional assessment** considers behaviour in its setting and improves the chance of a plan that can be managed psychologically and environmentally. It can consequently reduce, and sometimes remove, the need for psychotropic medication.

Functional behaviour analysis asks what consequence may be maintaining the behaviour. Challenging behaviour may serve three functional classes: obtaining a social positive reinforcer such as attention, escaping a demand through social negative reinforcement, or obtaining sensory reinforcement. These possibilities are hypotheses to test against observation, not labels to attach from a brief history. Parents, teachers and other caregivers are indispensable informants because they see the antecedents and consequences that a clinic visit cannot reproduce.

The behavioural plan should be least intrusive and culturally appropriate. The Indian guideline describes **three** linked levels: restructure the environment to alter antecedents, reinforce adaptive behaviour differentially, and prevent inappropriate reinforcement of problematic behaviour. Its basic premise is functional substitution. If a problematic behaviour communicates a need, the plan must create and reinforce a socially acceptable behaviour that serves the same function. Suppression alone leaves the original need unanswered and is likely to fail once the therapist has left.

- Define a small number of observable target behaviours in the settings where they occur, and prioritise those causing exclusion, stigma or interference with learning.
- Record antecedents, behaviour and consequences with carers, then decide which maintaining contingencies are plausible.
- Teach and reinforce an adaptive alternative that achieves the relevant function more safely.
- Change environmental triggers and caregiver responses together, then review whether the person's participation has improved.

Positive behavioural support is a useful framework because it combines reinforcement with changes to triggers, minimal use of aversive strategies and planned reactive responses such as distraction. Its components should not be applied as a menu of techniques. Selection follows the formulation. Techniques using negative reinforcement should not be used. A discipline-heavy behaviour programme is best developed and implemented by a behavioural psychologist or board-certified behavioral analyst, although psychosocial interventions delivered by non-specialist carers can produce better outcomes than no treatment or treatment as usual. In settings with few specialists, this supports task-sharing with supervision and training, not abandonment of formulation.

15.5 Medication stewardship and specialist liaison

GOOD TO REMEMBER

Medication treats a defined target symptom, coexisting mental disorder or physical contributor; it does not replace a formulation of behaviour. **Psychological intervention**, including functional behavioural analysis, should be considered first-line when no identifiable mental illness with clear treatment implications is present, except in the most serious or intractable behavioural disturbance. The same discipline applies when diagnostic information is inadequate: try psychosocial intervention before converting uncertainty into a prescription.

Antipsychotics remain the most widely prescribed psychotropic class in intellectual disability, yet their current positioning is as a later option for older children or adults when intensive behavioural intervention or other medication options have failed. This apparent contradiction is a warning about service reality, not a reason to disregard behavioural care. A functional formulation may reveal a modifiable trigger that makes medication unnecessary, whereas a clear psychiatric syndrome or severe risk may require simultaneous medical treatment and behavioural support.

When a dopamine antagonist is considered for behaviour that challenges, the NICE guideline NG11 identifies **three** alternative qualifying circumstances: psychological or other intervention alone has not changed the behaviour within an agreed time, treatment of a coexisting mental or physical problem has not reduced it, or risk is very severe. These are alternatives, not a checklist in which every condition must be met. Once medication is begun, prescribe the lowest possible dose and reassess whether continuation remains justified after target symptoms improve, when there is non-response, or when adverse effects affect functioning or quality of life.

> The management sequence is: clarify function and illness, strengthen environmental and behavioural supports, prescribe only for a defined target, and keep asking whether the drug is still needed.

Deprescribing needs the same care as initiation. The NICE guideline NG54 suggests slow withdrawal of antipsychotics for people without psychotic symptoms. This does not mean that withdrawal is consequence-free: discontinuation studies commonly, though not invariably, show re-emergence of problem behaviours. The appropriate response is preparation, monitoring of the original target behaviour, and readiness to revise environmental support, rather than assuming either that every drug must continue or that every drug can be stopped without difficulty. The UK STOMP programme promotes antipsychotic deprescribing but also shows a crucial pitfall: an antipsychotic may be replaced by another psychotropic rather than reducing overall medication burden.

Drug choice for a diagnosed condition may in general resemble practice in the wider population, but good prescribing practice is to start at lower doses and increase more slowly. This distinction resolves an apparent contradiction between using standard treatments and being more cautious in a person who may have communication barriers, neurological vulnerability or difficulty reporting adverse effects. A review should document the target symptom, benefit, adverse effects, family observation and whether non-drug supports remain active. Annual health checks are generally recommended for people with intellectual disability; they are health surveillance, not a substitute for medication review whenever treatment changes.

15.6 Continuity into adult life

Management succeeds when it survives transitions. A community-care model is judged less by its label than by the detail of planning and the commitment with which it is carried out. Planning needs both an understanding of the local population and an individual account of what this person can do, wants to do, and needs help to do. A named coordinator can prevent families from receiving inconsistent advice across health, education, social and voluntary-sector services.

Families often need additional help at puberty and school leaving; moving from child to adult services can be especially stressful. These periods should trigger anticipatory planning rather than a crisis referral. Reassessment and vocational guidance before school leaving protect against the common drift from education into inactivity. For school leavers with mild intellectual disability, vocational planning may lead to ordinary or sheltered work; adults with severe

disability are more likely to need the support of a day centre. The point is not to predict a person's future from a label, but to plan enough support for a real adult role.

Referral and linkage in India should map the person's needs to education and vocational routes, rehabilitation centres, national institutes and local nongovernment agencies. Whenever possible, link the family to agencies in their locality so that rehabilitation costs and travel do not quietly defeat the plan. The clinician should retain a longitudinal role: explain the plan repeatedly, detect emerging health or psychiatric problems, coordinate with the relevant team, and protect the person's right to participate in family and community life.

16 · Prevention of intellectual disability

Prevention is a life-course clinical task. Prevention of intellectual disability is not a promise that every case can be avoided. It is the organised reduction of preventable risk before conception, during pregnancy and birth, through the newborn period, and after disability is recognised. The examination value of the topic lies in distinguishing prevention of a condition from prevention of avoidable functional disadvantage once a person already has intellectual disability. A good answer therefore shows a framework, gives a few high-yield examples at the correct level, and retains ethical humility.

The field uses several overlapping taxonomies. The most useful approach in an intellectual-disability answer is to state the purpose of the intervention rather than force every programme into a label. Public health measures reduce exposure to risk; screening creates an opportunity for timely action; and habilitative care limits the secondary disabilities that arise when a person's needs are neglected. These are connected steps in one pathway, not competing lists.

16.1 What prevention means in intellectual disability

The conventional framework has **three** categories. **Primary prevention** acts before a disorder develops, so its clinical purpose is to reduce new cases. For intellectual disability, this means combining medical care with public-health, educational and social measures before an avoidable insult has produced disability. The point is causal timing: an intervention is primary when it removes or reduces the exposure before the outcome arises.

Secondary prevention aims to reduce the burden of established disease in the general terminology, but the intellectual-disability application is more clinically practical: recognise a condition that can lead to disability as early as possible and treat it before disability follows. This makes screening meaningful only when a result can lead to confirmation, treatment, counselling or follow-up. A test with no route to action may collect information, but it does not by itself achieve prevention.

Tertiary prevention concerns disability associated with an existing illness. In intellectual disability it is best understood as **habilitation**: helping the person attain the best attainable functioning despite the disability. This includes biomedical and sociocultural work, but it is not a synonym for cure. The target is participation, competence and protection from avoidable loss of function.

RULE **Rule:** Classify a preventive intervention by its intended point of action: before disability arises, while a remediable pathway is still open, or after disability is present to prevent additional functional loss.

PREVENTIVE LEVEL	CLINICAL QUESTION	INTELLECTUAL-DISABILITY APPLICATION	COMMON ERROR
Primary prevention	Can the causal exposure or pathway be avoided?	Genetic counselling, population health, safer pregnancy, nutrition, immunisation and prevention of toxic or traumatic injury.	Calling every antenatal test prevention without asking what action follows, or filing genetic counselling under care that comes after a diagnosis.
Secondary prevention	Can a disorder that may cause disability be found and treated early?	Early recognition and treatment of metabolic conditions or congenital hypothyroidism.	Equating screening with treatment.
Tertiary prevention	How can further functional disadvantage be reduced after disability is present?	Habilitation, education, family support and treatment of comorbid conditions.	Presenting support as an afterthought rather than prevention of secondary disability.

The alternative language of universal, selective and indicated prevention is also useful in public health. Universal work reaches whole populations, selective work addresses groups with increased risk, and indicated work is directed at people with individual evidence of heightened risk. Do not casually equate every use of indicated across classifications. The later Institute of Medicine model, published in 1994, uses indicated intervention for people with early or prodromal symptoms, whereas the earlier approach used it for asymptomatic people identified as higher risk. The boundary between selective and indicated work can be clinically blurred; the safer answer is to state the target population and the reason for intervention.

TRAP **Exam trap:** Primary, secondary and tertiary prevention are not interchangeable labels for antenatal, postnatal and rehabilitation services. Timing matters, but the decisive issue is what the intervention is meant to prevent.

16.2 Primary prevention before and during pregnancy

Primary prevention begins before pregnancy is recognised. Good preconception and antenatal care allows the clinician to identify modifiable risks, improve maternal health and support informed reproductive decisions without converting counselling into coercion. The Indian guideline specifically identifies malnutrition, diabetes, teratogenic drugs and substance use as maternal factors that may be treatable or preventable in subsequent pregnancies. A prior child with intellectual disability should prompt a respectful etiological review and a plan for the next pregnancy, not an assumption of parental fault. That review is where **genetic counselling** belongs, and it belongs here rather than later: the standard account of primary prevention opens with genetic counselling and continues with education of pregnant women about behaviours that may put the fetus at risk, early detection of fetal abnormalities during pregnancy, and good

obstetric and perinatal care. Counselling a couple before a further conception acts on the next pregnancy, so it removes the exposure before the outcome arises, which is the test this section has just set. Reading it as an after-the-fact service, because the conversation happens after a diagnosis in an older child, is the classic misplacement, and naming the primary preventive measures is a canonical examination question whose first answer is this one.

Risk reduction works through ordinary care. Primary measures include prevention of lead poisoning, avoidance of substance exposure during pregnancy, prevention of teenage pregnancy, improved neonatal care and injury prevention such as infant car seats. Prenatal vitamin supplementation is included because it prevents neural tube defects. The phrasing matters: prenatal supplementation should not be inflated into a guarantee against all developmental disorders. Likewise, childhood lead exposure is a risk factor for low IQ and intellectual disability, which supports environmental prevention without claiming that every exposed child will develop disability.

Maternal infections are another preventable pathway. Toxoplasmosis, cytomegalovirus, rubella and herpes are recognised maternal infections associated with infant morbidities that include intellectual disability. Rubella immunisation is therefore an important prevention measure. Prenatal care may begin before conception, including rubella immunisation for women without immunity. Prevention of rhesus sensitisation uses **anti-D antibody**. These examples should be used to show an integrated maternal-health strategy, not as a memorised infection list detached from antenatal care.

Alcohol deserves direct enquiry in all settings where it is accessible. In such societies, alcohol is the leading toxin cause of intellectual disability. Fetal alcohol syndrome has an incidence of **0.5-3 per 1000 births** and may account for as much as 3 percent of all intellectual disability. These are different denominators, and they must not be swapped. Apparent rates also depend on diagnostic practice: use of both physical signs and a history of heavy prenatal alcohol exposure produces lower rates than use of exposure history alone, with underdiagnosis and overdiagnosis possible at the two extremes.

Nutritional prevention has its own logic. Iodine deficiency is described as the most common preventable cause of learning disability worldwide. Where endemic iodine deficiency threatens pregnancy, periconceptional injection of **iodized oil** has attenuated endemic cretinism and subsequent intellectual disability in at-risk populations. A pharmacology source also states that daily maternal iodine intake of **150-200 microgram** can prevent endemic goitre and cretinism due to iodine deficiency, with iodised edible salt as the preferred delivery method. This is a prevention claim, not an invitation to prescribe an unsupervised supplement outside local obstetric and public-health practice.

one-third	over eighty	over 50
ID/GDD CASES WITH NONGENETIC CAUSES	POTENTIALLY TREATABLE DISORDERS IDENTIFIED	CONDITIONS DETECTED BY CONTEMPORARY NEWBORN SCREENING

The limits are as important as the opportunities. Nongenetic factors account for approximately one-third of ID/GDD cases, so environmental prevention is necessary but cannot remove all intellectual disability. At the same time, advances in therapeutics have identified over eighty potentially treatable disorders. This justifies careful diagnosis and prompt action without selling prevention as total control. Prevention priorities differ by setting: maternal and perinatal health are central public-health opportunities in low- and middle-income countries, while reduction of genetic causes of severe intellectual disability remains important in high-income countries. It is unlikely, however, that prevention can substantially alter the incidence of mild intellectual disability.

16.3 Antenatal screening, diagnosis and ethical care

Non-invasive prenatal screening is based on cell-free fetal DNA in maternal plasma. Its value is risk estimation and selection for counselling or diagnostic evaluation, not a diagnosis by itself. First-trimester screening combines an ultrasound marker with maternal blood testing. An ultrasound assessment for major malformations is often performed at approximately 20 weeks of gestation. These routes should be explained as complementary rather than as competing tests.

CLINICAL ROUTE	WHAT IT CAN CONTRIBUTE	TEACHING POINT
First-trimester screening	Risk assessment through ultrasound and maternal blood analytes.	A positive screen requires counselling and a diagnostic pathway.
Mid-trimester AFP testing	Screening for neural tube defects in the specified screening context.	An analyte result is not a diagnosis and is not specific to one disorder.
Amniocentesis	Diagnosis of chromosomal and metabolic disorders.	Use the test for the question it can answer.
Chorionic villus sampling	Chromosomal and molecular genetic studies.	Do not transfer amniocentesis metabolic role to CVS.

In one psychiatric-text account, amniocentesis or chorionic villus sampling is recommended for all women aged **35 years or more**. Another account frames amniocentesis as an indication-based decision for pregnancies with relevant prior or familial history, or a clinically significant current risk. The defensible clinical response is to know that recommendations are guideline- and context-dependent, establish the indication, explain benefits and limitations, and refer for current specialist obstetric or genetics advice rather than use a textbook threshold as a stand-alone rule.

Screening needs careful language. Maternal serum alpha-fetoprotein is a marker with more than one possible clinical explanation; it is not a disease label. For in-utero diagnosis of Down syndrome, the source describes a blood-based route, ultrasound assessment and invasive testing. These options differ in what they establish and what follow-up they entail. Carrier screening should be offered to all persons in high-risk populations, but named examples should follow locally relevant, verified services rather than stereotype or assumption.

Prenatal diagnosis followed by elective termination raises a boundary that clinicians should not obscure. It is a matter of deeply personal values because termination prevents childbirth rather than treating or preventing the disorder in the fetus. The psychiatrist's role is to support informed, non-directive decision-making, recognise family distress, avoid coercive framing, and ensure that a decision is not driven by misinformation about disability or by a lack of support for families who continue a pregnancy. The syndrome-specific genetics and recurrence discussion belongs with the relevant aetiology sections of this chapter.

16.4 Newborn screening and time-sensitive treatment

Newborn screening refers to tests undertaken within the first hours or days of life when early action can prevent severe health problems. Contemporary systems detect over 50 different conditions. A blood spot collected by heel stick is the established specimen method in the settings described in the source.

> **GOOD TO REMEMBER**
> An abnormal screen is a signal for confirmation, not a diagnosis or a moral judgement about the family. Communicate promptly and compassionately, then ensure confirmatory testing, treatment and longitudinal follow-up.

Congenital hypothyroidism illustrates the time-sensitive logic. With newborn screening, levothyroxine may be initiated by **4-6 weeks of life**. Screening of neonates is described as the best preventive strategy for cretinism. This should not be reduced to a prescription detail: early identification links a population programme to a child developmental outcome. The quoted treatment timing is not the time at which the blood sample must be collected.

The overall verdict on newborn metabolic screening is appropriately strong but bounded: it has been extremely successful in reducing the incidence and severity of some intellectual-disability syndromes. The qualifier some is crucial. A screening programme may also generate false-positive results, which can produce acute parental distress and unnecessary follow-up unless communication is prompt and compassionate.

Phenylketonuria is a classic screenable metabolic disorder, but its genetic mechanism and diet belong in the inborn errors of metabolism section. Here, the prevention lesson is narrower: early diagnosis and treatment of PKU, other metabolic conditions and congenital hypothyroidism are examples of secondary prevention because they intervene before disability develops.

16.5 Preventing secondary disability after diagnosis

Once intellectual disability is present, the preventable outcome is often loss of function rather than intellectual disability itself. Tertiary prevention therefore uses early support, education, family support and timely attention to mental or physical conditions that suppress functioning. This is habilitation in action. It safeguards developmental opportunity and reduces the additional disability created by untreated epilepsy, pain, sensory difficulty, psychiatric illness, behaviour problems, exclusion or caregiver exhaustion.

Do not wait for a crisis to begin support. Early intervention may start in the prenatal period by identifying high-risk pregnancies, arranging appropriate health care and addressing psychosocial adversities.

IN INDIA

For formal early intervention, the guideline names the District Early Intervention Centres of the Rashtriya Bal Swasthya Karyakram and the Anganwadi Centres of the Integrated Child Development Services. The preventive point is timely connection before a modifiable disadvantage becomes entrenched.

The service model, developmental methods and general evidence for early intervention are covered in the management section of this chapter.

Comorbidity management is central, not peripheral. Epilepsy affects 15-30 percent of persons with intellectual disability. Problematic behaviours may occur in 20-80 percent of the ID population. These figures do not authorise diagnostic overshadowing. A new behavioural change may be communication of pain, distress, environmental difficulty or mental illness. Prevention in tertiary care means looking actively for such contributors and treating them before they further reduce learning, relationships or family capacity. The psychiatric-comorbidity assessment itself is addressed in the dual-diagnosis section of this chapter.

Families may need genetic counselling alongside causal investigation and reproductive planning. It should be offered without blame and without making a risk discussion substitute for the family's own values or choices. The generic genetic-counselling process and diagnostic work-up are developed in the aetiology section.

16.6 Public-health priorities and realistic expectations

Prevention is most effective when it reaches the conditions in which children develop. ID risk factors are considerably increased in low-income countries, contributing to their increased rate of intellectual disability. This is why a prevention answer should move beyond rare disorders and include maternal health, nutrition, safety, infection control, toxic exposure and access to perinatal and neonatal care. It is also why prevention must be paired with equity: a family cannot implement advice that depends on services, food security or safe housing it cannot access.

Evidence supports interventions at more than one level. Programmes aimed at psychosocial stimulation were more effective than nutritional supplementation for improving both cognition and behaviour in growth-retarded children in the cited comparison. This does not diminish nutrition. It teaches that development depends on both biological provision and responsive social experience, and that a single nutrient-focused programme may not repair the effects of deprivation. Similarly, prevention programmes should be evaluated for whether they can be delivered consistently and whether they improve real developmental participation.

A comprehensive service begins with prevention and early detection and then connects families to the educational, health, social and practical supports that sustain participation. Prevention is therefore the first element of a continuum of care, not a separate campaign that ends after a

screening test. Appropriate referrals should follow the person across early, educational and adult life. The statutory eligibility and benefits themselves are dealt with in the disability certification and benefits section of this chapter.

MNEMONIC

PREVENT

Prepare before conception; **R**educe maternal, toxic and nutritional risks; **E**valuate antenatal and newborn screens; **V**erify a positive result; **E**ngage family support; **N**otice comorbidity; **T**arget function through habilitation.

The mature answer is neither fatalistic nor triumphalist. It recognises preventable pathways, acts early when an actionable condition is found, and continues to prevent additional disability through support and treatment. It also respects the person with intellectual disability as more than the outcome of a risk factor. Prevention should enlarge future choice, not become a language for blaming parents, narrowing reproductive autonomy or measuring a life only by what might have been avoided.

- Ask which preventable exposure or remediable condition is relevant at the present point in the person's life course.
- Make sure a positive screen has a confirmation, treatment and follow-up pathway.
- After diagnosis, look for treatable illness and avoidable barriers to participation rather than assuming that decline is inevitable.

For intellectual disability, prevention is a continuum: reduce avoidable risks before disability arises, detect and treat remediable conditions early, and use habilitation to prevent avoidable loss of function after diagnosis.

17 · Disability certification and benefits for intellectual disability in India (IDEAS, National Trust Act)

Certification is a clinical document with legal consequences. For a person with intellectual disability, it can convert an assessment into access to education, work, social protection and supported decision-making. The psychiatrist's task is therefore not merely to write a diagnosis. It is to distinguish clinical formulation from statutory quantification, obtain defensible evidence of functioning, explain what the certificate does and does not establish, and help the family navigate a system that can otherwise feel opaque.

The contemporary legal frame is the **Rights of Persons with Disabilities Act, 2016**, notified on **28 December 2016**. It replaced the **Persons with Disabilities (Equal Opportunities, Protection of Rights and Full Participation) Act, 1995**. Intellectual disability is a specified disability under this framework, alongside other conditions relevant to psychiatric practice. This section concerns the certification route for intellectual disability, its practical consequences, the limited role of IDEAS, and the National Trust framework for adulthood and guardianship.

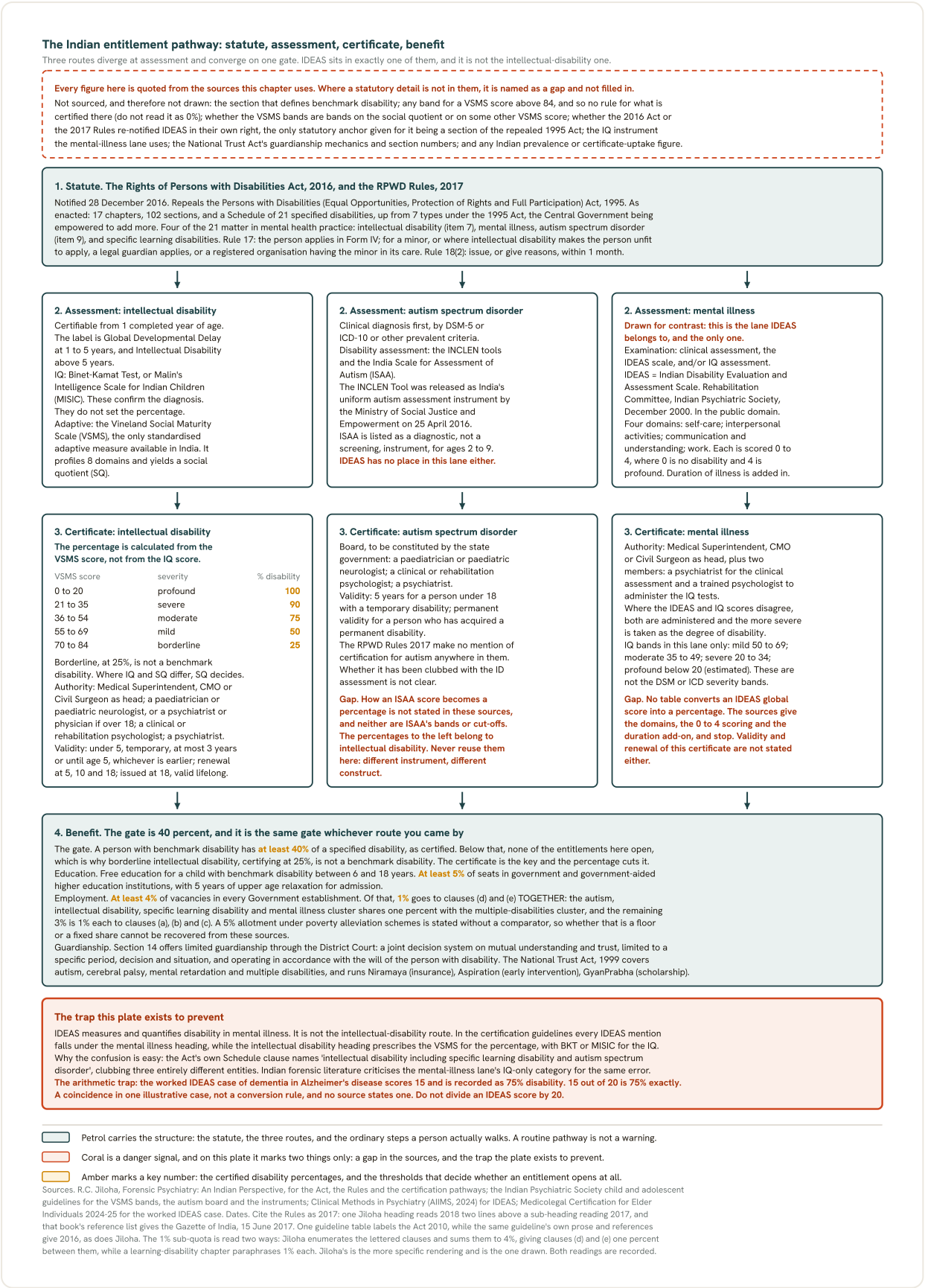


Fig 21.10 The Indian entitlement pathway: statute, assessment, certificate, benefit. One statute, three routes that diverge only at assessment, and one gate at 40 percent that all three converge on. The distinction the chapter turns on: IDEAS is the mental-illness disability instrument, not the intellectual-disability route, whose certified percentage comes from the VSMS score.

17.1 The legal frame: from diagnosis to enforceable participation

The Act is not a diagnostic manual. Its purpose is to translate disability into rights, duties and institutional remedies. It has 17 chapters and 102 sections; its Schedule names 21 specified disabilities, compared with 7 disability types in the earlier law. Within the Schedule, intellectual disability appears at serial 7 and autism spectrum disorder at serial 9. The nomenclature matters: the Act replaces **"mental retardation" with "intellectual disability"**. In clinical records and communication, use the current term consistently.

The statutory definition recognises limitation in intellectual functioning together with adaptive behaviour in everyday social and practical skills. That overlap with clinical assessment is important, but the questions asked are different. A clinical assessment asks what the person can do, what has changed, which supports work and whether there are treatable coexisting problems. A certificate establishes disability in the form required for rights and entitlements. It should never flatten the person into a percentage or be allowed to substitute for an individualised care plan.

RULE **Clinical severity and certified disability are related, but they are not interchangeable labels.** Clinical formulation guides care; statutory quantification establishes eligibility under a legal scheme.

For intellectual disability, the statutory schedule describes significant limitation in intellectual functioning together with limitation in adaptive behaviour. It does not legislate an IQ number, and a candidate should resist the temptation to supply one: the percentage on the certificate is derived from adaptive functioning, not from the IQ score. This is an easily missed distinction. A numerical cognitive test can corroborate intellectual impairment, but it does not itself produce the statutory percentage. Candidates who equate the two have confused an assessment construct with a legal outcome, and quoting a legal IQ cut-off that the statute never states is the sharpest form of that error.

- Use the diagnosis to organise clinical understanding and intervention.
- Use the certificate to document statutory disability and enable access to rights.
- Explain to caregivers that the certificate records function for a stated purpose, not the person's potential or worth.

17.2 The intellectual-disability certification assessment

Start with the correct question. The assessment must establish intellectual disability and then quantify disability in the prescribed manner. Careful administration and a credible developmental history are especially important; a rushed score, divorced from the person's actual opportunities and supports, is a poor foundation for a legal document.

IN INDIA

The Indian guideline identifies the VSMS as the currently available standardised adaptive-behaviour measure. The prescribed adaptive-functioning instrument is the **Vineland Social Maturity Scale (VSMS)**; it yields a **social quotient (SQ)** and an adaptive profile. Intellectual functioning is assessed using the **Binet-Kamat Test** or **Malin's Intelligence Scale for Indian Children**.

Cognitive testing in this pathway provides diagnostic confirmation, not percentage derivation. Where IQ and SQ suggest different grades, the SQ should guide the decision, because social maturity more closely reflects the ability to meet culture-appropriate demands of daily life.

GOOD TO REMEMBER

An implausible or discordant result should prompt review of the history, test conditions, language, sensory or motor barriers, educational opportunity, behaviour during testing and the informant's understanding of the items.

A statutory assessment should be clinically thorough even when its output is concise. Establish the developmental course and current adaptive abilities from an informant who knows the person across settings. Consider whether communication difficulty, behavioural disturbance, sensory impairment, physical illness or an untreated psychiatric disorder has distorted apparent function. Record observed abilities as well as deficits. The certificate process should protect against both overestimation, which deprives a person of support, and underestimation, which may unnecessarily restrict autonomy.

TRAP **Do not use an IQ score to calculate the intellectual-disability percentage.** In this certification pathway, the percentage follows the VSMS-based adaptive-functioning assessment; IQ testing confirms the intellectual impairment.

The categories below are statutory bands, not a substitute for the clinical severity discussion elsewhere in this chapter. They answer the specific legal question of what disability percentage is certified. The row should be read as a mapping: identify the applicable VSMS band, then document the corresponding percentage without improvising a conversion.

CERTIFICATION CATEGORY	VSMS SCORE BAND	CERTIFIED DISABILITY	PRACTICAL INTERPRETATION
Profound	0-20	100%	Highest statutory disability band
Severe	21-35	90%	Benchmark-disability threshold is met
Moderate	36-54	75%	Benchmark-disability threshold is met
Mild	55-69	50%	Benchmark-disability threshold is met
Borderline disability	70-84	25%	Does not meet benchmark-disability status

The borderline-disability row is clinically useful because it makes the eligibility boundary explicit. It is not a reason to dismiss need. A person may still need educational, vocational, family or mental-health support even when the statutory benchmark threshold is not met. Conversely, a benchmark certificate creates a legal entitlement pathway but does not tell the clinician which supports will be effective. Keep these two conversations separate and honest.

17.3 Application, medical authority and validity

The route should be explained before the family is sent from counter to counter. An application for a disability certificate is made in **Form IV**. When a person with intellectual disability cannot apply, a legal guardian may apply; an organisation registered under the Act may also apply where it has a minor in its care. This provision is practical rather than merely administrative: it avoids making access to certification contingent on an ability that is itself affected by the disability.

The medical authority is headed by the Medical Superintendent, Chief Medical Officer, Civil Surgeon or an equivalent state-notified authority. Its clinical membership includes a paediatrician or paediatric neurologist where available, with age-appropriate alternatives specified for adults, a clinical or rehabilitation psychologist, and a psychiatrist. The psychiatrist's contribution is more than signing a completed form. It includes diagnostic coherence, review of developmental and behavioural information, recognition of coexisting psychiatric illness, communication with the family and protection of the person's dignity during examination.

Certification can be issued after at least **1 completed year of age**. The label is age-sensitive: the guidance uses Global Developmental Delay from ages 1 to 5 years and Intellectual Disability after age 5 years. This distinction avoids premature certainty while preserving access to assessment and support. Do not delay a needed certificate simply because the child's development is still being clarified; use the appropriate certification label and plan review.

WHEN THE CERTIFICATE IS ISSUED	VALIDITY APPROACH	CLINICAL IMPLICATION
Before age 5 years	Temporary; it ends at the earlier of age 5 or 3 years.	Plan reassessment rather than treating the early certificate as final.
Between ages 5 and 17 years	Review is scheduled at ages 5, 10 and 18 years.	Make renewal part of transition planning, not a crisis near a deadline.
At age 18 years	Lifelong validity	Use the adult review to address future support and decision-making.

The medical authority must issue the certificate or communicate the reason for rejection within at most **1 month from receipt of the application**. In practice, clinicians can reduce avoidable delay by ensuring that reports identify the tool used, the functional basis of the percentage, the diagnosis appropriate to age and any need for follow-up. Keep a copy of the assessment record: it is clinically useful at renewal and can clarify an apparent discrepancy without making the family repeat an entire evaluation.

17.4 Benchmark disability and the rights that certification can activate

A **person with benchmark disability** has certified disability of at least **40%**. This is an eligibility threshold, not a clinical threshold. It determines access to several statutory provisions, whereas care should respond to need at every level of disability. Explain this plainly. Families often hear “eligible” as “severely disabled” or “ineligible” as “no help is possible”; both inferences are wrong.

40%	5%	4%	1%
BENCHMARK THRESHOLD	HIGHER-EDUCATION SEATS	GOVERNMENT VACANCIES	SHARED JOB-RESERVATION CLUSTER

A child with benchmark disability has a right to free education between 6 and 18 years. Government and government-aided higher-education institutions reserve at least 5% of seats for persons with benchmark disabilities, with an upper-age relaxation of 5 years for admission. In every Government establishment, at least 4% of vacancies are reserved. These provisions should not be presented as a promise of a particular educational placement or job. The clinician should document function accurately and direct the family toward the relevant institutional process rather than make an outcome guarantee.

The employment reservation has a subtle but examinable structure. Autism, intellectual disability, specific learning disability and mental illness form one listed cluster. That cluster does not receive a separate one-percent allocation by itself: it shares exactly 1% of vacancies with the multiple-disabilities cluster. This clarification prevents the common answer that incorrectly assigns the whole sub-quota to intellectual disability or to the listed cluster alone.

- Discuss educational entitlement early enough for the family to use it meaningfully.

- Frame reservation as an access mechanism, alongside realistic skill-building and accommodation.
- Check the current administrative pathway locally before advising on an application or placement.

17.5 IDEAS: important scale, wrong shortcut for ID certification

The acronym is a frequent source of error. IDEAS means Indian Disability Evaluation and Assessment Scale. It was developed by the Rehabilitation Committee of the Indian Psychiatric Society in December 2000. IDEAS is designed for disability due to mental disorders and is recommended for disability associated with mental illness. It is publicly available and remains important for psychiatric disability work.

IDEAS rates four functional domains: self-care, interpersonal activities, communication and understanding, and work. Each domain is scored from 0 to 4, from no disability to profound disability, and duration of illness is incorporated into the global score. These features explain why IDEAS may look superficially attractive in an adult with longstanding functional impairment. They do not make it the intellectual-disability certification instrument.

TRAP **IDEAS must not be substituted for the ID certification route.** Its place is the mental-illness pathway, whereas intellectual-disability certification follows its own prescribed assessment route.

This distinction is more than a technicality. Intellectual disability is a neurodevelopmental condition, and its certification requires an account of adaptive functioning. IDEAS includes illness duration, which answers a different question. Importing it into an ID certificate can create a legally vulnerable assessment and obscure the actual statutory basis of the percentage. Where intellectual disability and mental illness coexist, formulate and treat both conditions, but do not let the presence of psychiatric symptoms alter the prescribed ID percentage route.

17.6 Supported decision-making and the National Trust framework

Certification often brings families to the question they had postponed: who will help the person make decisions in adult life? Begin with the person's own preferences, abilities and participation. The **limited guardianship** model under the Act is offered by the District Court. It is a joint decision-making arrangement based on mutual understanding and trust, confined to a specified period and decision or situation, and intended to operate according to the person's will. This is conceptually different from taking over a person's life because support is convenient for others.

Clinicians should avoid treating guardianship as an automatic consequence of diagnosis or a certificate. Assess the decision at hand, the person's capacity to participate with support, the risks of coercion and the least restrictive way to enable a meaningful choice. Documentation should describe function and support needs rather than make sweeping declarations of incapacity. This approach also keeps the consultation aligned with rights, not merely family anxiety.

The **National Trust for the Welfare of Persons with Autism, Cerebral Palsy, Mental Retardation and Multiple Disabilities Act, 1999** covers four conditions: autism, cerebral

palsy, mental retardation and multiple disabilities. It creates a national body intended to support independent, community-close living and meaningful participation, rather than segregation or passive care. With an adult who has intellectual disability, the clinician must provide information about guardianship and National Trust provisions.

MNEMONIC

SUPPORT before substitution

Start with the person's wishes, identify the decision and the support required, protect against undue influence, and use the least restrictive lawful arrangement.

- Raise future decision-making before an emergency forces the issue.
- Include the adult with intellectual disability in the discussion in a communication-accessible manner.
- Separate financial or property concerns from a global assumption that the person cannot decide anything.

17.7 Documentation, advocacy and protection from procedural harm

Good certification is disciplined advocacy. The final document should make its reasoning auditable: diagnostic conclusion, adaptive assessment, IQ assessment where used, the statutory basis of the percentage, validity and the need for review. Avoid pejorative wording, casual estimates and unexplained contradictions between narrative and score. Discuss the result with the person and family in plain language. A certificate can open doors, but careless language can also follow a person into school, employment and adult services.

Rights under the Act have enforcement mechanisms. A first contravention of the Act or its rules attracts a **fine which may extend to Rs 10,000**, and a subsequent contravention a **fine of not less than Rs 50,000 and up to Rs 5 lakh**. Neither tier carries imprisonment, and the first-tier figure is a ceiling rather than a flat penalty. Fraudulently availing a benefit meant for persons with benchmark disabilities is a separate and graver offence, punishable with imprisonment which may extend to 2 years, or fine which may extend to Rs 1 lakh, or both. A separate group of offences of atrocities carries imprisonment of not less than 6 months and up to 5 years, and with fine, and their elements are examinable precisely because dropping them broadens the offence. Insult or intimidation qualifies when it is intentional, done with intent to humiliate, and in a place within public view. The sexual-exploitation limb requires the offender to be in a position to dominate the will of a woman or child with disability and to use that position to exploit her. The section runs to several further limbs, each with its own elements, and they are worth reading in the Act itself rather than in summary: a paraphrase that drops an element is precisely how these offences get overstated. Special Courts are designated in each district to handle violations.

The psychiatrist is not expected to become the family's lawyer. The clinical role is to recognise when rights may be at stake, document the relevant functional facts clearly, avoid false reassurance and direct the family toward appropriate legal or administrative assistance. For

broader assessment, service planning and protection issues, consult the Child psychiatry: assessment, treatment, services and protection notes. The practical aim is simple: certification should increase access, participation and safety without becoming a mechanism for unnecessary exclusion.

Must remember: In intellectual-disability certification, diagnose clinically, confirm intellectual functioning with the Binet-Kamat Test or Malin's Intelligence Scale for Indian Children, and calculate the statutory percentage from VSMS-based adaptive functioning - not from IDEAS, and not from the IQ score, which does not enter the percentage at all.

Autism: concept, causes and criteria

18 · Historical evolution (Kanner, Asperger, DSM-IV subtypes to spectrum)

Why this history matters. Autism did not move from obscurity to its current spectrum formulation simply by acquiring a new name. Each classificatory change reflected a different answer to a clinical question: is the presentation a distinct childhood syndrome, a developmental disorder with several recognisable patterns, or a dimensional condition whose boundaries cannot be reliably divided into neat subtypes? For the clinician, this history explains why older notes use unfamiliar terms, why families may still identify with a former label, and why current assessment must not be constrained by obsolete diagnostic partitions.

The examinable spine is therefore not a recital of dates. It is the movement from carefully described clinical prototypes, through a broad pervasive-developmental-disorder framework, to a single spectrum diagnosis. That movement also records an important correction in psychiatric thinking: explanatory models have to be accountable to observation and evidence, particularly when they risk locating blame in parents.

1943	11	1944	2013
FIRST PAPER	CHILDREN DESCRIBED	PARALLEL ACCOUNT	SPECTRUM SHIFT

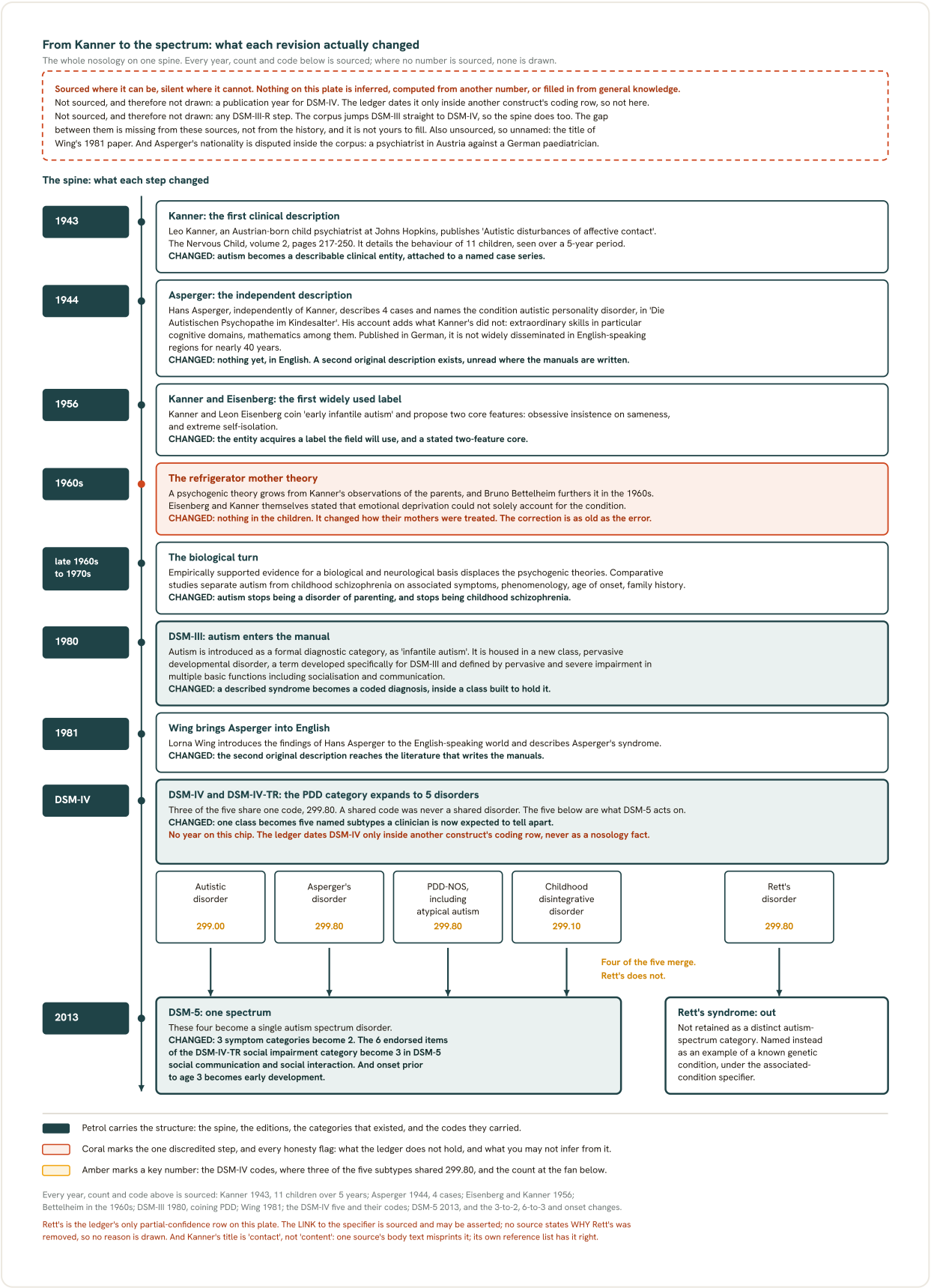


Fig 21.11 From Kanner to the spectrum: the nosological timeline and what each revision changed. The fan at the foot answers the examined question: four of the five DSM-IV subtypes merged into autism spectrum disorder and Rett's did not, surviving instead as an example of a known genetic condition under the associated-condition specifier. No year is drawn for DSM-IV and no DSM-III-R step is shown, because the sources cited hold neither.

18.1 From descriptive observation to a clinical syndrome

The starting point is the clinical description associated with **Leo Kanner, an Austrian-born child psychiatrist working at Johns Hopkins University**. He is credited with the first clinical portrayal of autism in the scientific literature. The importance of that contribution lies in the act of delineation: a set of children who might otherwise have been subsumed under broad and unhelpful psychiatric categories were described as sharing a distinctive developmental pattern. Good historical reading is not hero worship. It is an appreciation that a diagnostic entity begins when clinicians can show, with enough precision, what belongs together and what does not.

The paper, ***Autistic disturbances of affective contact***, concerned a series of **11 children** accrued over 5 years. It appeared in *The Nervous Child*, volume 2, pages 217-250. The title itself is instructive. Its language belongs to its period, but it directs attention to a disturbance in the child's way of relating rather than merely to disruptive conduct or low measured ability. That emphasis helped establish autism as a problem of developmental organisation and social relatedness, rather than a label for any child who was withdrawn, unusual, or difficult to manage.

A later follow-up is a useful corrective to retrospective romanticising of early accounts. Kanner's 1971 follow-up traced 9 of the original 11 children into adulthood; only two were eventually employed, and they remained single and lived with their parents. These observations should not be converted into a modern prognostic rule. They arose from a small historical cohort and a very different service context. Their value is conceptual: a diagnostic history should remain connected to the life course, participation, support, and family circumstances of the people being described.

RULE **Read the early descriptions as clinical prototypes, not as a checklist that limits whom autism can include.** A prototype establishes a recognisable pattern; later classification must test whether its boundaries remain valid in wider clinical populations.

18.2 Asperger's parallel account and its later influence

Independently of Kanner's work, **Hans Asperger** published an account based on **4 cases**. In his 1944 paper, *Die "Autistischen Psychopathen" im Kindesalter*, he used the term “autistic personality disorder”. The parallel descriptions matter because they prevent an overly simple origin story. Autism was not manufactured by a later committee out of a single author's observations. Different clinicians, working separately, identified related developmental phenomena, while giving different weight to their most striking features.

The feature that especially distinguished that account was attention to **extraordinary skills in particular cognitive domains**. This is often simplified into a stereotype of universal talent, which is clinically unhelpful. The historical point is narrower: ability can be strikingly uneven, and the presence of a particular strength does not erase social-developmental difficulty or establish a separate disease entity. Conversely, the absence of a striking skill never argues against autism. A mature formulation should describe the person's actual profile rather than infer ability from a historical label.

The work was published in German and was not widely disseminated in English-speaking settings until nearly 40 years later. **Lorna Wing**, in 1981, introduced these findings to the

English-language literature and described Asperger's syndrome. This delay helps explain why the label became prominent after many clinicians had already learned the original account of autism. It also explains why the label acquired cultural and personal significance for some patients and families. Respecting that significance does not require preserving a classification that later evidence found difficult to delimit consistently.

- Historical labels may illuminate how a person and family first understood a developmental profile.
- They should not be treated as a shortcut to current assessment or assumptions about cognition, language, education, or independence.
- When reviewing an old record, translate the label into a current description of developmental history, functioning, and support needs.

18.3 Early labels, aetiological error, and scientific correction

An influential early label emerged when **Kanner and Leon Eisenberg, in 1956, used "early infantile autism" and emphasised obsessive insistence on sameness and extreme self-isolation**. The label concentrated attention on early developmental presentation and on behavioural patterns that clinicians continue to recognise. Yet it also shows why diagnostic terminology should not be mistaken for a complete explanation. A name can draw attention to a pattern while still carrying assumptions about timing, causation, or typical presentation that later evidence will revise.

One of the most damaging examples was the **"refrigerator mother" theory, furthered in the 1960s by Bruno Bettelheim**. It interpreted autism through a psychogenic account of parenting and emotional deprivation. Its clinical legacy is not merely historical embarrassment. It can lead to guilt, adversarial relationships with caregivers, and neglect of the child's developmental and practical needs. The lesson for a psychiatrist is to separate careful inquiry about family relationships, which remains clinically relevant for every child, from unsupported causal attribution. Families require collaborative assessment, not retrospective blame.

Empirical work displaced this theory by providing **evidence for biological and neurological foundations, while comparative studies distinguished autism from childhood schizophrenia through associated symptoms, phenomenology, age of onset, and family history**. This was a methodological advance as much as an aetiological one. It required investigators to compare syndromes rather than rely on surface impressions, and to ask whether apparently similar social withdrawal had the same developmental meaning. The distinction remains clinically valuable: resemblance in a single symptom is not enough to establish diagnostic equivalence.

TRAP **Examination trap:** do not present the refrigerator-mother account as an historical "cause" that later became less important. It was a discredited psychogenic theory, not a competing aetiology to be balanced against contemporary evidence.

For present practice, history supports a disciplined stance. A developmental diagnosis does not make family context irrelevant, nor does it justify reducing the child to biology. It means that causal claims should match evidence, while formulation still attends to caregiver stress, communication, schooling, access to services, stigma, and the child's strengths. The causes of autism and the clinical features used for diagnosis are addressed elsewhere in this chapter; history here teaches why those domains must not be confused.

18.4 DSM-III and the pervasive developmental disorder framework

Autism entered a formal diagnostic manual in **1980**, when DSM-III used the label “infantile autism”. Formal recognition mattered because it placed the presentation within a reproducible diagnostic framework rather than leaving it dependent on individual clinical tradition. It also created the possibility that criteria could be studied, criticised, revised, and compared across settings.

The broader home created for it was **pervasive developmental disorder (PDD)**, a term developed specifically for DSM-III. It denoted pervasive and severe impairment across basic functions, including socialisation and communication. The word “pervasive” captured an attempt to distinguish a broad developmental organisation from an isolated symptom. Its limitation became clearer over time: a useful umbrella can inadvertently suggest that the conditions beneath it are naturally separate, even when the boundaries between them prove unstable.

The PDD framework nevertheless performed an important transitional task. It organised clinical observation around developmental patterns and made room for presentations that did not resemble a narrow prototype. It also created a common language for research and services. Candidates should understand this as a stage in nosology, not as a rival to the current diagnosis. The historical PDD term is still encountered in case records, educational reports, and older research, so it must be recognised and translated rather than dismissed as meaningless.

There is a broader lesson in this transitional category. Classification is most useful when it makes a clinician ask better questions, not when it supplies a prematurely final answer. The PDD framework encouraged clinicians to look across domains of development, but its named divisions could tempt them to treat a label as a complete account of the person. The later spectrum formulation retained the need for careful description while moving the centre of gravity from deciding which box fits best to explaining the individual's pattern. That is a more demanding task: it requires longitudinal history, direct observation, collateral information, and attention to functioning in ordinary settings.

> The key historical transition is from separately named pervasive developmental disorders to one autism spectrum diagnosis: the clinical task moved from choosing a former subtype to applying a common framework precisely.

18.5 DSM-IV: expansion into named subtypes

The DSM-IV and DSM-IV-TR PDD category contained exactly 5 disorders: autistic disorder, Asperger's syndrome, Rett's syndrome, childhood disintegrative disorder, and PDD-NOS. This

scheme is worth knowing because it shaped generations of clinical records and examination questions. It was also an attempt to represent heterogeneity, especially differences in developmental course and the degree to which an individual resembled the original prototypes. The weakness was not that heterogeneity was unreal; it was that named categories did not reliably carve that heterogeneity at its natural joints.

DSM-IV / DSM-IV-TR SUBTYPE	DIAGNOSTIC CODE
Autistic disorder	299.00
Asperger's disorder	299.80
PDD-NOS, including atypical autism	299.80
Rett's disorder	299.80
Childhood disintegrative disorder	299.10

Do not let the table mislead you into seeing the former codes as a clinical hierarchy. Codes record a classification decision; they do not tell you how a person communicates, what repetitive patterns are present, how development unfolded, or which supports are needed. The full diagnostic criteria, severity framework, and specifiers of current ASD are addressed elsewhere in this chapter. Here, the important point is that a subtype name in an older file is historical data to be interpreted, not a final formulation.

- Do not infer that former subtypes represented neatly separated biological entities.
- Do not convert a former subtype automatically into a contemporary severity statement.
- Do preserve the original developmental account, because it may contain useful information that a code alone cannot carry.

18.6 DSM-5: from subtype boundaries to a spectrum

The decisive revision came with **DSM-5 in 2013. Autistic disorder, childhood disintegrative disorder, Asperger's syndrome, and PDD-NOS were reconceptualised as a single autism spectrum disorder**. This was not a claim that all autistic people are alike. It was a claim that the prior labels did not consistently identify separate disorders, whereas variation could be described more meaningfully within a shared diagnostic framework.

The change also altered the architecture of symptoms: the 3 DSM-IV-TR symptom categories became 2 DSM-5 categories, and the item architecture beneath them was rebuilt. DSM-IV-TR required a total of 6 or more items across its 3 categories, with at least 2 drawn from the social-interaction category, which itself listed 4; DSM-5 carries 3 items in the single social-communication and social-interaction category. The threshold rule is worth reading twice, because it is widely misquoted as though the six were six social items: the six was the total across all three categories, and the social category alone never contained six. These are historical contrasts, not a substitute for learning current criteria. The critical conceptual shift was to bring social communication and social interaction together, while retaining restricted and repetitive

patterns as the other major domain. The diagnostic-criteria discussion should be used for the present rule set; history explains why it was reorganised.

The onset wording changed as well. DSM-IV-TR required onset before age 3, whereas DSM-5 specifies early development. This prevents an arbitrary historical cut-off from overriding a careful developmental history. It does not mean that onset history has become unimportant. Rather, the clinician has to establish whether the overall developmental pattern is compatible with ASD and whether later recognition reflects changing social demands, masking, missing early history, or another explanation.

RULE **Spectrum does not mean vague.** It replaces unstable subtype labels with a common diagnostic framework while requiring a more precise individual description of developmental history and current functioning.

18.7 Rett syndrome, coding legacies, and reading old records safely

The spectrum consolidation had an explicit exception. **Rett syndrome was not retained as a distinct autism-spectrum category; it is cited under the associated-condition specifier as an example of a known genetic condition.** This is a useful guard against a common historical error: assuming that every condition previously housed within PDD was simply renamed ASD. The detailed clinical account of Rett syndrome and childhood disintegrative disorder belongs with their dedicated discussion; the classification point here is that historical placement and present diagnostic status are not the same thing.

The manual pairs that reclassification with a differential instruction, set out where each of those syndromes is discussed in full. The classification principle is what belongs here, and it generalises: a symptom that appears during a particular phase of another condition must not be treated as sufficient evidence for a lifelong co-occurring diagnosis. Diagnostic overlap calls for full criteria, developmental context, and longitudinal reasoning.

Older ICD-10 documentation may use the following labels. The list is useful for record translation, but it must not be read as a DSM-IV row-by-row conversion.

ICD-10 CODE	LABEL PRINTED IN THE HISTORICAL PDD LISTING
F84.0	childhood autism
F84.1	atypical autism
F84.2	Rett's syndrome
F84.3	childhood disintegrative disorder
F84.4	overactive disorder associated with mental retardation and stereotyped movements
F84.5	Asperger syndrome
F84.8	other pervasive developmental disorders
F84.9	pervasive developmental disorders, unspecified

For the ICD-11 ASD stem, the back-mapping permits F84.0 childhood autism, F84.1 atypical autism, or F84.5 Asperger syndrome; use F84.1 when onset before age 3 is unclear, and use F84.5 regardless of onset when there is no general impairment in intellectual functioning or functional language. This is a coding bridge, not a licence to reintroduce obsolete diagnostic distinctions into the clinical formulation.

Finally, modern ASD encompasses historical terms that will appear in legacy material:

- early infantile autism, childhood autism, and Kanner's autism
- high-functioning autism, atypical autism, and PDD-NOS
- childhood disintegrative disorder and Asperger's disorder

GOOD TO REMEMBER

When encountering a historical label, first establish which classification system was used, then recover the descriptive evidence behind it, and finally apply the current framework without erasing the person's history. Taxonomy should improve clinical understanding, not replace it.

19 · Diagnostic criteria of ASD (DSM-5 dyad, ICD-11)

The diagnostic task is to recognise a pattern, not to count isolated oddities. Autism spectrum disorder is formulated from a persistent developmental pattern that is organised around **two** core domains. The value of the formal criteria is that they force the clinician to establish both the social-communication disturbance and the restricted or repetitive pattern, then to ask whether the pattern began developmentally and matters in the person's present life. This protects against diagnosing autism from one striking mannerism, a narrow interest, or social discomfort alone.

The criteria are best understood as a clinical argument. Symptoms must be described in their developmental context, linked to real-world functioning, and weighed against the person's general developmental level. The classification language is concise; the clinical work is not. A good answer therefore states the diagnostic structure first and then explains why each element is necessary. This section addresses the criteria in DSM-5-TR and ICD-11, before showing how their different formats lead to the same disciplined diagnostic reasoning.

2	3	at least 2	6A02
DSM CORE DOMAINS	CRITERION A ELEMENTS REQUIRED	CRITERION B ELEMENTS REQUIRED	ICD-11 PARENT STEM

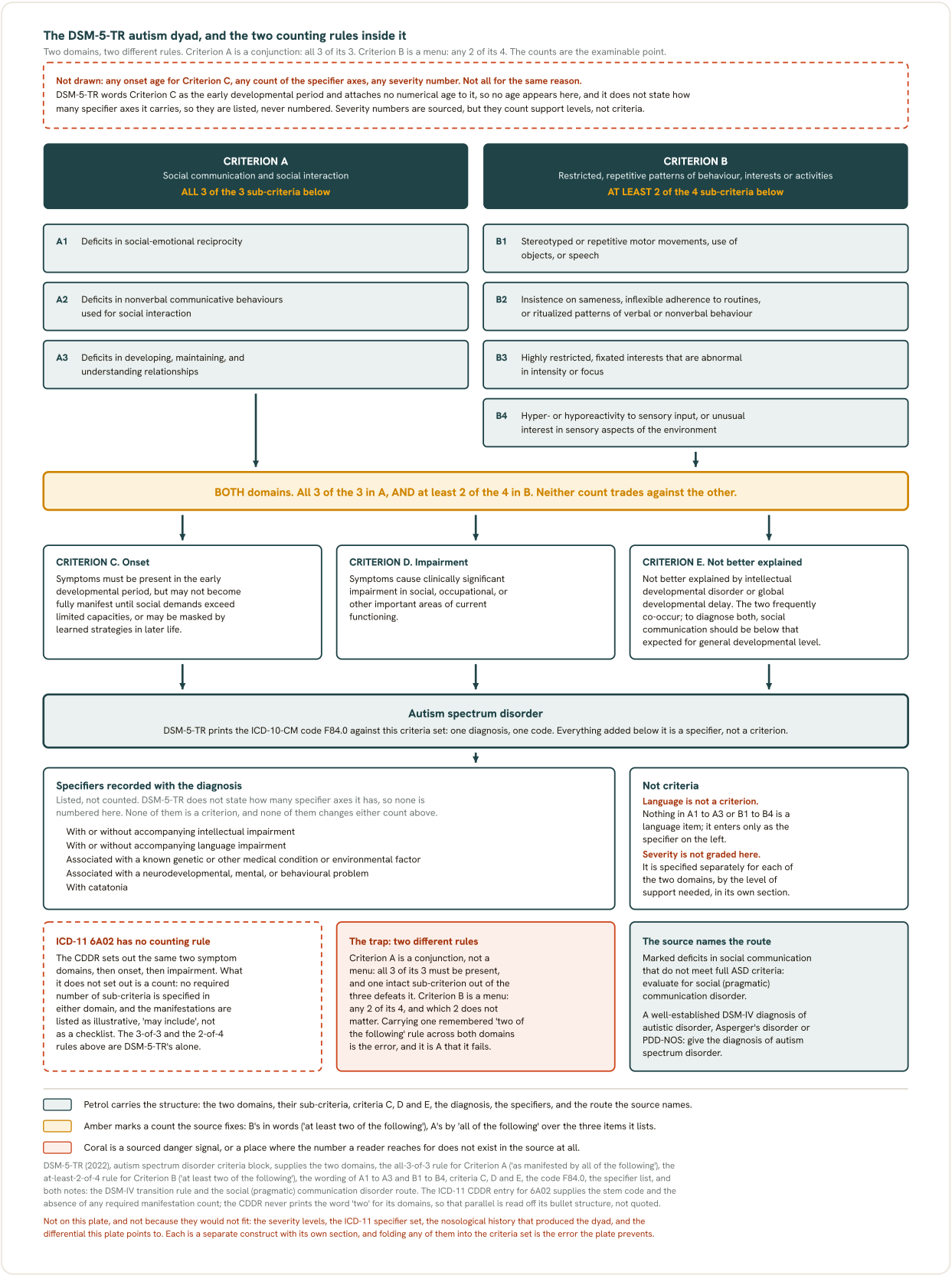


Fig 21.12 The DSM-5-TR autism dyad, and the two counting rules inside it. Two symptom domains, two different rules. Criterion A needs all 3 of its 3 sub-criteria, so one intact sub-criterion defeats it; Criterion B needs any 2 of its 4, and which 2 does not matter. Criteria C, D and E add onset, impairment and the not-better-explained rule against intellectual developmental disorder. Specifiers hang off the diagnosis, not the counts, where language and severity sit, outside the criteria set. ICD-11 6A02 has the same two domains and no counting rule, so these counts are DSM-5-TR's alone. No onset age is drawn: DSM-5-TR attaches none to the early developmental period, and the specifier axes are listed, never numbered, because it does not state how many there are.

19.1 The diagnostic architecture: a dyad with developmental and functional anchors

The DSM-5-TR dyad has one domain concerning social communication and social interaction, and another concerning restricted, repetitive patterns of behaviour, interests, or activities. The dyad is not merely a shorter checklist. It requires the examiner to decide whether social difficulty is part of a broader reciprocal-interaction disturbance and whether the behavioural pattern has the characteristic quality of repetition, inflexibility, restriction, or unusual sensory engagement.

A useful way to read the criteria is vertically as well as horizontally. Vertically, the clinician first establishes the core phenotype, then asks about developmental onset, present impairment, and competing explanations. Horizontally, each core domain must be examined across ordinary contexts rather than inferred from a single consultation. A person may use rehearsed social behaviours in a structured setting and still have difficulty when reciprocity, flexibility, or contextual understanding are demanded. Conversely, a single unusual habit without the wider pattern does not establish the diagnosis.

This structure also explains why a symptom inventory is insufficient. Checklists are useful prompts for enquiry, but their items are not interchangeable. An affirmative response about a repetitive act does not repair missing evidence from the social domain, and a history of social discomfort does not establish the repetitive domain. The clinician must decide whether each observation belongs to the required construct, whether it is persistent, and whether the observations form one developmental syndrome. That is the safeguard against both overinclusive diagnosis and the dismissal of a coherent but subtle presentation.

RULE **Diagnostic rule:** establish the complete pattern across both core domains before using onset, impairment, and exclusion criteria to confirm that it is autism spectrum disorder.

The printed DSM diagnosis carries the code F84.0. In an examination answer, however, the code is less important than showing the logic of the dyad. The examiner should be able to see that the candidate understands why a social-communication presentation without the required restricted or repetitive pattern is not simply labelled autism by default.

19.2 Criterion A: social communication and social interaction

Criterion A concerns deficits that are persistent and span the social-communication field. DSM-5-TR requires **all three** listed components. This matters because conversational difficulty, poor eye contact, or social isolation in isolation can arise for many reasons. The criterion asks for a coherent disturbance in reciprocity, in the use of nonverbal communication for social purposes, and in relationships.

- **Deficits in social-emotional reciprocity** concern the give-and-take of social exchange. The clinical question is whether the person can initiate, respond to, and sustain a reciprocal interaction in a manner appropriate to the context.
- **Deficits in nonverbal communicative behaviours used for social interaction** concern how nonverbal signals are used to regulate and share interaction, rather than a superficial judgement about one gesture or one gaze behaviour.

- **Deficits in developing, maintaining, and understanding relationships** require attention to the person's grasp of social expectations, adjustment of behaviour to context, and capacity to participate in relationships.

The phrase “social communication” should not be reduced to spoken language. A fluent speaker can have marked difficulty with reciprocity, social interpretation, or relationships, while a person with limited speech may show a clear wish to engage and share. The diagnostic question is how social communication functions between people. **Language impairment is not itself a core DSM-5-TR criterion**; its specifier framework is addressed in the severity-level and specifier discussion in this chapter. Detailed language phenomenology belongs with the clinical features of ASD.

Clinical interpretation. Criterion A is often easiest to misread when an observer relies on a socially compelling moment during an interview. The purpose is not to decide whether a person can make contact at all; it is to understand the consistency, flexibility, reciprocity, and context-sensitivity of social communication over development. A single competent performance, particularly in a familiar or highly structured interaction, is not by itself a substitute for that broader account.

For the same reason, Criterion A should not be phrased as a personality judgement. Reserve descriptive language for observable interaction and its functional meaning. Ask how the person shares attention, responds when another person changes the topic, reads the interpersonal situation, and adapts communication to the relationship. Such questions convert a loose impression of being “socially awkward” into evidence that can be placed, or not placed, within the criterion. They also make it easier to explain the reasoning to the person and family without reducing the assessment to a label.

19.3 Criterion B: restricted and repetitive patterns

Criterion B captures a style of behaviour and interest that is repetitive, circumscribed, inflexible, or unusually related to sensory experience. DSM-5-TR requires **at least two** of the listed manifestations. The threshold deliberately prevents a single preference, hobby, or repetitive action from carrying the diagnosis. What counts is the pattern's persistence, context, intensity, and relationship to the rest of the clinical picture.

The manifestations include stereotyped or repetitive motor movements, use of objects, or speech. The point is not simply that behaviour is repeated. Repetition must be interpreted as part of the characteristic pattern, rather than as an isolated self-soothing act, play preference, or response to a transient stressor.

A further manifestation is insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behaviour. Here, the clinical task is to ask what happens when an expected sequence changes. The diagnostic relevance lies in the rigidity and the way the need for predictability shapes functioning, not in whether a household happens to have routines.

The domain also includes highly restricted, fixated interests that are abnormal in intensity or focus. A strong interest should be evaluated for its quality and consequences, not judged by whether its topic seems unusual to the clinician. An ordinary subject can become diagnostically relevant when engagement is unusually narrow or dominant; an unusual subject can be entirely non-pathological when it is flexible and proportionate.

Finally, DSM-5-TR includes **hyper- or hyporeactivity to sensory input, or unusual interest in sensory aspects of the environment**. This places sensory experience within the restricted and repetitive domain. It does not mean that sensory sensitivity alone is diagnostic. Its value is greatest when it helps explain a broader pattern of behaviour, distress, avoidance, fascination, or difficulty adapting to ordinary environments.

MNEMONIC

Memory spine: A is all, B is a selected set

Establish every social-communication element in A, then establish the required threshold within B. Use this as a recall aid only after understanding what the observations mean clinically.

TRAP **Examination trap:** do not diagnose ASD from social-communication deficits alone. The restricted and repetitive domain is required, and it must be demonstrated rather than assumed.

19.4 Criteria C to E: timing, impact, and developmental explanation

The core phenotype is necessary but not sufficient. Criterion C requires that symptoms are present in the **early developmental period**. Recognition may occur much later because demands become more complex or because a person has learned strategies that conceal difficulty. The criterion is therefore about developmental presence, not the age at which a family, school, or clinician first names the concern. A late recognition narrative should prompt careful developmental reconstruction, not an automatic rejection of ASD.

Criterion D requires **clinically significant impairment** in social, occupational, or other important areas of current functioning. Impairment is the bridge from descriptive traits to a disorder. It should be considered in the person's actual environment, including the effort needed to maintain functioning and the consequences when familiar supports are absent. The clinician should avoid both extremes: treating every difference as disorder, or dismissing substantial hidden burden because outward performance appears adequate.

Criterion E says that the presentation must not be better explained by intellectual developmental disorder or global developmental delay. It also permits co-occurring diagnoses when **social communication is below that expected for general developmental level**. This is a comparative judgement, not a shortcut based on an intelligence score. The issue is whether the social-communication profile is disproportionately affected relative to the person's overall developmental functioning. Intellectual disability and its assessment are covered in the relevant sections of this chapter.

- **Timing:** establish a developmental history that can account for the current pattern, including why it became visible when it did.
- **Impact:** describe what the pattern changes in daily participation, relationships, education, work, or other important roles.
- **Explanation:** decide whether the social-communication disturbance exceeds what would be expected from the person's general developmental level.

The transition note is clinically important: an individual with a well-established DSM-IV diagnosis of autistic disorder, Asperger's disorder, or PDD-NOS should receive the diagnosis of autism spectrum disorder. This is a diagnostic transition rule, not an invitation to recreate older subgroup boundaries in current formulation. For a person with marked social-communication deficits who does not meet the full ASD criteria, the DSM directs that they should be evaluated for social (pragmatic) communication disorder. The full differential is considered elsewhere in this chapter.

19.5 ICD-11: essential features and a clinically judged threshold

ICD-11 places ASD under the parent stem 6A02. Its essential features have two required symptom domains, structurally paralleling the DSM dyad. The practical message is the same: social-communication disturbance and restricted or repetitive patterns are both indispensable to the syndrome.

The important structural contrast is that ICD-11 does not set a numerical sub-criterion threshold within either domain. Instead, manifestations are illustrative rather than a required count. This calls for disciplined clinical judgement, not diagnostic laxity. The clinician must still establish that the required domains are convincingly represented and persistent. A list of examples is not a licence to infer a domain from a vague or isolated concern.

When working from ICD-11, document the evidence that makes each domain essential rather than simply reproducing several examples. This keeps the record auditable: another clinician should be able to see why the pattern was judged to meet the domain, how it has persisted developmentally, and how it affects functioning. The same discipline is useful when DSM criteria are used. Counting is a decision aid, not a replacement for a coherent account of the person's development and current life.

The ICD onset requirement is that onset occurs during the developmental period, usually becoming apparent early, although characteristic symptoms may become fully manifest only when social demands exceed the person's capacities. Its functional requirement is significant impairment in important areas of functioning; diagnosis remains appropriate when adequate functioning is achieved only through exceptional effort. This wording is especially useful for presentations in which compensation can obscure the burden of the condition.

ICD-11 also notes that a true regression pattern may typically begin between **1 to 2 years of age**, while loss of previously acquired skills is rarely observed above **3 years of age**. This is a narrow

classification observation, not a substitute for the clinical assessment of regression or its differential diagnosis.

19.6 DSM-5-TR and ICD-11: compare the logic, not merely the wording

DIAGNOSTIC AXIS	DSM-5-TR	ICD-11	CLINICAL CONSEQUENCE
Core structure	Two core domains	Two essential symptom domains	Both require a dyadic phenotype rather than a single-domain formulation.
Within-domain threshold	Every Criterion A component is required; Criterion B requires its stated minimum.	No required sub-criterion count is specified.	DSM is more operationally counted; ICD relies more explicitly on judgement about the essential pattern.
Developmental timing	Symptoms are present in the early developmental period.	Onset occurs during the developmental period.	Recognition can be delayed without converting a developmental condition into an adult-onset one.
Functional significance	Clinically significant current impairment is required.	Significant impairment remains relevant despite exceptional compensatory effort.	Assess current consequences and effort, not symptoms in abstraction.

The classifications should therefore not be treated as rival definitions. They are two formats for the same central clinical problem: determining whether a developmental social-communication pattern is accompanied by the required restricted or repetitive pattern and causes meaningful difficulty. DSM makes the internal threshold more visible. ICD makes the need for judgement in a variable phenotype more visible. In either system, the best formulation is evidence-based, developmental, and functionally anchored.

19.7 A disciplined way to write and defend the diagnosis

GOOD TO REMEMBER

In practice, begin by naming evidence under each core domain. Then state the developmental course, the present functional impact, and the developmental comparison required when intellectual disability or global developmental delay is relevant. This sequencing prevents the common error of beginning with a favourite label and searching selectively for confirming behaviours.

A concise formulation can therefore be written as a chain of reasoning: the person has evidence for the social-communication domain; evidence for the restricted or repetitive domain; a developmental history compatible with the condition; current functional consequences; and no more complete developmental explanation for the social-communication profile. If any link is weak, the right response is to clarify that link rather than to stretch another criterion. This

approach is especially valuable when the presentation is compensated, when history is incomplete, or when more than one developmental difficulty is present.

Do not turn the criteria into a proxy severity scale. Severity levels and accompanying intellectual or language impairment are specifiers, not replacements for the core diagnostic argument. Likewise, screening and diagnostic instruments may organise information, but they do not remove the need to establish the criteria. Their selection and use are addressed in the assessment section of this chapter.

> ASD requires the dyad, developmental presence, clinically meaningful impact, and a developmental explanation that does not account for the social-communication profile more fully.

The final formulation should make its reasoning transparent. It should show why the observed pattern belongs together, why it is developmental, why it matters now, and why the chosen diagnosis is more accurate than a partial description. That is the difference between reproducing a checklist and using a diagnostic classification responsibly.

20 · Epidemiology and changing prevalence of ASD

Epidemiology is more than a prevalence figure. In autism spectrum disorder, the number reported by a survey depends on whom it reaches, how cases are found, which diagnostic concept is used, and whether a record, a service register, or direct assessment establishes the diagnosis. The examination task is therefore not to recite one figure as though it were universal. It is to state the population and method to which a figure applies, distinguish occurrence from detection, and explain why apparently conflicting estimates may all be credible within their own settings.

Autism is diagnosed across the life course, but ascertainment is shaped by developmental surveillance, referral pathways, educational provision, clinical expertise, and families' ability to reach assessment. These influences make epidemiology clinically relevant. They determine which children come to attention, which adults remain unidentified, and how a service should interpret a rising caseload. Prevalence describes the burden recorded in a defined population; it should not casually be converted into a claim about causation or a proof that the underlying incidence has changed.

20.1 Reading an ASD prevalence estimate

Prevalence is the proportion of a defined population meeting a case definition at a stated time or over a stated period. For ASD, it is an output of both the condition in the population and the process used to recognise it. A direct, structured assessment can identify people who have never entered services, whereas an administrative dataset captures only those who have acquired a diagnostic label. Neither approach is automatically superior in every setting: each answers a somewhat different question.

A useful distinction is between a population survey, a clinical series, and a service count. A clinic denominator is strongly influenced by referral, affordability, and local diagnostic practice. A

school or disability register may be especially sensitive to eligibility rules. A population study can reduce some of these effects, but still depends on participation and on the screening and diagnostic pathway. Thus, when comparing papers, the candidate should ask: who was sampled, what constituted a case, and how was that case established?

Case definition is equally important. Research that asks whether a person meets a current diagnostic formulation is not directly equivalent to a historical study that used a narrower formulation. Nor is a parent-reported label equivalent to an assessment made after direct observation and developmental history. The point is not to rank methods in the abstract. It is to understand the direction in which each method may shape a result and to avoid comparing unlike estimates as though they measured precisely the same construct.

For clinical readers, this also explains why a service's experience can diverge from published population data. A tertiary service may receive people with complex presentations, delayed recognition, or accompanying needs that make referral more likely. A community survey may capture people whose needs have not brought them to a specialist. Both can be valuable sources of information, but neither should be used to invalidate the other without considering its sampling frame. Epidemiology becomes clinically useful when it trains that habit of interpretation.

RULE **Never quote an ASD frequency without naming its population, period, and ascertainment pathway.** The same label can yield different observed frequencies when the pathway into the numerator changes.

Ascertainment means the process by which potential cases are found and verified. It is not a minor methodological footnote. A wider net may identify people whose presentations were previously overlooked; an altered threshold may change who qualifies; and better access to assessment may turn an unmet need into a recorded diagnosis. These processes can change measured prevalence even when the underlying population risk has not changed.

- State whether the estimate is local, national, or multinational.
- Identify whether it is a survey estimate, a diagnosed-rate estimate, or a service-derived estimate.
- Describe a temporal increase as a change in **measured prevalence** unless the evidence separately establishes an increase in incidence.

20.2 Contemporary frequency: use the denominator carefully

Published figures support a broad conclusion: ASD is not rare in contemporary clinical and public-health practice. They do not support treating one jurisdiction's figure as the world's figure. The DSM-5-TR describes a reported frequency in the United States of **between 1% and 2%**. In the same source, the median global prevalence is **0.62%**, while prevalence across non-United States countries has approached 1%. These are estimates from differently assembled evidence, not interchangeable constants.

1-2%	0.62%	approaching 1%	3:1
UNITED STATES REPORTED FREQUENCY	MEDIAN GLOBAL PREVALENCE	NON-UNITED STATES REPORTS	WELL-ASCERTAINED EPIDEMIOLOGICAL SEX RATIO

The Kaplan & Sadock's Comprehensive Textbook of Psychiatry gives a current estimate of about 69 per 10,000, or about 1 in 145 children. Lewis's Child and Adolescent Psychiatry describes 6.6 per 1,000 as a reasonable overall estimate. The immediate lesson is not to calculate a false reconciliation between these reports. Rather, each estimate needs its source and denominator attached when it is quoted.

IN INDIA

An international or United States surveillance number may be useful as context, but it must not be relabelled as an Indian prevalence estimate. The Indian Psychiatric Society guideline cites a United States surveillance estimate of 1 in 68; its provenance must travel with the number. A responsible answer acknowledges the need for locally representative ascertainment rather than importing a denominator from another health and education system.

20.3 Sex and gender: distinguish epidemiology from diagnosis

Sex ratio is often quoted as a compact epidemiological fact, but it carries an important methodological warning. In well-ascertained epidemiological samples, the global male:female ratio is 3:1. That figure should not be conflated with the ratio among people who receive a diagnosis. The DSM-5-TR states that ASD is diagnosed between three and four times more often in males, while the ICD-11 Clinical Descriptions and Diagnostic Requirements describes males as diagnosed at a 4:1 male:female ratio.

The difference between a diagnostic ratio and an epidemiological ratio matters because diagnosis is a social and clinical event. Referral expectations, recognition of less stereotyped presentations, language and cognitive profile, and co-occurring difficulties can all shape who crosses the threshold into a dataset. Therefore, a male predominance in a clinical register must not be interpreted automatically as a direct measure of the sex distribution of autistic traits in the wider population.

Other textbook summaries similarly report a male excess, but their scope should be retained. Kaplan & Sadock's Comprehensive Textbook of Psychiatry reports on the order of three to five boys for every girl in clinical and epidemiological samples, and notes that the predominance is greater among those with higher cognitive ability. Lewis's Child and Adolescent Psychiatry reports that boys are affected between three and four times more often. These formulations should be presented as source-specific descriptions, not as permission to under-recognise ASD in girls or women.

TRAP **Examination trap:** a diagnosed male:female ratio is not automatically a population prevalence ratio. Name the numerator before interpreting the ratio.

20.4 Why recorded prevalence has risen

The historical contrast is striking, but the correct inference is nuanced. Early epidemiological studies described autism as roughly 1 per 2,000, whereas current estimates are much higher. This temporal pattern establishes that the recorded frequency has changed. It does not, by itself, identify why it changed. Trends are especially difficult to interpret when the diagnostic category, awareness, service incentives, and methods of finding cases evolve together.

REPORT	FIGURE AND INTENDED USE	INTERPRETIVE BOUNDARY
Early studies	Roughly 1 per 2,000	Historical baseline, not a current estimate.
Kaplan & Sadock	About 69 per 10,000, or about 1 in 145 children	Contemporary textbook estimate.
Lewis	6.6 per 1,000	Reasonable overall estimate in that text.
Indian Psychiatric Society guideline	1 in 500 historically, compared with 1 in 68	Both figures are historical comparison and United States surveillance context, not Indian population prevalence.

Secular increase refers to an apparent change across time. It may reflect a real change in occurrence, an altered measurement process, or both. Lewis interprets the **apparent rise** through evolving diagnostic concepts, broader recognition of the spectrum, and access to relevant services, rather than as proof that population occurrence has increased. This is a disciplined reading of time trends: the conclusion should remain proportionate to what the underlying studies can establish.

The evidence base has expanded substantially. Kaplan and Sadock refers to over 60 epidemiological studies. More studies improve the visibility of variation and methodological limitations; they do not erase those limitations. An increased research base should make the clinician more careful about denominators, not more casual about generalisation.

20.5 Mechanisms that inflate or alter measured frequency

Diagnostic broadening or change in diagnostic practice can alter measured frequency. When a category is conceptualised differently, people previously placed outside it, or given another developmental label, may now meet the contemporary definition. Awareness also matters: families, teachers, and clinicians who recognise a broader range of presentations are more likely to refer and document concerns. Service availability can have a similar effect, because visible assessment and educational support make diagnosis more feasible and more relevant to a family.

Diagnostic substitution is a related, but more specific, phenomenon. In the United States in particular, a single diagnostic label is commonly used to identify students for service eligibility. As awareness rose, schools and parents became more likely to use the autism label; estimates can consequently be inflated when service records, rather than direct assessment of children, establish caseness. This is not a claim that every increase is artificial, nor should this United States-specific mechanism be exported uncritically to all countries.

Kaplan and Sadock likewise regards **the measured rise** as partly shaped by evolving diagnosis, professional practice, educational policy, and public recognition; higher rates often occur where awareness and services are more available. It leaves the possibility of a true incidence increase unresolved. That restraint is the correct clinical posture. A service observing more diagnosed children may be seeing a mixture of changing need, changing recognition, and changing access.

MNEMONIC

FRAME the rising count

Find out how cases were found; **R**eview the diagnostic rule; **A**sk about awareness and access; **M**ap service and educational incentives; **E**xpress uncertainty about true incidence.

- Do not call a rise an epidemic merely because a surveillance figure rose.
- Do not dismiss all increase as artefact merely because ascertainment has improved.
- State which mechanisms are supported by the source and which remain unresolved.

20.6 Ethnoracial, social, and cultural interpretation

Within United States child data reported in the DSM-5-TR, prevalence is lower in African American children at 1.1% and Latinx children at 0.8% than in White children at 1.3%, even after socioeconomic resources are taken into account. This is an observed comparison in that source. It should not be rewritten as established proof that the difference is wholly caused by underdiagnosis or differential ascertainment. The same source says reported prevalence may be affected by misdiagnosis, delayed diagnosis, or underdiagnosis, which is a cautious possibility rather than a completed causal explanation.

Ascertainment bias is the distortion introduced when some people are more likely than others to be identified, referred, or recorded. It is a useful hypothesis when interpreting group differences, but it is not a substitute for evidence. A good answer separates the observed distribution from proposed explanations and avoids converting a plausible clinical concern into a settled epidemiological conclusion.

Social-class arguments require the same historical discipline. **Kanner's original observation** involved highly achieving, high-social-status parents, but Kaplan and Sadock regards the sample as likely biased by case ascertainment and notes that epidemiological studies have generally not found such an association. The lesson extends beyond ASD: impressions formed from a selective clinic population should never be mistaken for a population association.

20.7 Applying epidemiology in clinical and service work

What the clinician should do with a rate. Epidemiology should improve case-finding and service planning, not replace individual assessment. A family does not become more or less likely to need a careful developmental evaluation because the clinician has memorised a national estimate. Conversely, a service cannot plan workforce, assessment capacity, or educational liaison responsibly if it assumes that a historical clinic count captures all people who may need support.

GOOD TO REMEMBER

Treat an apparent rise in caseload as an audit question, not a reflex conclusion. Check referral, assessment pathways, education or disability processes, age groups, and the catchment population. This helps distinguish increased recognition from a changed denominator, so planning is based on demand visible to the service.

At the level of a research appraisal, ask whether the study has separated screening from confirmation and whether non-participants might differ from participants. Ask whether a diagnostic record had to pre-exist for inclusion, and whether the study would identify people with a subtle or previously unrecognised presentation. Such questions are transferable across settings. They also keep the clinician from treating an attractive headline figure as more precise than the study design permits.

When counselling families, use population data with humility. Explain that frequency figures vary across settings and methods, and that an epidemiological association cannot determine the cause of one person's ASD. Avoid implying blame from parental characteristics, social position, or the fact that a diagnosis became visible after a change in school or service access. The aetiology of ASD, including genetic and environmental architecture, belongs in the etiological discussion of this chapter; epidemiology here explains how cases are distributed and counted.

For a written answer, a compact but high-quality structure is: define prevalence and ascertainment; quote a source-qualified contemporary estimate; state the male predominance while distinguishing diagnosed from epidemiological ratios; then explain the rise through diagnostic concepts, awareness, practice, educational policy, and service access. End with the unresolved point: changing measured prevalence alone cannot settle whether underlying incidence has changed.

The rise in recorded ASD prevalence is real as a measurement trend; its causes are multiple, and it must not be equated automatically with a proven rise in true incidence.

20.8 High-yield synthesis

The epidemiology of ASD is best understood as a conversation between population occurrence and the systems that recognise it. Contemporary estimates demonstrate substantial need, while their variation demonstrates why source, denominator, geography, and ascertainment must be stated. Sex ratios have the same requirement: a well-ascertained epidemiological ratio and a diagnosed-rate ratio answer different questions. Historical increases should prompt examination of diagnostic change, awareness, education, services, and record-based case definition before claims are made about causation.

This approach protects against opposite errors. Minimisation assumes that ASD is uncommon because an older estimate was lower. Overstatement treats every rise in a recorded rate as proof of a new environmental cause. The clinician's role is to recognise the public-health burden, assess

the individual in front of them, and communicate uncertainty precisely. That combination is more useful than a detached list of figures and more defensible than a single dramatic number.

21 · Etiology of ASD (genetic, neurobiological, environmental)

Aetiology is plural, not a single cause. Autism spectrum disorder arises through developmental pathways in which inherited liability, rare genetic events, brain development and environmental exposures may intersect. The clinical task is therefore not to offer families a simplistic cause, but to explain that association is not destiny, that aetiological formulation can identify relevant medical conditions, and that a cause is not required before support can begin. The examination task is similar: distinguish evidence for familial aggregation from proof of an individual mechanism, and distinguish a risk factor from a cause.

The evidence is strongest for a substantial genetic contribution, but ASD is genetically heterogeneous. Its neurobiology is likewise a set of converging developmental observations rather than a diagnostic test. Environmental findings need particularly careful reading because many exposures are correlated with parental characteristics, obstetric events and ascertainment. A good answer therefore moves from genetic architecture to neurodevelopmental correlates, then appraises environmental association and vaccine misinformation without conflating any of these levels of explanation.

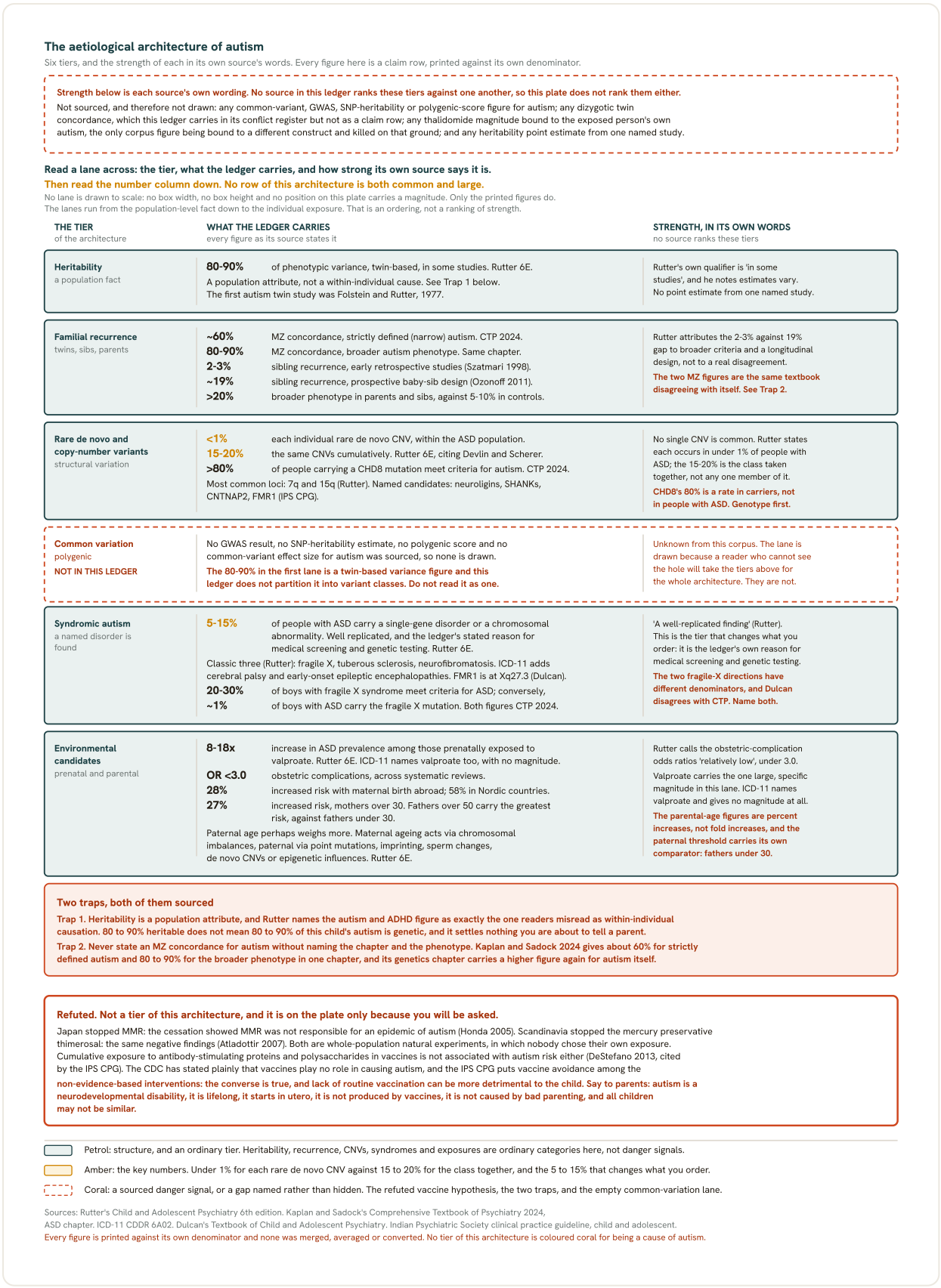


Fig 21.13 The aetiological architecture of autism. Six tiers, each printed with the strength its own source claims for it and against its own denominator. The plate teaches one shape: no row beneath twin-based heritability is both common and large, and the one row the ledger ties to a clinical action is syndromic autism. The common-variation lane is drawn empty because no such figure was sourced, and a hole in the map is not a hole in the world. The two traps and the refuted vaccine hypothesis are marked on the plate itself.

21.1 What an aetiological formulation means in ASD

Aetiological heterogeneity means that similar clinical phenotypes can arise through different causal routes. In practice, this prevents the error of treating all ASD as one disease with one lesion. Some presentations occur alongside a recognisable medical or genetic disorder; others reflect the combined effect of many inherited and developmental influences.

> **GOOD TO REMEMBER**
>
> Use the formulation to connect a person's developmental history, physical examination and family pattern to an appropriate medical work-up; choice of ASD genetic tests belongs to the ASD medical workup section.

RULE **Read causation at the right level:** a population estimate of genetic influence does not tell us what caused ASD in one child.

Heritability is a population statistic: it describes the share of variation in a population that is associated with genetic differences under the conditions studied. In some studies, twin-based estimates for autism lie **between 80-90** percent of phenotypic variance. This is compelling evidence that genetic factors matter, but it does not mean that this proportion of any individual's ASD was "caused by genes," nor does it make environmental influences irrelevant. Heritability can change when a population or its exposures change.

Historical twin work made the importance of genetic factors visible after **Folstein and Rutter, 1977**. Twin and family designs are complementary. Twin concordance asks whether co-twins share the phenotype; family studies ask whether traits cluster among relatives. Neither design, on its own, identifies a gene or predicts the outcome for an individual pregnancy. A careful answer also states whether it is discussing narrowly defined autism or the broader phenotype.

approximately 60%	80-90%	approximately 19%	5-15%
MZ CONCORDANCE: NARROWLY DEFINED AUTISM	MZ CONCORDANCE: BROADER AUTISM PHENOTYPE	PROSPECTIVELY FOLLOWED INFANT SIBLINGS	SINGLE-GENE DISORDER OR CHROMOSOMAL ABNORMALITY

21.2 Familial aggregation and the broader phenotype

For strictly defined autism, monozygotic twin concordance is approximately 60%. When the outcome includes the **broader autism phenotype**, concordance is between 80-90%. The broader phenotype refers to subthreshold autistic-like traits in relatives, not to an assertion that every relative has ASD. This contrast is clinically instructive: genetic liability may be expressed as traits rather than as the full diagnostic phenotype, and the phenotype being measured changes the estimate.

Older retrospective family studies reported sibling recurrence between 2-3%. In prospectively followed infant siblings, the estimate is approximately 19%. These figures should not be presented as a contradiction. The later design observes siblings from birth and works with broader contemporary diagnostic ascertainment; both features can increase detection. For

counselling, it is more honest to explain that familial recurrence is elevated and that an individual family needs a personalised assessment rather than a memorised population figure.

Subthreshold autistic-like traits occur in above 20% of parents and siblings of ASD probands, compared with between 5-10% in control families. Such aggregation supports a dimensional account of inherited liability. It must not be turned into blame: a parent's communication style, temperament or isolated trait is not evidence that parenting caused ASD.

TRAP **Examination trap:** do not quote the broader-phenotype twin figure as the concordance for narrowly defined autism, and do not use an early retrospective sibling estimate as though it came from a prospective infant-sibling design.

21.3 Genetic architecture: common liability, rare variants and syndromic ASD

Genetic architecture describes the kinds of genetic contribution found across a condition, rather than seeking one "autism gene." ASD includes polygenic liability, rare inherited or de novo variation, copy-number variation and recognised syndromic disorders. These categories can overlap in a person, and the presence of a genetic finding should be interpreted alongside the phenotype. A result may clarify associated medical surveillance and family discussion, but it does not reduce the child to a laboratory label.

ARCHITECTURE	WHAT IT CONVEYS CLINICALLY	IMPORTANT DISTINCTION
Familial liability	Traits and diagnoses may aggregate in relatives.	Aggregation is not a single-gene explanation.
Rare de novo copy-number variation	A rare genomic event may contribute substantially in an affected individual.	Each individual rare event is below 1% of the ASD population, yet such variants cumulatively account for between 15-20%.
Chromosomal abnormality	May place ASD in a broader medical and developmental formulation.	Reported abnormalities involve many loci; 7q and 15q are described as common sites.
Syndromic association	Prompts attention to syndrome-specific physical and neurological features.	Association does not imply that every person with ASD has a syndrome.

Recognisable syndromic associations include fragile X, tuberous sclerosis, and neurofibromatosis. ICD-11 additionally lists cerebral palsy and early-onset epileptic encephalopathies among medical disorders associated with ASD, alongside chromosomal abnormalities and neurocutaneous disorders. These are associations that enrich clinical assessment; syndrome-specific genetics, phenotype and recurrence belong to their respective disorder sections.

Copy-number variation refers to a genomic deletion or duplication. The useful clinical contrast is frequency against collective contribution: any one rare de novo CNV occurs in **below 1%** of the ASD population, but the group taken together may account for between 15-20%. This is why a negative search for one famous variant never excludes a genetic contribution, and why genetic architecture cannot be understood by memorising a short gene list.

- Candidate genes named in the Indian guideline include neuroligins, SHANKs, CNTNAP2 and FMR1.
- FMR1 is the fragile X gene, and it is the single commonest monogenic contributor a candidate is asked to name here; its locus, repeat mechanism and phenotype are taught with the syndrome itself.
- CHD8 mutations illustrate a genotype-first association: above 80% of carriers meet diagnostic criteria for autism spectrum conditions.

Direction of a proportion is a frequent exam trap. Between 20-30% of boys with fragile X syndrome meet criteria for ASD, whereas approximately 1% of boys with ASD carry the fragile X mutation. These are different denominators and neither can be inverted. This distinction is also why a syndrome association supports targeted clinical consideration, not indiscriminate causal certainty.

21.4 Neurobiological findings: correlates, pathways and limits

Neurobiology informs models; it does not diagnose ASD. Studies of neurotransmitters, brain growth, imaging and postmortem tissue have identified group-level differences. They are heterogeneous, often developmentally timed, and may be affected by sampling, clinical variation and methodology. The appropriate conclusion is that ASD involves altered neurodevelopmental processes, not that any one scan, blood measure or neuropathological feature establishes the diagnosis.

Peripheral hyperserotonaemia is the most consistently replicated biochemical observation: elevated serotonin in blood and platelets is reported in between 25-50% of children and adolescents with autism. It is peripheral, not a direct measure of cerebrospinal fluid or brain serotonin. A candidate should therefore resist the tempting but invalid inference that it specifies a universal central neurotransmitter abnormality or tells the clinician how to treat an individual child.

Structural findings have focused on development and timing. In the first MRI study, total brain tissue volume in ASD was approximately 6% larger than in controls. **Megalocephaly** may be observable as young as 2 years of age, but was not present at birth in the cited observation. The teaching point is not that all autistic children have enlarged heads. It is that a developmental trajectory may differ after birth, and a cross-sectional image cannot be read as a universal sequence.

Postmortem studies describe increased neural packing, reduced cell size in the limbic system, reduced cerebellar Purkinje-cell number and altered cortical minicolumn organisation, as well as macroencephaly and altered brain-growth trajectories. Such work is constrained by small samples and tissue quality. It must be kept separate from in-vivo imaging. In a voxel-based meta-analysis, grey matter reduction was reported in the bilateral amygdala-hippocampus complex and bilateral precuneus, with a small increase in the middle-inferior frontal gyrus. These are group findings, not a radiological signature for clinical diagnosis.

21.5 Synaptic pruning as a mechanistic example

Synaptic pruning is a useful model for linking cellular findings with developmental circuitry. In the cited postmortem study, typically developing controls lost approximately 40% of dendritic spines across the age span, while individuals with ASD lost half as much as controls. The observation supports a hypothesis of less efficient pruning in at least some ASD tissue. It does not establish that deficient pruning is the sole pathway to ASD, nor that it is measurable in routine practice.

In the linked mechanistic work, **mTOR hyperactivity** was associated with impaired autophagy and disrupted synaptic pruning. In mice, rapamycin, an mTOR inhibitor, corrected pruning deficits and ASD-like behaviours. This is an important translational distinction: a correction in an animal model is not a recommendation for standard ASD treatment. The pathway is being examined in the tuberous-sclerosis subgroup, but the same source cautions that rapamycin is unlikely to become standard treatment for ASD. Mechanism-guided research is valuable precisely because it identifies testable subgroup hypotheses, not because it offers a universal cure.

MNEMONIC

Formulate, do not flatten

family pattern, genomic architecture, developmental brain evidence and exposure history belong in one formulation, but none should be promoted alone into “the cause.”

21.6 Environmental and perinatal associations: how to appraise them

Risk factor means an exposure or characteristic associated with a higher probability of outcome; it is not automatically a necessary or sufficient cause. Obstetric complications have some consistency across reviews, but associated odds ratios tend to be **below 3.0**. This magnitude and the possibility of confounding demand restraint. Complications may be on a causal pathway, may mark an earlier developmental difference, or may share determinants with ASD risk. The clinician should neither dismiss a careful obstetric history nor use it to assign parental blame.

Prenatal valproate exposure is a clinically important exception because it is recognised as a teratogenic exposure. ASD prevalence among those exposed prenatally is between 8-18 times that of the general population. ICD-11 also recognises prenatal valproate as an ASD risk factor. This is a risk statement about prenatal exposure, not permission to alter an individual’s medication without specialist review. In clinical practice, risk communication must balance maternal illness, treatment alternatives and reproductive planning.

Parental age and migration findings illustrate why context matters. Maternal birth abroad is associated with a risk increase of exactly 28% in the general estimate, while the association in immigrant mothers in Nordic countries is a risk increase of exactly 58%. These findings may reflect a complex mixture of migration-related exposures, access to care, social context and ascertainment; they must never be used to stigmatise migrant families.

- The greatest paternal-age risk is reported for fathers over the age of 50, compared with fathers below 30 years.

- For mothers over the age of 30 years, the estimated increased risk is exactly 27%.
- Evidence supports advanced parental age as a risk factor, with a possibly greater paternal contribution. Proposed pathways differ: maternal ageing is linked with chromosomal imbalance, while paternal ageing is linked with point mutation, imprinting, sperm-cell changes, de novo CNVs and epigenetic influences.

The professional response to these associations is prevention-oriented but non-punitive: obtain a sound exposure history, coordinate medication decisions before conception where possible, and avoid retrospective certainty. Most importantly, do not imply that ordinary parental conduct produced the child's ASD.

21.7 Vaccines, parenting and the duty to correct misinformation

This is a safety conversation, not an optional controversy. Claims that vaccines cause autism can lead to delayed or absent vaccination and can damage trust at the point when families need clear guidance. The evidence should be explained calmly, directly and without amplifying the myth through excessive repetition. The same principle applies to blaming parenting: it is inaccurate, harmful and clinically obstructive.

Natural experiment evidence is particularly useful when an exposure changes across a whole population. The Japanese cessation of MMR showed that MMR was not responsible for an epidemic of autism. Separately, Scandinavian cessation of the mercury preservative thimerosal gave the same negative finding. These are distinct questions and should not be blurred: MMR and thimerosal are not interchangeable exposures.

TRAP **Examination trap:** do not write "vaccines are controversial causes." The supported conclusion is that vaccines do not cause autism; withholding routine vaccination can harm the child.

IN INDIA

The Indian guideline, citing the CDC, rejects vaccine causation of autism and cautions that missing routine vaccination can harm the child. It also cites evidence that increasing exposure to vaccine antibody-stimulating proteins and polysaccharides was not associated with autism risk. These statements permit an unambiguous clinical recommendation while still making room for parents' fear to be heard respectfully.

When communicating a diagnosis, frame ASD as a lifelong neurodevelopmental disability with prenatal origins. Explain clearly that neither vaccination nor poor parenting produces it, and that children need not look alike clinically. This framing validates uncertainty without inventing a culprit. It also redirects attention toward assessment of strengths, support needs, associated medical conditions and intervention planning.

21.8 Clinical synthesis and examination answer

A complete aetiology answer begins with multifactorial neurodevelopmental causation and then separates evidence streams. Describe strong familial and twin evidence without treating heritability as individual causation. Explain the range from polygenic liability to rare CNVs,

chromosomal abnormalities and syndromic ASD, while referring genetic-test selection to the ASD medical workup. Use neurobiological observations as developmental correlates, state their limitations, and appraise environmental findings by study design, effect magnitude and confounding. End by explicitly correcting vaccine and parenting myths, because they are clinically consequential errors rather than harmless opinions.

For families, the formulation should be usable. It should say what is known, what remains uncertain, and what can be acted upon now. A syndrome-associated presentation may change surveillance and counselling; a prenatal exposure may affect future reproductive planning; neither changes the need for developmental support. Avoid giving a long list of alleged causes, which often leaves parents with blame rather than understanding.

ASD aetiology is best understood as heterogeneous neurodevelopmental causation: genes and developmental biology predominate, environmental associations require cautious causal inference, and vaccines and parenting do not cause autism.

The clinical picture of autism and how it is recognised

22 · Clinical features and core domains of ASD

The clinical task is to recognise a pattern, not to count isolated oddities. Autism spectrum disorder becomes visible when differences in social communication are understood alongside restricted, repetitive or inflexible patterns of behaviour and experience. A child may be quiet, highly verbal, bright, anxious, disruptive, apparently aloof, or warmly attached to familiar people. None of those impressions settles the question. The useful clinical formulation asks how the person shares attention, uses communication with another mind in view, manages reciprocity, and responds when routine, expectation or sensory input changes.

Recognition matters because the presentation is developmental and contextual. The same behaviour can have different meanings at different developmental levels, and a behaviour that looks unusual in a brief consultation may be an adaptive response to uncertainty. History, direct observation and reports from the settings in which the person lives should therefore be brought together. The diagnostic criteria are addressed separately; this section concerns the clinical phenotype that makes those criteria intelligible at the bedside.

approximately 20%	12-24 months	12-24 months	10-50%
REPORTED REGRESSION OR DEVELOPMENTAL STAGNATION	USUAL RECOGNITION WINDOW	USUAL WINDOW FOR SKILL LOSS, WHEN PRESENT	ESTIMATED RANGE FOR SAVANT OR SPLINTER SKILLS

22.1 Recognising a developmental social-communication difference

Autism is usually recognised through a qualitative difference in how social information is noticed, interpreted and used. The central issue is not whether the person wants company in an abstract sense. It is whether interaction is organised as a reciprocal exchange in which another person's

attention, knowledge, feeling and response alter one's own communication. A person may seek proximity, enjoy affection, or initiate contact for a need, yet still have difficulty sustaining the back-and-forth adjustments that make interaction feel shared.

Social reciprocity includes initiating interaction, responding contingently, sharing interests and affect, and allowing the partner's response to shape the next move. In young children this may be seen as limited showing, bringing, pointing out or checking back after an event. In older children and adults it may appear as a one-sided conversation, a tendency to return rapidly to a preferred topic, or difficulty recognising when the listener is confused, bored or trying to change direction. These are descriptions of the quality of exchange, not moral judgements about empathy, attachment or motivation.

A careful history separates capacity from performance. Some people communicate much less when overloaded, unfamiliar with the examiner, tired, frightened or expected to respond rapidly. Others have learned scripts that make initial interaction appear fluent but that do not readily flex when the conversation departs from the script. The clinician should look for developmental continuity and for the effort required to maintain apparently typical social behaviour. A socially successful encounter does not erase a pervasive developmental difficulty, just as an awkward encounter does not establish one.

RULE **Interpret the social behaviour in its communicative context: the question is how the person shares a mind with another person, not whether eye contact or sociability is simply present or absent.**

GOOD TO REMEMBER

At the bedside, ask the caregiver for ordinary examples of sharing an achievement, reacting to another person's distress, joining group play, managing a misunderstanding or telling a story to a listener who does not already know it. Then observe the response to an unexpected but benign shift in topic or activity: does the person repair communication, seek clarification, withdraw, perseverate or become distressed? This helps distinguish a stable developmental pattern from a situational reaction.

CLINICAL DOMAIN	WHAT TO EXPLORE	HOW IT MAY PRESENT IN CONSULTATION	INTERPRETIVE CAUTION
Reciprocal exchange	Initiation, response, turn-taking and sharing	Conversation may be asymmetrical, unusually passive, or driven by an immediate need	Shyness and anxiety can reduce performance without creating the developmental pattern
Non-verbal integration	Gesture, facial expression, gaze and timing	Signals may be limited, atypical, poorly coordinated with speech, or difficult to read	Do not convert one gaze style into a diagnosis
Relationships	Peer interest, social imagination and adaptation to roles	Rules of friendship may be understood literally; group interaction may be hard to enter or sustain	Solitude may be a preference, a coping strategy, or both
Repetition and inflexibility	Patterns, routines, focused interests and sensory responses	Distress, persistence or repetitive action becomes prominent when expectations shift	Determine function and impairment before labelling a preference pathological

22.2 Non-verbal communication and shared attention

Non-verbal communication is not a decorative adjunct to speech. Gesture, posture, facial expression, gaze, distance, timing and prosody carry much of the meaning of social exchange. In autism, the difficulty may lie in producing these signals, coordinating them with words, noticing those used by others, or inferring the unstated message they convey. The pattern is variable. Some individuals use abundant gesture but in an idiosyncratic or poorly integrated way; others have a relatively neutral facial style yet understand familiar relationships well.

Joint attention is especially useful clinically because it captures the difference between requesting and sharing. A person can point to obtain an object while finding it harder to point simply to make another person notice something interesting. Similarly, a child may follow a hand because it predicts access to a desired item but not because the other person's focus is meaningful in itself. The history should ask about following another's attention and actively directing it. Direct observation can then assess whether the person shifts attention between an object and a partner, checks the partner's reaction, or incorporates the partner into the experience.

Do not reduce this to an eye-contact test. Gaze is influenced by culture, anxiety, visual ability, temperament and the demands of the interaction. Sustained eye contact may be uncomfortable, distracting or consciously performed. Conversely, apparently good eye contact may coexist with marked difficulty using its social meaning. A better formulation is that visual social signals can be used differently, sometimes with little coordination between gaze, gesture and spoken language.

- Explore whether gestures are used to share as well as to request.
- Observe whether facial expression and voice match the intended message.
- Ask how misunderstandings are repaired when the listener does not follow.

- Consider whether social signals are missed, over-read or interpreted literally.

TRAP **Examination trap: neither reduced eye contact nor a preference for solitude is a sufficient clinical shortcut. The developmental pattern across reciprocal communication, non-verbal integration and flexibility is what carries meaning.**

22.3 Language phenomenology: form, use and reciprocity

Language in autism must be described at the level of use, not merely the presence or absence of words. The language phenomenology of autism includes differences in developmental progress, conversational reciprocity, prosody, literal interpretation and the ability to adjust communication for another person's perspective. Language ability is heterogeneous. Some people have very limited spoken language; others have extensive vocabulary and grammatically complex speech while finding the social use of language persistently difficult. Language impairment is considered separately as a specifier and should not be confused with the social-communication phenotype itself.

Pragmatic language concerns what speech does between people. It includes selecting relevant information, recognising implied meaning, taking turns, maintaining an appropriate topic, judging what the listener knows, and repairing breakdowns. A child may answer factual questions accurately but not volunteer information that would make the account coherent for someone else. An adult may speak fluently at length about a preferred interest yet have trouble sensing the reciprocal rhythm of conversation. These patterns are often most evident in unstructured conversation rather than in direct question-and-answer exchanges.

Echolalia refers to repetition of heard language. It may be immediate or delayed, but the clinician should not assume that it is meaningless. Repeated phrases can request, protest, regulate emotion, preserve a familiar sequence, or communicate an association that is not obvious to the listener. The clinical task is to determine the communicative function and the degree to which repetition substitutes for flexible, generative language. Similarly, pronouns may be used in an unusual perspective, and speech may have an atypical rhythm, volume or melody. Such features are useful only when understood as part of the wider developmental pattern.

History taking should locate the change in ordinary life. Ask how the person tells a story, joins a conversation already in progress, copes with jokes and indirect requests, or reacts when someone asks an ambiguous question. A family may report that the person is articulate but seems not to "read the room". That everyday observation can be more clinically revealing than a formal vocabulary performance. Equally, a bilingual environment should not be mistaken for a cause of autism or used to excuse a pervasive social-communication concern. The relevant question is whether the pattern is present across the languages and relationships available to the person.

For the intervention plan, speech and language therapy belongs with the broader support approach described in the relevant section of this chapter. At assessment, the immediate responsibility is more basic: document what communication achieves, what breaks down, and what support makes the person's intention understandable.

22.4 Restricted patterns, insistence on sameness and sensory experience

Restricted and repetitive behaviour is clinically broader than obvious motor stereotypy. It includes repetitive movements or speech, a marked need for predictability, unusually intense or narrowly focused interests, and atypical responses to sensory input. Its common thread is a reduced ease with variation or a distinctive way of regulating attention, arousal and meaning through repetition. The behaviour may be quietly internalised and therefore missed in an articulate adolescent or adult whose outward presentation appears composed.

Insistence on sameness is often best recognised through the consequences of disruption. The person may rely on a fixed route, sequence, object arrangement, conversational script or rule. A change may produce distress, anger, shutdown or prolonged preoccupation, not because the preference is trivial but because predictability is serving an organising function. Obtain concrete examples from home, school, workplace and travel. Then ask whether the routine can be modified with preparation, whether the response is proportionate, and whether the restriction limits participation or causes conflict.

Sensory reactivity may involve unusually strong, weak or seeking responses to sound, touch, light, smell, taste, movement or visual detail. It should be elicited as lived experience rather than treated as a box to tick. A child who covers their ears may be in pain, anticipating noise, avoiding a social demand, or doing more than one of these. An adult who avoids crowded places may be managing sensory overload, social uncertainty, panic, or a combination. The formulation must link trigger, response, function and consequence.

- Ask what the person repeatedly does, says, collects, watches or arranges.
- Ask what must happen in a particular order and what follows if it does not.
- Explore focused interests for intensity, flexibility and impact, rather than judging the topic itself.
- Map sensory triggers, protective strategies and the effect on ordinary participation.

Focused interests can be a route to competence, pleasure, identity and connection. They become clinically relevant when their intensity or inflexibility constrains functioning, blocks reciprocal engagement, or makes transition difficult. This distinction protects against pathologising expertise or temperament. It also enables the clinician to use an interest respectfully as a bridge during assessment rather than treating it only as a symptom.

MNEMONIC

Clinical spine - SHARE

Explore **S**ocial reciprocity, **H**ow communication is used, **A**daptation to change, **R**epetitive or focused patterns, and **E**xperience of sensory input. It is an enquiry sequence, not a diagnostic shortcut.

22.5 Developmental regression and developmental stagnation

Developmental regression means loss of abilities that were previously acquired and used, whereas developmental stagnation refers to a plateau in expected gains. Both deserve a careful timeline. A report may be imprecise because a skill was inconsistent, context-bound or only recently emerging. The clinician should therefore ask for examples of the earlier ability, its ordinary use, the sequence of change, and what else changed around the same period. In autism, regression or stagnation is reported in approximately **20%** of cases.

Autism symptoms are typically recognised between **12 and 24 months**. Where previously acquired social or language skills are lost, the loss itself also typically occurs between **12 and 24 months**. These statements describe distinct clinical questions: recognition concerns when the pattern comes to notice, while regression concerns the timing of actual skill loss. Keeping that distinction prevents a history of late recognition from being incorrectly recorded as regression.

Loss of previously acquired skills is usually described during the second year of life and is rarely observed after **3 years of age**. When it occurs, the loss most often concerns language use and social responsiveness. If the reported course is later or broader, document it precisely rather than forcing it into an autism narrative. If a loss occurs after 3 years of age, it is more likely to include changes in cognitive and adaptive functioning, continence, sleep, language, social ability, emotion and behaviour. Such a presentation needs separate evaluation; the differential diagnosis of regression is addressed elsewhere in this chapter.

Regression is emotionally potent for families. They often remember a before-and-after story, and it is important neither to dismiss that account nor to turn it prematurely into an aetiological explanation. Clarify whether the child lost a functional word, stopped responding to name, ceased sharing interest, became less socially responsive, or simply failed to add new skills. Review contextual changes and establish whether the apparent loss was global or selective. The record should preserve the caregiver's observations in concrete behavioural language, because that is more useful than the label alone.

> A plateau and a loss are not interchangeable: establish the prior skill, its functional use and the sequence of change before calling a course regressive.

22.6 Cognitive style, uneven ability and exceptional islands of skill

There is no single cognitive profile that defines autism. What is clinically common is an **uneven profile of abilities**, including in people whose general intellectual ability is average or high. A person may have striking factual knowledge, rapid pattern recognition or strong rote learning alongside difficulty applying ability flexibly in everyday social and practical contexts. The gap between demonstrated intellectual skill and adaptive functioning can be conspicuous. This is why a superficially impressive verbal or factual performance should not end the assessment of support needs.

Several cognitive accounts have helped clinicians describe aspects of the phenotype: **theory of mind**, weak central coherence, and executive dysfunction. Difficulty representing another

person's perspective can illuminate social misunderstandings. A detail-focused processing style can illuminate difficulty deriving the broader context. Problems with planning, shifting set, organising and inhibiting a habitual response can illuminate inflexibility. None should be used as a universal explanatory key. The cognitive theories do not establish a unitary cognitive origin for all manifestations of autism.

Savant or splinter skills describe competencies that stand out above the individual's general level of ability or above population expectation. Estimates vary with how such skills are defined, ranging between **10% and 50%** of people with autism. Their presence does not negate disability in another domain, and their absence has no diagnostic implication. Clinically, ask what the skill enables, whether it is enjoyable or burdensome, and whether it can be incorporated into education, work, self-advocacy or engagement.

Mental-state, contextual-integration, executive and motor features often discussed in autism are descriptive aids, not a substitute for developmental assessment. They can explain why an individual may know a rule yet fail to use it in a fast-moving social situation, or complete a complex task yet struggle with the unstructured demands around it. The practical lesson is to look for discrepancies: between knowledge and use, verbal expression and reciprocal conversation, performance in a preferred task and performance under change, or stated independence and actual day-to-day functioning.

22.7 A disciplined clinical formulation

A high-quality formulation is developmental, functional and contextual. It anchors current observations to the early pattern, describes what the person can do independently and what becomes difficult, and identifies environmental adjustments that alter performance. Home, educational, occupational and peer settings may reveal different aspects of the same difference. This framing matters when a person camouflages difficulties or when family routines have hidden impairment in consultation.

Use examples rather than labels wherever possible. "Talks at length about one topic and does not notice attempts to change it" is more clinically useful than "poor reciprocity" alone. "Becomes unable to continue when the usual route changes" is more useful than "rigid". Concrete descriptions allow later review, enable communication across disciplines and protect against diagnostic overshadowing. They also help the person and family identify accommodations without requiring the clinician to infer motives from a behaviour.

Changes from the person's usual baseline need particular care. Do not attribute new distress, marked withdrawal, loss of function, self-injury, agitation, sleep change or altered eating automatically to autism. Medical and psychiatric comorbidity, including catatonia, is considered in the medical comorbidities and workup section of this chapter. The clinician's immediate job is to recognise a baseline change and investigate it as a change, rather than accepting it as an inevitable expression of the developmental condition.

The assessment must also leave room for the person's strengths, preferences and account of their own experience. A formulation that records only deficits will miss the conditions in which

communication works, the interests through which rapport can be built, and the strategies that already reduce overload. The aim is not to make every distinctive preference disappear. It is to understand the pattern well enough to distinguish difference from impairment and to identify where support can make participation more possible.

23 · Severity levels and specifiers (with or without ID, language impairment)

Severity is a statement about current help, not a shorthand for ability. In autism spectrum disorder (ASD), a useful formulation has to keep several descriptions in view at once: the intensity of core autistic difficulties, intellectual functioning, language functioning, associated conditions and the person's changing circumstances. Collapsing these into one label makes the record look tidy but makes clinical planning less accurate. The diagnostic formulation should therefore preserve distinctions that matter to the person, family and team.

The immediate examination value is equally practical. A severity level describes the support needed for the core autistic presentation. It does not quantify intelligence, does not replace a language description, and does not exhaust the associated clinical picture. Specifiers make those separations visible. They turn a broad diagnostic name into a clinically communicable account of the individual sitting in front of the clinician.

3	2	3
DSM-5-TR SUPPORT LEVELS	DOMAINS RATED SEPARATELY	ICD-11 FUNCTIONAL-LANGUAGE LEVELS

23.1 What an ASD severity level is intended to convey

Start with the question behind the label. The DSM-5-TR severity framework is a description of **support need in the core autistic domains**. It asks how much assistance is currently required because of social-communication difficulty and because of restricted, repetitive patterns of behaviour. It is thus a functional clinical description, not a global ranking of the person.

There are **three** named support levels: **Level 1, requiring support**; **Level 2, requiring substantial support**; and **Level 3, requiring very substantial support**. The wording is deliberately about the assistance needed. A level should prompt the clinician to ask what supports permit participation, communication, flexibility and safety, rather than being treated as a fixed essence of the person.

RULE **Rule:** Record ASD severity as support need in the core domains, and record intellectual and language impairment separately.

That distinction matters because observed difficulty can be shaped by context. A familiar, predictable setting with effective communication supports may reveal capacities that are obscured in an unfamiliar assessment room. Conversely, a change in demands, sensory load, illness, distress or available support may expose need that was not apparent previously. The severity description must therefore be anchored in current functioning and meaningful participation, not in a superficial impression formed from a brief encounter.

The phrase “severity” can mislead candidates because the same word is used elsewhere in developmental psychiatry. Intellectual disability has its own severity classification, which is dealt with in the severity classification of intellectual disability section. The ASD framework answers a different question. It does not establish the presence of intellectual impairment and it does not substitute for an assessment of adaptive functioning. Keeping the questions separate prevents a common category error.

23.2 Rate the domains separately, not the person globally

DSM-5-TR asks that the level of support be recorded separately for **two** domains. One rating concerns social communication; the other concerns restricted, repetitive behaviour. This is not clerical fussiness. These domains can create quite different practical barriers, and their support requirements may not move together.

A person may need intensive help to manage inflexibility, transitions and repetitive behaviour while communicating more effectively in a structured interaction. Another may have relatively less conspicuous repetitive behaviour in a short consultation yet need extensive support to sustain reciprocal communication across daily settings. A single global level would conceal that pattern and can lead the receiving team to organise support around the wrong problem.

The social-communication rating asks how much support is needed for reciprocal communication. The restricted and repetitive behaviour rating asks how much support is needed for rigidity, repetitive behaviour and related interference. The intellectual-impairment specifier asks whether intellectual impairment accompanies ASD, while the language-impairment specifier asks whether language impairment accompanies it and, if so, what current verbal functioning is like. None of these questions can be answered by simply borrowing the answer to another.

GOOD TO REMEMBER

In a clinical note, record the two domain ratings with the observed barriers and the supports that alter them. A later change in level may reflect a change in support need, setting or the effect of effective intervention; it is not a prognosis.

TRAP **Classic trap:** Do not derive the ASD level from an IQ score. Intellectual impairment is carried by a separate specifier, not by the ASD severity level.

Equally, do not infer the support level merely from speech quantity. Language impairment and social-pragmatic difficulty overlap in everyday presentation but are not interchangeable descriptions. The language phenomenology of ASD belongs with the clinical features of ASD section; here the task is to show how language is captured in the diagnostic formulation without converting it into a core-severity score.

23.3 DSM-5-TR specifiers: add the clinically relevant dimensions

Specifiers prevent the broad ASD diagnosis from becoming clinically empty. They should be stated positively where present and negatively where absent, after assessment rather than by assumption. The first pair addresses accompanying intellectual and language impairment: **with**

or without accompanying intellectual impairment, and **with or without accompanying language impairment**.

The intellectual specifier is not an invitation to re-grade intellectual disability within the ASD note. It flags a co-occurring dimension that changes communication, assessment interpretation, educational planning and the kind of support likely to be required. The underlying assessment and severity classification of intellectual disability should be documented using their own standards. In particular, an autistic person's social difficulty should not be mechanically treated as evidence of global adaptive limitation.

The language specifier is similarly an addition, not a restatement of ASD itself. When language impairment is present, the clinician describes the current level of verbal functioning. This avoids the unhelpful shorthand of treating all language difficulty as equivalent. It also guards against the opposite error: assuming that fluent speech rules out significant social-communication disability. The diagnostic criteria section explains why language impairment is not itself a DSM-5 core criterion; the present specifier tells the reader whether it accompanies the syndrome.

Other specifiers place ASD in its wider clinical setting. The formulation may state that it is associated with a known genetic or other medical condition or environmental factor. This wording records an association or context; it must not be casually converted into a causal conclusion. It may also state that ASD is associated with a neurodevelopmental, mental, or behavioural problem. The full map of co-occurring conditions and how to detect them belongs in the medical comorbidities and workup in ASD section.

- Use the intellectual-impairment specifier to make accompanying intellectual impairment explicit, not to estimate it from presentation.
- Use the language-impairment specifier to record an accompanying language problem and current verbal functioning.
- Use the known-condition wording to state context accurately without claiming causation that the evidence has not established.
- Use the associated-problem wording to ensure that important co-occurring neurodevelopmental, mental or behavioural difficulties do not disappear behind the ASD label.

For the candidate, this is a disciplined form of diagnostic writing. "ASD" alone describes the syndrome but not the clinically relevant modifiers. A complete formulation makes visible which parts of the presentation are core, which are accompanying impairments, and which require parallel assessment or treatment. That distinction also protects against diagnostic overshadowing, where all distress or disability is wrongly attributed to autism.

23.4 Catatonia: a specifier that requires active clinical attention

Do not let a specifier become a footnote. DSM-5-TR permits the specification **with catatonia**. This should prompt careful assessment of a change from the individual's usual state, including

motor behaviour, responsiveness, activity and functional capacity. A diagnostic label should never replace a longitudinal account of what has altered and what the family or carers have observed.

When catatonia accompanies ASD, DSM-5-TR directs use of the additional code **F06.1**. It is a separate diagnostic code, not an ASD severity modifier and not a reason to raise a support level by default. The distinction is clinically important: support-level language describes core autistic support needs, whereas catatonia identifies an associated syndrome that requires its own recognition and management pathway.

The period of greatest risk described in DSM-5-TR is the adolescent years. That fact should sharpen clinical vigilance without turning ordinary developmental change into a diagnosis. When there is concern, document baseline, timing and trajectory, seek collateral from people who know the individual well, and evaluate the wider clinical context. Recognition belongs with the ASD comorbidity assessment; treatment is addressed in the pharmacotherapy in ASD section.

> A support level is not a catch-all label: ASD severity, accompanying intellectual impairment, accompanying language impairment and catatonia each answer different clinical questions.

23.5 ICD-11: characterisation through intellectual development, language and loss of skills

ICD-11 takes a differently structured approach. It does not supply DSM-style support-need severity levels. Instead, ASD is characterised through specifiers relating to **intellectual development, functional language and loss of previously acquired skills**. This is not a minor terminological difference. A clinician transferring information between systems must not invent a support level from an ICD-11 code, nor assume that an ICD-11 language category is a DSM support rating.

The intellectual-development axis is expressed as **with or without disorder of intellectual development**. In ICD-11, a co-occurring disorder of intellectual development is diagnosed separately rather than folded into the ASD diagnosis as an inline descriptor. When assessing this co-occurrence in ASD, greater weight should be given to the intellectual, conceptual and practical aspects of adaptive functioning than to social skills. This protects against mistaking social difficulties intrinsic to autism for evidence of a broader adaptive disorder.

The **functional-language axis** has **three** levels. It distinguishes mild or no functional-language impairment, impaired functional language, and complete or almost complete absence of functional language. The middle category concerns a person unable to use more than very short spoken expressions. These categories primarily describe functional expressive language, including nonverbal expression where relevant; they do not replace assessment of the pragmatic communication difficulty that is central to ASD.

FRAMEWORK	WHAT IT RECORDS	CLINICAL READING
DSM-5-TR ASD severity framework	Current support need in each core domain	Use to communicate the assistance required for core autistic difficulties
DSM-5-TR intellectual specifier	Presence or absence of accompanying intellectual impairment	Keep intellectual impairment distinct from ASD support level
DSM-5-TR language specifier	Presence or absence of accompanying language impairment	Describe current verbal functioning when impairment is present
ICD-11 ASD characterisation	Intellectual-development, functional-language and loss-of-skills specifiers	Do not translate this structure into a support-level rating

ICD-11 also records whether there has been loss of previously acquired skills. The suffixes 6A02.x0 and 6A02.x1 distinguish absence from presence of such loss. This coding distinction should not be used to teach the phenomenology or differential diagnosis of regression, which are considered elsewhere in the chapter. Its value here is to show that ICD-11 preserves a clinically salient developmental course feature within the diagnostic characterisation.

23.6 ICD-11 coding: use the matrix, do not guess it

The ICD-11 code reflects the combination of intellectual-development and functional-language descriptors. It is safer to determine the descriptors first, then select the matching code, rather than attempting to recall a code from an impression of severity. The following are selected, explicitly stated combinations. They are examples for accurate use, not a substitute for the full coding matrix.

DOCUMENTED COMBINATION	ICD-11 CODE	POINT TO RETAIN
Without disorder of intellectual development, with mild or no functional-language impairment	**6A02.0**	This is a coding combination, not a DSM support level.
Without disorder of intellectual development, with impaired functional language	6A02.2	Language description and intellectual-development status are both needed.
With disorder of intellectual development, with mild or no functional-language impairment	6A02.1	Co-occurring intellectual-development disorder is represented in the code.
With disorder of intellectual development, with impaired functional language	6A02.3	Do not infer support need from this code.
With disorder of intellectual development, with complete or almost complete absence of functional language	6A02.5	Use the language descriptor that is actually documented.

ICD-11 provides 6A02.Y for other specified ASD when the parameters do not apply, and 6A02.Z for unspecified ASD when the parameters are unknown. These are not interchangeable expressions of uncertainty. Good documentation reduces unnecessary use of the unspecified category while respecting genuine limits of available information.

23.7 A practical method for writing the formulation

Write the formulation in layers. First establish the diagnosis using the diagnostic criteria. Then describe the current ASD support need in each core domain when using DSM-5-TR. Next state the intellectual and language specifiers, followed by relevant associated-condition and associated-problem specifiers. Finally, record catatonia when present and use the appropriate additional code. This order mirrors clinical reasoning: syndrome, core impact, accompanying impairments, then associated context.

- State the diagnostic system being used so that the reader knows whether a support-level or coding-matrix framework applies.
- For DSM-5-TR, record the two core-domain support ratings separately and add a short functional explanation of each.
- State whether accompanying intellectual impairment and accompanying language impairment are present, without converting either into an ASD level.
- Record associated conditions and associated problems as clinical context, and keep association distinct from causal attribution.
- For ICD-11, establish intellectual-development status and functional-language status before selecting the code and any loss-of-skills suffix.

This layered method also improves communication across disciplines. It tells a speech and language clinician that a language issue is recognised without pretending that language alone explains the autistic presentation. It tells an educational or disability team that intellectual impairment requires its own assessment. It tells the medical team that an associated condition needs parallel attention. Above all, it avoids turning a diagnostic label into a description that is too vague to guide care.

When information is incomplete, state what has and has not been assessed rather than filling the gap with an assumed level. Diagnostic precision is not achieved by false certainty. A provisional formulation can still be useful if it clearly separates observed support needs from unassessed intellectual functioning, language status or associated conditions. The next assessment question then becomes obvious.

23.8 High-yield discriminations

The strongest answers distinguish neighbouring ideas before they list them. ASD severity is a support-need description for the core autistic domains. Intellectual impairment and language impairment are accompanying specifiers. ICD-11 uses a different characterisation system, based on intellectual development, functional language and loss of previously acquired skills rather

than DSM-style support levels. These are complementary ways of making heterogeneity visible, not rival measures of a single latent "severity".

In practice, the formulation should remain revisable. Support needs can change with environment, development, communication access, co-occurring clinical problems and the fit between the person and available services. Revision is not inconsistency when it reflects a better account of current functioning. What should remain stable is the discipline of recording each dimension for what it means, rather than allowing a memorable label to do the work of assessment.

24 · Screening and diagnostic tools (M-CHAT, ADOS, ADI-R, CARS, ISAA, INCLEN)

Tools organise evidence; they do not substitute for clinical judgment. Autism assessment is often difficult not because there is a shortage of instruments, but because instruments answer different questions. A brief parent questionnaire can identify a child who needs closer attention; an observed interaction can make social communication and restricted behaviour more visible; a developmental history can establish whether the pattern has been present across time; and a disability assessment can document functional impact for statutory purposes. Treating all of these as interchangeable produces familiar errors: diagnosing from a screen, or mistaking an administrative score for the diagnostic formulation.

For the candidate and the clinician, the useful organising question is therefore not, "Which scale proves autism?" It is, "What decision is this instrument designed to support?" A positive screen changes the urgency of assessment. A clinician-administered diagnostic measure can sharpen an already careful formulation. An Indian disability instrument serves a further, legally important function after diagnosis. This sequence protects children from delayed support while preserving the clinician's responsibility to weigh developmental history, direct observation, informant accounts, context, and alternatives.

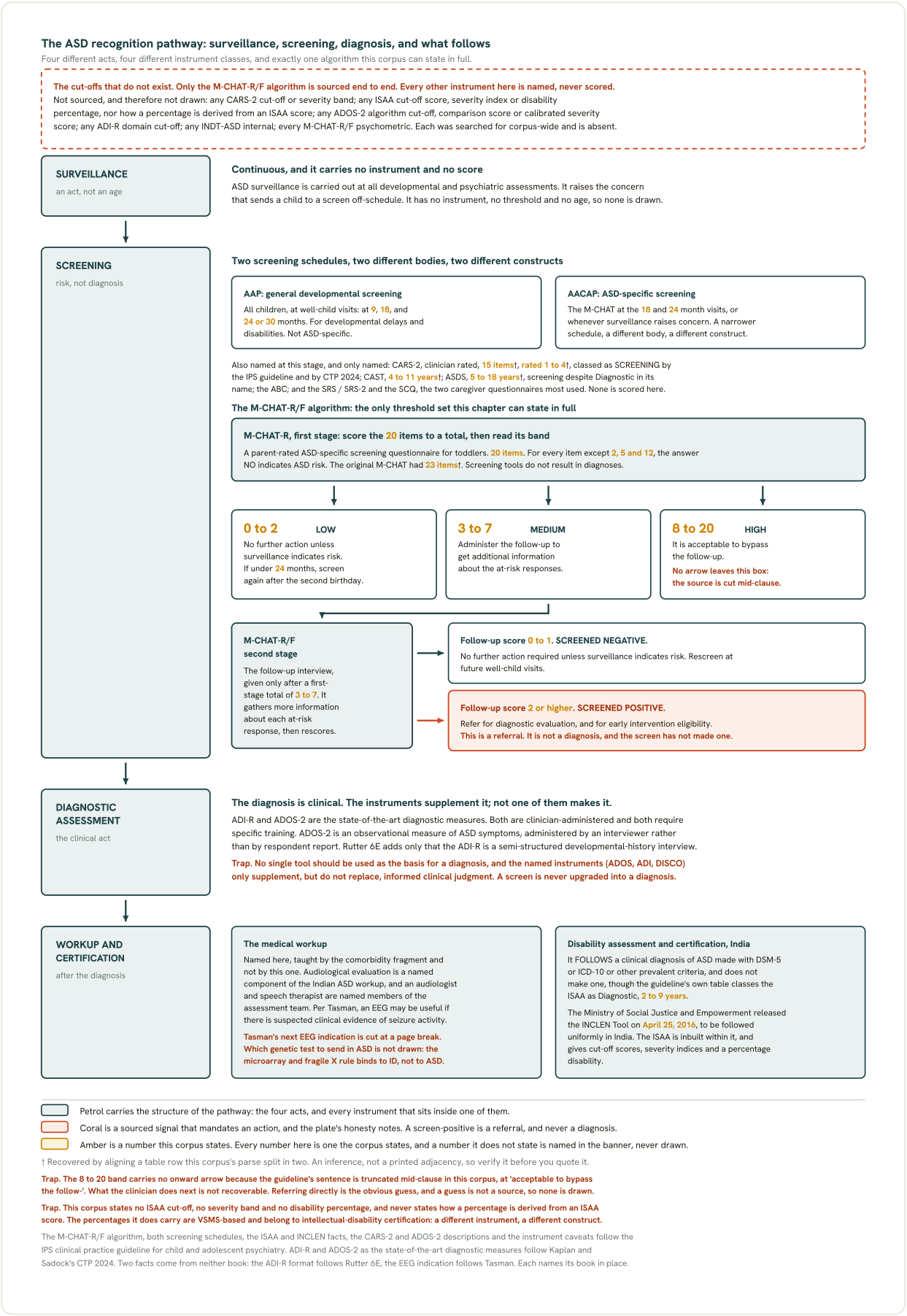


Fig 21.14 **The ASD recognition pathway.** Surveillance raises the concern, screening estimates the risk, the diagnosis is clinical, and both the workup and the Indian certification route follow it. Only the M-CHAT-R/F algorithm is sourced end to end; every CARS-2, ISAA, ADOS-2 and ADI-R cut-off is named on the plate and left undrawn.

24.1 A functional taxonomy of autism instruments

Screening is a process for finding children whose developmental profile warrants further assessment. Its purpose is sensitivity to concern, not a final statement about diagnosis. A screen can be negative when concerns remain clinically persuasive, and it can be positive because another developmental or behavioural difficulty needs assessment. Conversely, the absence of a score should never close a history that is concerning to parents or a clinician. **The M-CHAT-R/F** is a parent-rated, autism-specific toddler screen with a second-stage follow-up interview; it is not a diagnostic instrument.

RULE **Rule:** Match the instrument to its decision, then return the result to a clinical formulation rather than allowing the score to become the formulation.

CLINICAL QUESTION	USEFUL INSTRUMENT CLASS	WHAT THE RESULT SHOULD DO
Is there sufficient concern to escalate assessment?	Parent-rated autism-specific screen	Trigger follow-up questioning, surveillance, or referral according to the algorithm.
Can autism-related behaviour be sampled directly?	Observational assessment	Add direct clinical observations to the history and examination.
What was the developmental course?	Developmental-history interview	Clarify onset, change over time, and examples across settings.
Does a scale indicate concern or quantify observed features?	Clinician-rated screening scale	Support communication and follow-up, without becoming the diagnosis.
What is required for disability documentation in India?	Post-diagnostic disability assessment	Quantify disability and support access to certification-related entitlements.

The distinction is clinically productive. A questionnaire brings the caregiver's longitudinal experience into the consultation. An observation samples what happens under clinical conditions, but cannot by itself reproduce every home, school, or peer context. A history interview may expose a meaningful developmental trajectory, but its interpretation depends on the quality of the informant account and the clinician's questions. Each contributes a different kind of evidence. Diagnostic confidence comes from their convergence with informed judgment, not from collecting the largest possible pile of forms.

Informed clinical judgment remains central: diagnostic instruments supplement it and no single tool should provide the sole basis for diagnosis. This is not a vague appeal to intuition. It means the clinician must decide whether reported and observed features form a coherent autism pattern, whether they are developmentally meaningful, and whether another explanation better accounts for the presentation. The full assessment framework belongs with the Child psychiatry: assessment, treatment, services and protection notes; the present task is to understand what each autism-specific tool can and cannot add.

24.2 M-CHAT-R/F: using a screening algorithm safely

A screen is an invitation to think again. The M-CHAT-R/F is designed for toddlers and is parent rated. The revised checklist contains **20 items**. Its follow-up interview matters because an initial answer may reflect misunderstanding of the question, an unusual circumstance, or a concern that becomes clearer when a clinician asks for examples. In practical terms, the first stage identifies responses needing clarification; the follow-up turns a crude count into a more clinically usable account of risk responses.

0-2	3-7	2 or higher	8-20
LOW FIRST-STAGE BAND	FOLLOW-UP REQUIRED	POSITIVE AFTER FOLLOW-UP	FOLLOW-UP MAY BE BYPASSED

At a first-stage total of **0-2**, the result falls in the low-risk band. No further algorithmic action is required unless surveillance itself indicates risk. For a child younger than 24 months, with repeat screening after the second birthday, the guidance is to screen again. This is a useful reminder that a reassuring questionnaire is not permission to ignore a parent's developing concern, a developmental history, or a change in social engagement.

GOOD TO REMEMBER

A first-stage total of **3-7** is the medium-risk band and calls for the follow-up interview. Do not call this initial score the diagnostic threshold. It is a decision point for gathering better information about at-risk answers. After follow-up, a score of **at least 2** is screen positive and should lead to referral for diagnostic evaluation and consideration of early-intervention eligibility. A follow-up total of 0-1 is screen negative; ongoing surveillance and rescreening at future well-child visits remain appropriate when concerns arise.

At a first-stage total of 8-20, out of a possible **20**, it is acceptable to bypass the follow-up stage. The important interpretive point is that bypassing an interview is a rule within a screening algorithm, not a licence to pronounce a diagnosis from the questionnaire. In any referral, record the informant's examples, what was observed in clinic, and why the referral is being made. This makes the result clinically portable rather than a detached number.

Scoring direction is an easily missed technical trap. On most items, a response indicating risk is “no”; the exceptions are items 2, 5 and 12, where “yes” indicates risk. A clinician should not attempt to reconstruct the scoring from recollection while interpreting a parent form. Use the approved scoring algorithm, check that the correct revised version is being used, and document whether the score is before or after follow-up. That last distinction prevents a medium-risk first-stage score from being misreported as a positive screen.

TRAP **Examination trap:** A positive follow-up screen warrants diagnostic evaluation; it is not itself a diagnostic threshold.

Autism-specific screening is recommended at 18 and 24 months and whenever surveillance raises concern, alongside surveillance at developmental and psychiatric assessments. This differs from the schedule for general developmental screening, which is recommended at 9, 18, and 24 or 30

months during regular well-child visits. The distinction matters because a general screen and an autism-specific screen answer overlapping but not identical questions. The normal developmental context and milestones that make a particular response concerning are developed in the Normal child development and milestones notes.

24.3 Diagnostic instruments: observation, history, and trained use

Diagnostic measures are not simply longer screening scales. The ADI-R and ADOS-2 are described as state-of-the-art diagnostic measures and require specific training for clinician administration. Training is not ceremonial. It is needed to standardise how prompts are given, how behaviours are recognised and coded, and how the resulting information is used without overriding the clinical context. An untrained, improvised administration can create an appearance of precision while reducing the value of the observation.

ADOS-2 is an interviewer-administered observational measure of autism-spectrum symptoms. Its contribution is direct sampling: the assessor sees how communication, reciprocity, play, flexibility, and other relevant behaviours emerge in interaction. That is valuable when caregiver descriptions and clinic impressions diverge, when the child presents differently across contexts, or when the clinician needs well-described behavioural examples for a multidisciplinary discussion. It remains a sample, however. A calm consultation, a familiar caregiver, fatigue, anxiety, unfamiliar language, or the child's strategy for coping with novelty can all influence what is observed.

ADI-R is a semi-structured instrument for obtaining a developmental history. It is particularly useful conceptually because autism is not diagnosed from a single cross-sectional interaction. The interviewer asks the caregiver to move from labels such as “he does not mix” to concrete examples: what the child did, in what situation, whether the pattern was sustained, and how it changed. A good developmental history distinguishes a stable developmental style from a recent change and makes it possible to test whether apparently isolated symptoms belong together. This also protects against treating a questionnaire response as though it were already a clinical observation.

These instruments should be read as complementary rather than competitive. One asks what behaviour can be observed now; the other helps establish developmental meaning through a knowledgeable informant. Neither removes the need to consider communication ability, sensory or physical factors, environmental demands, and alternative developmental formulations. Nor should their absence be used to deny care where an experienced clinician can make a sound diagnosis. Their special value lies in disciplined, reproducible evidence when the assessment is complex, contested, or requires detailed communication across services.

MNEMONIC

H-O-L-D the hierarchy

History, **O**bservation, **L**inkage across contexts, then **D**iagnostic judgment. It is a sequence for thinking, not a substitute scoring rule.

24.4 Rating scales and caregiver questionnaires: useful, but bounded

Rating scales create a compact record of behaviours and can support screening, communication, and longitudinal review. Their apparent simplicity is also their risk. A numerical total can look more authoritative than a detailed narrative, yet a rating depends on who observed the behaviour, what comparison they used, and what setting was captured. The score should lead back to examples. If a scale and the clinical account disagree, investigate the discrepancy rather than automatically privileging either source.

INSTRUMENT	CLASSIFICATION AND RESPONDENT	STRUCTURE STATED IN THE GUIDELINE
CARS-2	Screening, clinician rated	15 items rated 1-4
CAST	Screening, parent rated, ages 4-11 years	37 yes/no items
ASDS	Screening, parent rated, ages 5-18 years	50 yes/no items
Autism Behavior Checklist	Screening, parent rated	57 items rated 1-4

CARS-2 is classified as a screening measure, even though it is clinician rated. That distinction is worth making explicitly because clinician administration alone does not convert a scale into a diagnostic instrument. Likewise, the word “Diagnostic” in the Asperger Syndrome Diagnostic Scale can mislead: the guideline classifies it as screening. Candidates should classify an instrument by its validated clinical role, not by the confidence suggested by its name.

Caregiver questionnaires are especially helpful when clinic behaviour is unrepresentative of home or school. The commonly used instruments include the Social Responsiveness Scale, including SRS-2, and the Social Communication Questionnaire. They can focus the interview and identify domains requiring examples. They do not turn the caregiver into the diagnostician. The clinician's task is to determine the meaning of an endorsed difficulty, its developmental context, its consistency across settings, and the effect of communication level or situational stress.

24.5 ISAA, INCLEN, and the Indian certification pathway

Diagnosis and certification answer different questions. The **Indian Scale for Assessment of Autism** is listed as a diagnostic instrument for children aged 2-9 years. Its stated format is a parent interview with 40 items rated 1-5. It produces cutoff scores, severity indices, and percentage-disability output that can assist certification. These capabilities make it important in Indian practice, but they should not be allowed to collapse the distinction between a clinical diagnosis and the later task of documenting disability for statutory purposes.

The ISAA is inbuilt within the INCLEN Tool; this is a containment relationship, not a claim that the two names are interchangeable. The **INCLEN Diagnostic Tool for Autism Spectrum Disorder** was released for uniform autism assessment in India by the Ministry of Social Justice and Empowerment, Department of Empowerment of Persons with Disabilities on April 25, 2016. In an examination answer, this is best presented as an Indian assessment and certification framework rather than as an alternative to clinical reasoning.

IN INDIA

Clinical diagnosis precedes disability assessment. After an established clinical diagnosis using recognised diagnostic criteria, the INCLEN tools and ISAA are used for disability assessment. It is therefore incorrect to write that a child "has autism because the ISAA is positive." A scale result may support certification-related documentation, but the diagnosis must already rest on a clinical assessment. Conversely, an appropriate clinical diagnosis should not be withheld simply because certification has not yet been completed.

Certification quantifies disability and can facilitate scheme benefits and insurance eligibility. It therefore has material consequences for a family, and careful documentation is part of good clinical care. The required board comprises a paediatrician or paediatric neurologist, a clinical or rehabilitation psychologist, and a psychiatrist. This multidisciplinary composition reinforces the point that administrative assessment should be clinically anchored rather than reduced to a form-filling exercise.

Certificate validity depends on the nature of the disability. For an individual under 18 years of age with temporary disability, validity is 5 years; where a permanent disability has been acquired, the certificate has permanent validity. When discussing this with families, distinguish clearly between the diagnosis, the disability assessment, and the certificate. They are connected steps but not synonymous labels. The statutory context is the Rights of Persons with Disability Act, and the instrument's output should be explained as serving this practical pathway.

TRAP **Examination trap:** Do not use an ISAA or INCLEN disability assessment as though it replaces the clinical diagnosis of autism.

24.6 Integrating results in the consultation

A robust assessment is transparent about the status of each piece of evidence. State whether a result came from a parent screen, a caregiver questionnaire, clinician rating, direct observation, developmental-history interview, or disability assessment. Then state what it adds and what remains uncertain. This simple discipline prevents a referring clinician from reading a "positive" score as definitive and prevents a receiving clinician from discarding valuable information because it is not itself diagnostic.

- When a screen is positive, explain that it identifies a need for thorough diagnostic assessment and does not apply a diagnostic label.
- When a score is negative but surveillance remains concerning, return to the history, settings, and direct observation rather than treating the score as a veto.
- When trained diagnostic measures are used, document that their findings were integrated with clinical judgment rather than reported as a free-standing conclusion.
- When disability documentation is needed, establish the clinical diagnosis first and then proceed through the appropriate Indian assessment pathway.

The practical consequence is a more humane and accurate conversation with caregivers. Rather than saying “the test says autism” or “the test is normal,” the clinician can say what was observed, what the caregiver described, what the instrument contributes, and why further assessment or follow-up is appropriate. This approach also makes room for uncertainty without inaction. Where developmental concerns are real, referral and support planning need not wait for a family to understand every technical distinction among questionnaires, interviews, observation schedules, and certificates.

> A positive autism screen changes the next clinical action; it does not, by itself, make the diagnosis.

25 · Medical comorbidities and workup in ASD (epilepsy, sleep, GI, genetics)

Autism does not protect a person from ordinary illness, and it can make illness harder to detect. A change in behaviour, communication, participation or self-care may be the presenting language of pain, sleep disruption, seizures, anxiety or another co-occurring condition. The assessment therefore has two linked tasks: recognise the developmental autism phenotype and ask what else may be contributing to current impairment. This is especially important when the person has limited spoken language, difficulty describing internal states, or a history in which distress has repeatedly been attributed to autism itself.

Co-occurring conditions are not a miscellaneous appendix to diagnosis. They alter risk, family burden, educational participation and the interpretation of behaviour. A useful formulation begins with the person's usual baseline and asks what has changed, in which setting, with what bodily or emotional correlates, and what reliably relieves or worsens it. The clinician should bring together caregiver observations, the person's own account where possible, developmental history, medication exposure, sleep and elimination history, dietary pattern, and a focused physical and neurological examination. The aim is not indiscriminate testing. It is a hypothesis-led medical workup that can explain a new problem and identify needs that autism alone cannot explain.

10-15%	up to 53%	7-24%	more than 50%
ESTIMATED EPILEPSY IN CHILDREN WITH ASD	WITH A FREQUENT SLEEP PROBLEM	WITH SIGNIFICANT CHRONIC GASTROINTESTINAL COMPLAINTS	ESTIMATED TO MEET ADHD CRITERIA IN SOME SAMPLES

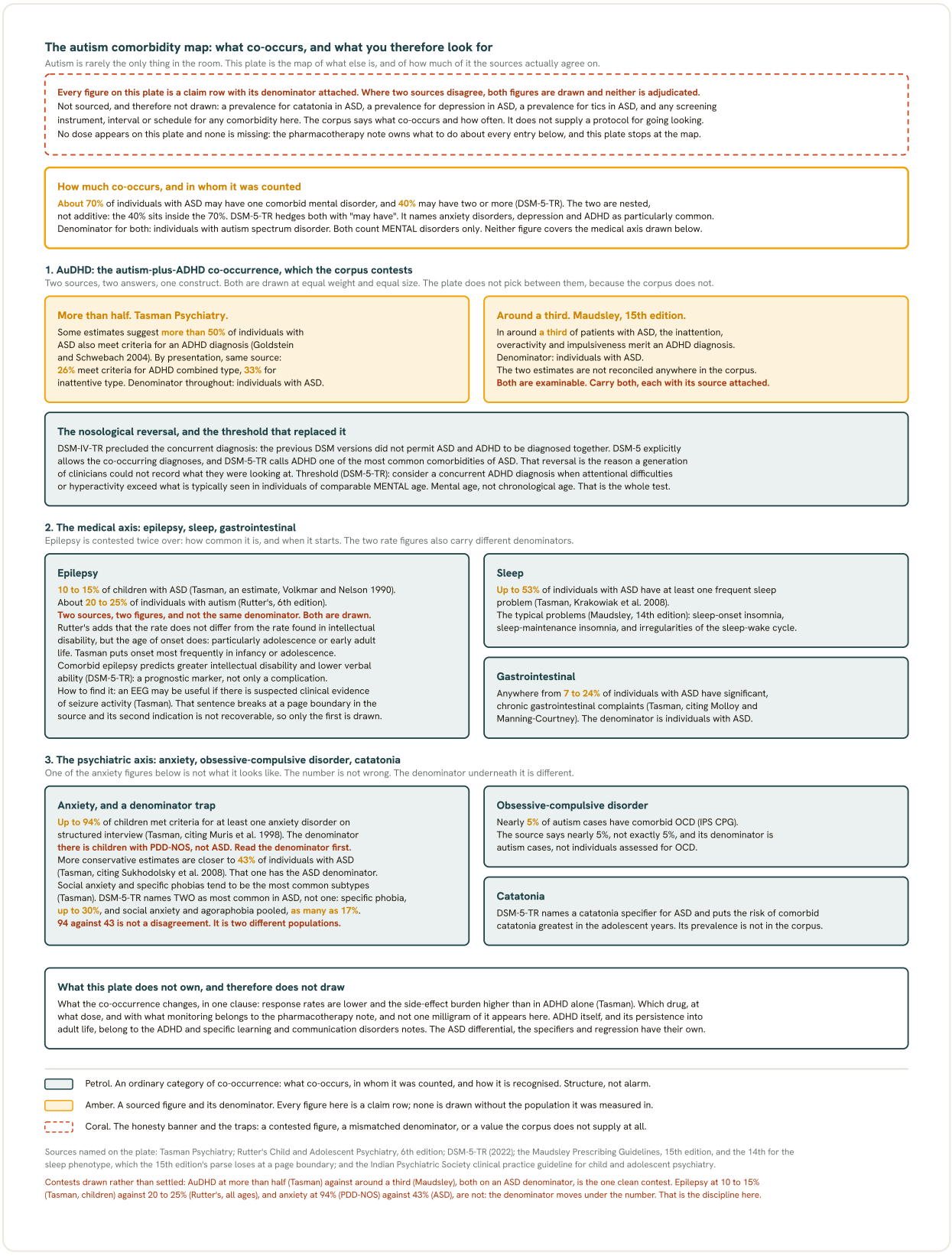


Fig 21.15 **The autism comorbidity map.** What co-occurs with autism, medical and psychiatric, with every figure carried alongside the population it was counted in. AuDHD leads the plate because the sources contest it: more than half of individuals with ASD (Tasman) against around a third (Maudsley), drawn side by side at equal weight rather than reconciled, with the DSM-IV exclusion that DSM-5 reversed and the comparable-mental-age threshold that replaced it. Epilepsy is contested in rate and in age of onset, though its two rate figures also differ in denominator, children against all ages. Anxiety at 94% against 43% only looks contested: the denominators are PDD-NOS and ASD respectively, and telling the two cases apart is the point. Prevalences for catatonia, depression and tics in ASD are not in the sources, so none is drawn.

25.1 A clinical method for finding comorbidity

Start with the referral question, but do not let it narrow the enquiry prematurely. A child described as "more autistic" may in fact be sleeping poorly, constipated, frightened, in pain, having unrecognised seizures, depressed, overwhelmed by a new demand, or experiencing an adverse effect of treatment. An adult who has always required support may develop a new loss of function that deserves the same respect as new loss of function in any other patient. Baseline is therefore a clinical instrument: establish usual communication, mobility, eating, sleep, continence, interests, activity level, self-injury, emotional expression and ability to tolerate ordinary transitions before interpreting change.

Behavioural equivalent is a practical term for a bodily or emotional problem expressed through observable behaviour when direct reporting is limited. It should prompt enquiry, not provide a conclusion. Agitation may reflect discomfort, fear, overstimulation, an interpersonal conflict, a seizure-related event or an emerging psychiatric syndrome. Similarly, apparent non-compliance can be a communication about an inaccessible demand or an aversive physical state. Ask caregivers for a sequence rather than a global adjective: what happens first, what follows, how long it lasts, whether awareness changes, and what restores the person to baseline.

CLINICAL SIGNAL	QUESTIONS THAT SHARPEN THE HISTORY	WHY IT MATTERS	IMMEDIATE ROUTE
New episodic change	Was there altered awareness, a motor event, a fall, a period of confusion, or a return to usual functioning?	Distinguishes a possible neurological event from a behavioural description alone	Document the event carefully and consider neurological assessment
New irritability or self-injury	What has changed in sleep, bowels, eating, activity, medication, demands or sensory load?	Physical discomfort and environmental strain are common explanatory possibilities	Examine, identify urgency, and investigate the leading hypothesis
Functional decline	Which specific skill has changed, how abrupt was the change, and is it present across settings?	Prevents a serious change being absorbed into a static developmental label	Assess medical, neurological and psychiatric contributors in parallel
Internalising distress	What situations are avoided, feared, anticipated or endured with marked distress?	Anxiety may be obscured by shutdown, rigidity, anger or somatic complaint	Make a symptom formulation before assigning a label

RULE **Any marked change from baseline deserves a fresh medical and psychiatric formulation; do not accept autism as its explanation until competing explanations have been considered.**

25.2 Epilepsy: recognise events, assess risk and use EEG selectively

Epilepsy is an important ASD comorbidity, but its clinical relevance lies in more than its frequency. Estimates in children with ASD place comorbid epilepsy between **10% and 15%**.

Another estimate for individuals with autism is about **20% to 25%**. These figures should not be treated as a prediction for an individual patient. They do, however, justify a low threshold for taking a paroxysmal history seriously, particularly when family descriptions include abrupt episodes that differ clearly from the person's usual repetitive behaviour, sensory response or emotional dysregulation.

The age pattern has clinical value. Onset is reported most often in infancy or adolescence, while onset in adolescence or early adult life is particularly distinctive when autism is compared with intellectual disability. The overall epilepsy rate in autism does not differ from that found in intellectual disability; the timing of onset is the more useful discriminating observation. A routine visit should therefore include a longitudinal question about events across development, rather than only asking whether seizures are occurring now.

Paroxysmal event is preferable to the word seizure until the history supports a conclusion. Ask whether the event occurs from sleep, during illness, after a particular trigger, or in a stereotyped sequence. Repetitive movements, staring, withdrawal, panic, dissociation and behavioural shutdown can all be misread if the sequence is not reconstructed.

Explain to families who expect a single investigation to explain autism or every behavioural difficulty that the question for the test is narrow: whether the event pattern raises a neurological concern that the EEG can help clarify. When events are recurrent or the account is unclear, contemporaneous documentation and neurological collaboration are often more informative than retrospective labels.

GOOD TO REMEMBER

When a new event is reported, reconstruct it with a witness before naming it a seizure: onset, behaviour, awareness, responsiveness, colour change, injury, duration, recovery and return to baseline. A caregiver-recorded video can improve phenomenology. An EEG may be useful when there is clinical evidence suggesting seizure activity; it does not replace a careful event history and must be interpreted in its clinical context.

Comorbid epilepsy is associated with greater intellectual disability and lower verbal ability. This association should guide anticipatory support, not deterministic counselling. It signals that communication access, safety planning and caregiver education may need particular attention. It does not mean that every person with epilepsy will follow the same developmental course, nor does it license therapeutic pessimism. Seizure management itself should be coordinated with the relevant neurological service; syndrome-specific seizure teaching belongs to the relevant syndrome sections rather than to a general ASD workup.

TRAP **Examination trap: do not request an EEG merely because a person has ASD. Request it for a clinically credible paroxysmal history, and document the event phenomenology that created that suspicion.**

25.3 Sleep disturbance: make the phenotype visible before treating it

Sleep problems are common enough to merit routine enquiry rather than waiting for a complaint. Up to **53%** of individuals with ASD have at least one frequent sleep problem. The common clinical patterns are **sleep-onset insomnia**, **sleep-maintenance insomnia** and **sleep-wake cycle irregularity**. Naming the pattern matters because a family report of "not sleeping" may refer to a prolonged settling period, repeated waking, a shifted schedule, an early waking pattern, or a combination.

Ask for the sequence around sleep rather than an estimate of total sleep alone. Establish the evening routine, time of attempted sleep, latency, night waking, nocturnal activities, waking time, naps, weekend variation, bedroom conditions, caregiver responses, daytime sleepiness and change from baseline. Ask directly about pain, bowel discomfort, breathing concerns, seizures, itching, medication timing, caffeine exposure, activity, screen use and anxiety. This identifies reversible drivers and clarifies whether the intervention must change the environment, the daytime rhythm, an associated condition, caregiver contingency, or a biological sleep signal.

Melatonin has shown benefit in **17 studies** in children with ASD. The dosing figures come from a narrower base and should be quoted as such: a meta-analysis of five of those studies is what reports good efficacy across doses of **1 and 10 mg**.

IN INDIA

For sleep disturbance in ASD, Indian guidance states a melatonin range between 1 and 10 mg at night.

Treatment periods in the cited evidence ranged from 14 days to over 4 years, which shows that use has not been confined to a single brief interval, not that every patient should receive long-term treatment.

Where epilepsy is also present, the guidance describes melatonin as carrying a low risk of seizure precipitation. This is reassuring but does not remove the need to document the seizure history, current neurological treatment and temporal relation between any new symptom and a medication change. The broader pharmacological management of target symptoms is addressed separately. Here the principle is to treat sleep as a clinical syndrome with contributors, not as an inevitable feature of autism.

For sleep, identify the pattern and its maintaining conditions before adding a treatment. A precise sleep history often prevents both under-treatment of discomfort and over-treatment of behaviour.

25.4 Gastrointestinal symptoms and bodily distress

Gastrointestinal complaint may present as a direct report, altered food intake, agitation around meals, a new sleep problem, avoidance of the toilet, or self-injury. Significant chronic gastrointestinal complaints are reported in between **7% and 24%** of individuals with ASD. It

should encourage active enquiry, not a presumption that every distress behaviour is gastrointestinal.

The clinical history should make eating and elimination discussable without embarrassment. Ask about appetite, food selectivity, texture or smell sensitivity, pain behaviour, swallowing concerns, nausea, vomiting, stool pattern, toileting avoidance, soiling, bleeding, weight change and the relation of symptoms to medication or routine disruption. In a person who cannot describe pain conventionally, a caregiver's recognition of a distinct grimace, posture, guarding, meal-related distress or night waking may be the most useful starting point. Examination and referral should be guided by the symptom pattern and urgency, as in any patient.

It is useful to separate a sensory preference from a medical symptom without making them adversaries. A narrow diet may be a stable sensory accommodation, a consequence of pain, an anxiety-driven avoidance, a learned family adaptation, or several of these together. The same is true of toileting refusal. The formulation becomes clinically useful only when it identifies function, discomfort, nutritional impact and the practical barriers to change. Abrupt dietary restriction, apparent pain, dehydration, bleeding, persistent vomiting or a significant decline in functioning should not be normalised as autism.

- Ask directly about bowel, bladder, appetite and pain at each meaningful change in behaviour.
- Use familiar caregiver observations as data, then test them against a focused history and examination.
- Distinguish a long-standing preference from a new restriction or a new distress signal.

25.5 Genetic and general medical workup: phenotype-led, not reflexive

Phenotype-led genetic evaluation means that genetic assessment is anchored to the individual rather than to a generic autism label. The history should include developmental course, medical events, neurological symptoms, family history and any pattern suggesting a broader developmental condition. The examination should attend to growth, physical features, skin, neurological signs and other findings that may make a syndromic explanation more likely. This approach improves the quality of the referral question and helps the family understand why genetic assessment is being considered.

Genetic testing is part of the ASD medical workup, but its value is not exhausted by producing a laboratory result. A result may refine an aetiological formulation, prompt surveillance for associated medical issues, inform reproductive discussion through an appropriate genetics service, and connect a family with relevant supports. Conversely, an uninformative result does not invalidate the person's autism diagnosis or their need for support. Consent should cover what the investigation can and cannot answer, the possibility of uncertain findings, and the implications of receiving information that concerns relatives.

The psychiatrist's role is to recognise when a broad medical formulation is needed, obtain a careful developmental and family history, coordinate referrals and translate results into a person-

centred plan. It is not to assign a syndrome from a single physical feature or to promise that testing will identify a cause in every case.

- Record the phenotype and the family question that prompted referral.
- Explain what a result may clarify and what it may leave uncertain.
- Coordinate interpretation with genetics and other relevant services.

Hearing and vision concerns, nutritional status, sleep, bowel function, medication effects and neurological symptoms should be considered alongside genetic assessment. This prevents the common error of pursuing an etiological explanation while missing a treatable contributor to day-to-day distress. A medical workup is successful when it makes the clinical picture more intelligible and improves care coordination, even when no single cause is established.

25.6 AuDHD: distinguish co-occurrence from diagnostic substitution

AuDHD refers to the co-occurrence of autism and ADHD. ADHD is one of the most common comorbidities in ASD, and some estimates suggest that **more than 50%** of individuals with ASD also meet criteria for an ADHD diagnosis. The clinical task is not to decide which label "explains everything". It is to establish whether attentional difficulties, impulsivity or hyperactivity represent an additional, impairing syndrome rather than an understandable consequence of overload, anxiety, poor sleep, learning demands or the social environment.

This is a major nosological correction for candidates to understand. DSM-IV-TR did not permit concurrent ASD and ADHD diagnoses; DSM-5 permits co-occurring diagnoses. The earlier exclusion still shapes clinical habits. In one direction, marked activity or distractibility is dismissed as part of autism. In the other, social difficulty is assumed to follow from ADHD alone. Both shortcuts can delay a formulation that makes sense of the person's whole pattern.

Assess attention and activity across settings and against the person's developmental level. A concurrent ADHD diagnosis should be considered when attentional difficulties or hyperactivity exceed what is typical for individuals of comparable mental age. Ask what happens in a low-demand preferred activity, in an unstructured group setting, during a task that is understood but not preferred, and after a poor night of sleep. Gather collateral from the settings where demands differ. This separates sustained regulation difficulty from a problem that appears only under a particular sensory, social or academic load.

The co-occurrence changes management planning because sleep, anxiety, communication demands and environmental predictability can all alter attention and activity. Establish the target impairment, its baseline, the family's priorities and the measures by which benefit and adverse effects would be judged. Pharmacological treatment selection for target symptoms is addressed in the pharmacotherapy section; ADHD itself, including adult persistence, is addressed in the ADHD and specific learning/communication disorders notes. The key point here is that dual diagnosis is clinically permissible and often clinically clarifying.

MNEMONIC

Clinical spine - CHANGE

When behaviour worsens, check **C**ourse from baseline, **H**ealth and pain, **A**rousal and sleep, **N**eurological events, **G**astrointestinal and other bodily symptoms, and **E**motional or attentional comorbidity. It is a sequence for broadening the formulation, not a substitute for assessment.

25.7 Anxiety, obsessive-compulsive phenomena and catatonia

Anxiety disorder is common in ASD, with a more conservative estimate of **43%** in individuals with ASD. Social anxiety and specific phobias tend to be the most common subtypes. The assessment must not mistake all avoidance for autism-related social difference or all insistence on sameness for anxiety. Ask what the person fears will happen, what they anticipate, whether distress appears before the event, what safety behaviours are used, and whether the pattern is new, pervasive or situation-bound.

Obsessive-compulsive symptoms demand a similarly functional distinction. Repetitive acts in autism may be enjoyable, regulating or linked to predictability, whereas compulsions are more often driven by distress and an attempt to reduce a feared outcome. The distinction can blur, particularly when the person cannot easily report intrusive thoughts. Explore the emotional state before and after the act, the consequences of interruption, the degree of choice, and whether the behaviour has acquired a feared meaning. Nearly 5% of autism cases have comorbid OCD, so the possibility should be actively considered rather than hidden within a description of repetitive behaviour.

Catatonia requires recognition because new marked slowing, reduced initiation, mutism, posturing, agitation, loss of previously established function or other conspicuous motor-behavioural change may be misattributed to autism. The core safeguard is temporal: a substantial change from the person's own baseline is a clinical event. Obtain a careful timeline, assess medical and neurological contributors, seek collateral, and involve appropriate specialist services where concern is significant. Do not wait for a person to fit a stereotyped image of catatonia before taking severe change seriously.

TRAP **Examination trap: repetitive behaviour is not automatically OCD, and withdrawal is not automatically autism. Establish the preceding emotion, perceived purpose, degree of distress and change from baseline.**

25.8 Integrating findings into a safe, useful formulation

The completed assessment should answer practical questions: what is longstanding, what is new, what is impairing, what needs urgent medical attention, and which professional needs to act next? Document observations in behavioural language, preserve uncertainty where it remains, and specify the evidence supporting each concern. A formulation such as "new nocturnal waking with apparent abdominal discomfort and daytime irritability" is more actionable than "behavioural problem in ASD". It allows the family, psychiatrist, paediatric or medical colleague, school and therapist to work from the same clinical picture.

Finally, explain the formulation to the person and family without implying blame. Families often arrive after being told either that every problem is autism or that autism explains nothing. Both positions are unhelpful. Autism provides a developmental context. It does not erase the need to recognise pain, epilepsy, sleep difficulty, gastrointestinal symptoms, genetic questions, ADHD, anxiety, OCD or catatonia. Care improves when the clinician can hold both truths at once: the person's baseline neurodevelopmental profile matters, and a real change from that baseline always deserves to be understood.

Managing autism and its outcome

26 · Differential diagnosis of ASD

Differential diagnosis is a longitudinal task. Autism spectrum disorder (ASD) is not established by an isolated socially atypical behaviour, a language delay, or a repetitive act observed in clinic. The clinician asks whether the social-communication pattern is pervasive, whether restricted or repetitive behaviour is present across development, what the child could do before the concern emerged, and whether the proposed alternative explains the whole presentation more parsimoniously. The same exercise determines whether an apparent alternative is instead a co-occurring condition that deserves its own diagnosis and treatment plan.

For examination answers, organise the differential around the **developmental trajectory**: an early and pervasive neurodevelopmental pattern supports ASD; a setting-bound difficulty, a later change after relatively typical development, or a symptom driven by fear, psychosis, sensory loss, neurological illness, or adverse care conditions should redirect assessment. Collateral developmental history, direct observation across settings, and attention to communication without spoken language are often more informative than a checklist answer. The task is not to make every unusual behaviour autistic, nor to deny ASD when another condition is also present.

RULE **Core rule.** Diagnose ASD when its characteristic social-communication and restricted or repetitive pattern is the best account of the developmental presentation; add another diagnosis when that condition independently meets its threshold or requires separate clinical attention.

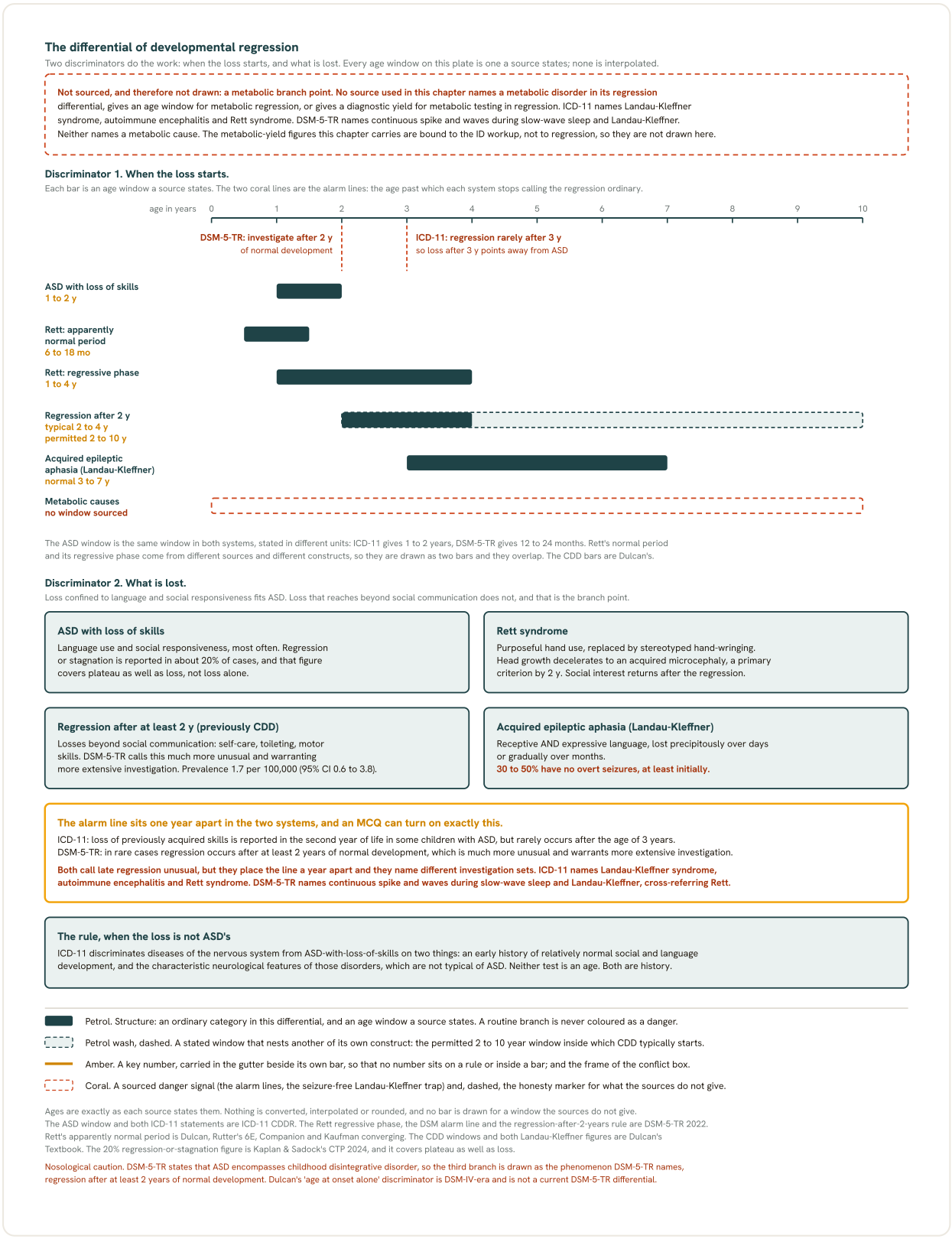


Fig 21.16 The differential of developmental regression. The branch points that separate autism with loss of skills, Rett syndrome, regression after at least two years (previously childhood disintegrative disorder) and acquired epileptic aphasia, drawn on the two discriminators that actually do the work: the age at which the loss starts, and what is lost. The ruler carries only age windows a source states, and the alarm lines show ICD-11 and DSM-5-TR placing the boundary of the ordinary a full year apart. A metabolic branch point is named on the banner and left undrawn: no source used here supplies one, and the metabolic yields this chapter carries are bound to the intellectual-disability workup rather than to regression.

26.1 A practical frame for the differential

Start by examining social communication, restricted or repetitive behaviour, pervasiveness across settings, and any loss of acquired capacity. Was early social communication qualitatively atypical, or merely delayed in proportion to overall development? Are repetitive behaviours, unusual interests, inflexibility, or sensory-linked patterns truly present, including historically? Is the pattern evident across relationships and environments? Does the time course suggest ASD or a neurological, psychiatric, or environmental cause? This frame prevents the common error of treating an isolated surface feature as diagnostic.

Use function, not appearance alone. Repetition may be self-organising, enjoyable, anxiety-reducing, an attempt to avoid an aversive demand, or a response to an intrusive thought. Social withdrawal may reflect a pervasive social-communication difference, fear of scrutiny, low mood, psychosis, or an attachment-related adaptation. The meaning of the behaviour, its developmental onset, and what changes it are more discriminating than the behaviour's name.

- Obtain a timeline of social reciprocity, communication, play, interests, daily routines, and any losses of skill.
- Ask about the same behaviour at home, school, and with familiar and unfamiliar people.
- Separate a core ASD feature from a secondary reaction to anxiety, frustration, communication failure, or environmental change.
- Look for independent evidence that an alternative disorder is present, rather than using it as a label for a difficult symptom.

TRAP **Examination trap.** A child who does not speak in a particular setting does not automatically have ASD. Pervasiveness of the social-communication difficulty and the presence or absence of restricted or repetitive behaviour decide the direction of enquiry.

26.2 ADHD and intellectual developmental disorder

ADHD and ASD can be difficult to discriminate. Interrupting, intruding into personal space, excessive volume, impatience, and poorly modulated interaction can create social difficulty in ADHD. These behaviours should not be mistaken for the qualitative reciprocity and communication pattern of ASD. The discriminating history is the developmental course, together with whether restricted or repetitive behaviours and unusual interests are absent. When they are absent and the social difficulty tracks inattention, impulsivity, or hyperactivity, ADHD is the better explanation.

These disorders may also coexist. In a person with ASD, a concurrent ADHD diagnosis should be considered when attentional difficulty or hyperactivity exceeds what is expected at a comparable mental age. This is a clinical comparison against developmental capacity, not a shortcut based on chronological age. Do not use the presence of ASD to explain away disabling inattention, and do not use the presence of ADHD to dismiss a developmental history of restricted interests and pervasive social-communication difference. ADHD itself is covered in the ADHD and specific learning/communication disorders notes.

The boundary with intellectual developmental disorder demands the same developmental comparison. In intellectual developmental disorder without ASD, social-communicative skills generally do not show an apparent discrepancy from other intellectual skills. ASD is justified when social communication and interaction are substantially more impaired than the developmental level indicated by nonverbal abilities. Observe how the child uses gaze, gesture, affect, shared attention, and nonverbal problem solving, rather than equating sparse speech with autism. This distinction is especially difficult before symbolic communication has emerged and when repetitive behaviour occurs in the context of developmental disability.

ICD guidance makes the same point operationally: assign both diagnoses, with the relevant specifier, when social-communication deficits are greater than would be expected from intellectual functioning. In this setting, adaptive assessment should not let ASD-related social difficulty artificially inflate the apparent global developmental impairment. The conceptual and practical evidence, the developmental record, and repeated assessment matter greatly. The full differentiation of intellectual disability, global developmental delay, and borderline intellectual functioning is addressed elsewhere in this chapter.

26.3 Language, pragmatic communication, and selective mutism

A **developmental language disorder** can produce frustration, reduced participation, and secondary social difficulty. It is not usually accompanied by abnormal nonverbal communication or restricted and repetitive patterns. A child with language disorder may therefore show a strong wish to engage, communicative use of gesture and facial expression, and reciprocal response despite impaired spoken language. The assessment should be designed so that social intent can be seen without demanding verbal fluency.

Social (pragmatic) communication disorder is the nearer diagnostic neighbour because social use of communication is impaired. The separator is the absence of restricted or repetitive behaviour and interests. Before making this diagnosis, enquire carefully about such behaviour in both the past and the present. If ASD criteria are met, ASD takes diagnostic precedence; it is not appropriate to divide an ASD presentation into separate social-communication and repetitive-behaviour diagnoses. Conversely, pragmatic language impairment alone does not justify adding developmental language disorder to ASD. Individuals with a primarily pragmatic language disorder may still initiate and respond appropriately to social and emotional cues and share interests, while lacking the typical restricted or repetitive pattern seen in ASD.

Selective mutism is a disorder of context, not a global absence of communication. Early development is not typically disturbed; communication can be appropriate in some settings; and even where the child is mute, reciprocity is not impaired and restricted or repetitive patterns are not present in the way expected in ASD. Ask what the child does at home, with familiar relatives, and in school. Selective mutism shows normal language and social communication in specific environments but not others, whereas ASD-related social-communication deficits and restricted or repetitive patterns are evident across situations and contexts. Anxiety and ASD can coexist, so a clear setting effect should prompt assessment for both rather than a hurried either-or decision.

PRESENTATION	QUESTION THAT RESOLVES IT	PATTERN FAVOURING THE ALTERNATIVE
Developmental language disorder	Can social intent be demonstrated nonverbally?	Reciprocal nonverbal communication without the restricted or repetitive ASD pattern.
Social pragmatic communication disorder	Has restricted or repetitive behaviour ever been present?	Social-communication impairment without that behavioural pattern.
Selective mutism	Does communication become normal in a safe, familiar setting?	Context-specific mutism with preserved reciprocity.

26.4 Attachment-related presentations, deprivation, and disruptive behaviour

Severe social deprivation can produce **quasi-autism**, an autism-like presentation that must not be assumed to establish a lifelong neurodevelopmental diagnosis without careful history. A key discriminating observation is substantial improvement after movement to a more nurturing environment. Where there is no reliable account of earlier social and communicative development, the distinction may remain difficult. Longitudinal observation is then clinically more honest than premature certainty.

Reactive attachment disorder and ASD share social withdrawal and low reciprocity, but their organising patterns differ. ASD shows atypical language development, whereas delayed language in reactive attachment disorder is not stereotyped or otherwise deviant; restricted or repetitive behaviour is not a feature of reactive attachment disorder; and children with ASD can show attachment behaviour with caregivers at least some of the time despite their social-communication difficulty. A history of severe neglect or maltreatment is required for reactive attachment disorder and is very unusual in ASD. The diagnosis should therefore rest on both the caregiving history and the behavioural phenotype, not on either alone.

Disinhibited social engagement disorder may also enter the differential when a child approaches unfamiliar adults indiscriminately. That feature is not decisive, because some children with ASD may also show it. Restricted, repetitive, and inflexible patterns are not typical of disinhibited social engagement disorder, and a marked reduction in symptoms after a nurturing environment is provided supports that diagnosis. The clinical task is to avoid a false split between adversity and neurodevelopment: adversity may be clinically crucial whether or not ASD is ultimately present.

Oppositional behaviour needs a functional formulation. In oppositional defiant disorder, the social-communication and restricted or repetitive pattern characteristic of ASD is absent. Explosive rages in ASD are often linked to a specific trigger - a disruption of routine, aversive sensory input, anxiety, or interruption of a rigid sequence - rather than a wish to be provocative or spiteful as required for an oppositional account. Ask what occurred immediately before the outburst and what the child was trying to preserve, escape, or communicate. Demand avoidance

may be prominent in ASD, but it should not be mistaken automatically for a separate disruptive disorder.

26.5 Repetitive behaviours: OCD, stereotypies, and tics

The same act can look repetitive while serving very different psychological functions. In **obsessive-compulsive disorder**, compulsions are performed in response to intrusive thoughts, commonly to relieve anxiety. In ASD, repetitive motor acts, insistence on sameness, or ordering behaviour may be experienced as pleasurable and reinforcing. Ask the person, in developmentally appropriate language, what happens if the behaviour is interrupted: is there fear of a feared consequence, a drive for relief or completeness, sensory regulation, enjoyment, or distress at change? The answer is informative but not self-sufficient, because insight and the ability to report internal experience vary.

Resistance is another useful but imperfect axis. People with OCD more often consciously resist compulsive urges, whereas the characteristic social-communication deficits of ASD are not features of OCD even when repetitive behaviour is prominent. Adolescents and adults with ASD may nevertheless attempt to suppress behaviour they know is socially unwelcome. Therefore, neither resistance nor lack of resistance should be converted into an isolated diagnostic test. Evaluate the cognitive antecedent, emotional consequence, developmental context, and the rest of the ASD phenotype.

Stereotypic movement disorder is not added merely because repetitive movements are visible in someone with ASD. A separate diagnosis is not made when the movement is better explained by ASD; it can be appropriate when stereotypy causes self-injury and becomes a specific treatment focus. The practical point is to describe the movement carefully, assess injury and impairment, and decide whether it needs additional clinical attention rather than multiplying labels.

Differentiate stereotypies from **tic disorders** by phenomenology. Tics tend to be less stereotyped, briefer, later in emergence, and associated with premonitory sensory urges; unlike ASD repetitive behaviours, they are not generally experienced as soothing. Direct questioning about an urge, temporary suppression, fluctuation, and relief is useful when language permits. Co-occurrence remains possible, so the aim is accurate description and not forced diagnostic exclusivity.

MNEMONIC

Meaning before movement

For any repetitive act, ask: what precedes it, what does it achieve, what happens if it is blocked, and does the broader social-developmental pattern support ASD? This is a reasoning sequence, not a diagnostic acronym.

26.6 Anxiety, psychosis, and personality pathology

Anxiety disorders overlap with ASD because social withdrawal and repetitive behaviours can be core ASD features but can also express anxiety in either presentation. The central question is

whether social functioning becomes broadly intact when anxiety is low and the context is familiar. In **social anxiety disorder**, interaction with familiar people or in non-anxiety-provoking situations does not show the pervasive impairment expected in ASD; persistent restricted, repetitive, or inflexible patterns are also not features of social anxiety alone. A patient may have both conditions, and avoidant behaviour should then be formulated in terms of both developmental style and fear.

Childhood-onset **schizophrenia** is usually preceded by normal or near-normal development, and hallucinations and delusions are defining features that do not belong to ASD. Its prodrome can nonetheless resemble ASD through social impairment, unusual interests, or odd beliefs. Questions about psychotic symptoms require concrete follow-up: an affirmative literal answer may refer to ordinary media rather than a perceptual experience. Establish the source, location, controllability, conviction, and functional consequence of the reported experience. If ASD and schizophrenia each meet criteria, diagnose both rather than making one erase the other.

In adults without major language or intellectual impairment, personality disorder can resemble ASD through detachment, eccentricity, unusual preoccupations, or restricted affect. The most helpful discriminator is the early developmental course: lack of imaginative play, restricted or repetitive behaviour, and sensory sensitivity in childhood point toward ASD rather than a personality-based explanation. Adult social style alone is an unreliable retrospective diagnostic shortcut. Seek informant history and records where available, and distinguish a lifelong developmental pattern from interpersonal adaptations that emerged later.

26.7 Regression and neurological red flags

Regression changes the diagnostic posture. Loss of skills can occur in ASD. The syndrome-specific accounts belong with their dedicated discussion.

> **GOOD TO REMEMBER**
>
> In ASD, loss of skills is reported in the **second year of life** in some children and rarely occurs after the age of **3 years**. A later loss, especially after a relatively normal early social and language history, should trigger a differential that includes acquired epileptic aphasia, autoimmune encephalitis, Rett syndrome, and other neurological causes.

Medical causes are distinguished from ASD with loss of skills through the history of relatively normal development and the neurological features that are not typical of ASD on their own. Look beyond social communication. Loss of self-care, toileting, motor ability, or other acquired competence makes an encephalopathic process more concerning and warrants more extensive medical investigation than an uncomplicated ASD assessment. Where autistic features are attributable to a medical condition, ICD guidance allows a secondary neurodevelopmental syndrome diagnosis rather than ASD. The distinction matters because immediate management and prognosis can change.

Landau-Kleffner syndrome is an important mimic when apparent autism follows loss of receptive and expressive language after a period of normal development. That period may be **3 to**

7 years. An absent seizure history does not exclude it: **30 to 50%** of affected children may initially have no overt seizures. A careful neurological history and appropriately directed investigation are therefore essential in acquired language regression.

30 to 50%
LANDAU-KLEFFNER SYNDROME MAY INITIALLY LACK OVERT SEIZURES

26.8 Sensory impairment and closing clinical synthesis

Severe hearing impairment, severe visual impairment, maltreatment, developmental coordination disorder, epileptic encephalopathy, mood disorder, conduct disorder, and other behavioural conditions belong in a broad ASD differential when the presentation requires it. Their presence should slow diagnosis rather than become a reason to abandon developmental assessment. Hearing assessment is a named component of ASD workup, and audiology with speech and language expertise belongs in the multidisciplinary team for this reason. The technical differentiation of hearing impairment from developmental language disorder should not be imported uncritically as an ASD rule.

End with a positive formulation: which features establish ASD, which features point to a differential, which conditions may coexist, and what further observation or investigation is required before certainty is justified. This separates a thoughtful assessment from a list of labels. It also protects families from both diagnostic delay and diagnostic overreach. The full child psychiatric assessment battery, services and protection are covered in the Child psychiatry: assessment, treatment, services and protection notes.

> In ASD differential diagnosis, developmental course, pervasiveness across settings, the function of repetitive behaviour, and evidence of neurological regression are more useful than any isolated symptom.

27 · Rett syndrome and childhood disintegrative disorder

Why these conditions matter. Both **Rett syndrome** and childhood disintegrative disorder can present to the psychiatrist as a child who has lost development and now appears autistic. That surface resemblance is clinically consequential but diagnostically incomplete. Rett syndrome is a distinctive genetic and neurological condition with a characteristic course and physical phenotype. Childhood disintegrative disorder is a historical diagnostic construct describing unusually marked regression after a prolonged period of apparently typical development. A careful longitudinal history prevents the error of treating every regression as simply another presentation of autism.

The exam task is not merely to recall labels. It is to identify the pattern of loss, look for the accompanying neurological signs, understand what has changed in current classification, and arrange a proportionate medical assessment. The broader differential diagnosis of developmental regression is addressed in the differential diagnosis of ASD section; here the emphasis is on the two named entities and on the clinical reasoning that separates them from ASD.

99%	about 1 per 10,000	four	1.7 per 100,000
RETT CASES DUE TO SPORADIC MUTATION	REPORTED PREVALENCE IN GIRLS	CLASSIC RETT COURSE PHASES	CDD PREVALENCE ESTIMATE

27.1 Rett syndrome: identity, genetics and diagnostic stance

Rett syndrome was first described by Andreas Rett, an Austrian paediatrician, in Austria in 1966. It is an X-linked dominant condition caused by pathogenic change in MECP2, located at Xq28. MeCP2 binds methylated DNA and participates in the regulation of gene expression and chromatin structure. Its loss disrupts neuronal maturation and synaptogenesis. This mechanistic framing is useful because it explains why the syndrome is not just a behavioural regression syndrome: its evolution includes motor, autonomic, skeletal and epileptic manifestations.

Most cases arise anew rather than being inherited. The proportion attributed to a **99%** sporadic mutation is a memorable genetic fact, while the wider clinical point is that family history may be unrevealing. The syndrome is reported chiefly in females. A hemizygous mutation has been described as incompatible with life in a male fetus, but this should not be recited as an unqualified explanation for the sex distribution because the literature contains a contrary account of that mechanism. The clinically safer formulation is that rare affected males may have severe neonatal encephalopathy, and rare male cases have been reported with a concomitant XXY abnormality.

The syndrome is a major cause of intellectual disability in girls, described as the second most important cause after Down syndrome. Frequency figures require care because sources use different denominators and epidemiological constructs. A reported prevalence is **about 1 per 10,000 girls**; an incidence estimate is approximately 1 per 10,000 female live births. These should never be swapped in an answer. In practice, their shared message is rarity combined with high clinical importance.

RULE **Rett syndrome remains a clinical diagnosis.** Consensus clinical criteria establish the syndrome; genetic testing supports the diagnosis but does not establish it in isolation.

This principle avoids two errors. First, a compatible molecular result does not replace phenotype and developmental trajectory. Secondly, a negative or non-diagnostic result must not cause a clinician to abandon a convincing clinical syndrome. Variant forms exist, including a very rare preserved speech variant in which mutation carriers may communicate well and have mild intellectual disability; asymptomatic carriers have also been reported. Such variation makes clinical synthesis more important, not less.

27.2 The Rett developmental trajectory

The diagnostic spine is a period of apparently ordinary early development followed by loss of acquired skills and a changing neurological phenotype. The early apparently normal period is reported as **6-18 months**. Delays may become apparent around 6 months, but this is not a substitute for listening closely to the parents' account of the developmental sequence. In the

regressive phase, typically between ages 1 and 4 years, a child may look strikingly autistic. The key question is what happens next, and what accompanies the loss.

The course is conventionally understood as **four** classic phases. Rather than forcing stage labels onto a history, describe the sequence: an early apparently normal period, a short plateau, rapid deterioration of motor and speech abilities, a later plateau that can last for years, and subsequent deterioration of the motor system. This sequence is more clinically useful than a list of labels because it connects the family narrative to examination findings and anticipates changing care needs.

Loss of **purposeful hand use** is particularly discriminating. It is replaced by stereotyped hand-wringing, hand-washing, hair-pulling, clapping, flapping or twisting movements. These movements are not an incidental repetitive behaviour. They represent the substitution of a stereotypy for previously functional manual activity, and therefore should be elicited with a before-and-after history. Hand stereotypies first appear at about 18 months. Bruxism, mouthing and body twisting can accompany the hand movements.

Head growth is also informative. Head circumference can be normal at birth, then head growth decelerates, producing **acquired microcephaly**. The adjective matters: it distinguishes a postnatal deceleration from a head that was already small at birth. Significant microcephaly has been stated to emerge by 2 years of age, although head-growth slowing is not universal. A normal head-growth record therefore does not by itself dispose of the diagnosis; it makes the whole pattern, rather than one sign, more important.

TRAP **Examination trap.** Do not call a child autistic solely because she has social withdrawal, language loss and stereotypies during Rett regression. Ask specifically about loss of purposeful hand use, slowing of head growth, gait change and the later social course.

There is a further reason this distinction matters. Social interest often diminishes during the regressive period but later improves; many affected individuals develop an intense eye gaze or staring. Thus, an early autistic appearance need not remain the dominant social phenotype. In the current manual, ASD should be considered in Rett syndrome only when all ASD diagnostic criteria are independently met. This is a disciplined use of co-diagnosis, not a reflexive relabelling of the syndrome.

27.3 Neurological, autonomic and systemic phenotype

The neurological examination should deliberately seek a pattern, rather than recording isolated abnormalities. Gait dyspraxia or apraxia, truncal ataxia, gait and trunk movement apraxias, muscle wasting and dystonia are described in Rett syndrome. These features change the clinical question from “is this autism?” to “what neurological disorder is producing regression and autistic-appearing behaviour?” A child who can no longer use her hands purposefully and develops an abnormal gait demands this broader formulation.

Breathing abnormalities are another helpful clue. Episodes of hyperventilation and breath-holding can alternate. Their importance is practical as well as diagnostic: they should not be

assumed to be wilful behaviour or anxiety without a careful clinical account. Reflux and swallowing problems may reflect autonomic pathology. Acrocyanosis and trophic skin changes of the feet add to the autonomic and vascular phenotype. These physical features often explain why a narrowly psychiatric interview is insufficient.

Epilepsy is a frequent complication, but numerical statements in this area differ between sources. One source reports over 90% of girls with Rett syndrome having epilepsy, with seizures usually beginning around 4 years; another reports a lower range beginning at a different age. The defensible clinical learning point is that seizure history must be actively sought and that later changes in behaviour, alertness or function should not automatically be attributed to the developmental disorder. The Rett-specific seizure burden belongs in the medical formulation, not as a generic ASD comorbidity statistic.

Scoliosis is common over the course of the syndrome, reported in about 87% of females by 25 years of age. Sleep disturbance also deserves direct inquiry: 85% of preschool children have been reported to have disturbed sleep with night waking and paroxysmal or continuous laughter. These are not peripheral details. Sleep disruption, immobility, seizures, feeding difficulty and spinal deformity can each alter daily function, caregiver strain and the safety of medication choices.

Cardiac risk requires particular prescribing attention. There is an increased risk of arrhythmias associated with a **prolonged QT interval**. In Rett syndrome, avoid prokinetic agents, antipsychotics, tricyclic antidepressants, certain antiarrhythmics, anaesthetic agents and certain antibiotics where this risk is relevant. This is not an instruction to leave distressing symptoms untreated. It is a reminder that a psychiatric prescription belongs within a medical plan that considers cardiac conduction and concurrent treatment.

Despite the apparent downhill trajectory, Rett syndrome is **not a neurodegenerative disorder**. The neuropathological account is one of developmental arrest in infancy rather than progressive inflammatory, demyelinating or degenerative destruction. That distinction protects against a common conceptual mistake: later motor deterioration is real, but it does not prove a conventional neurodegenerative process.

27.4 Classification and the autism boundary

Rett syndrome is no longer included as an ASD or pervasive developmental disorder subtype in DSM-5. This differs sharply from childhood disintegrative disorder, which was absorbed into ASD. The historical placement of Rett beside autistic disorders should therefore be taught as classification history, not current nosology. The separation has validating evidence: MeCP2 mutation is not found in other autism spectrum disorders, making Rett a distinct genetic and neurological entity rather than simply a severe ASD subtype.

The current ASD diagnosis nevertheless contains a useful documentation route. The specifier **associated with a known genetic or other medical condition or environmental factor** can be applied when the individual has a known genetic condition, with Rett syndrome named as an example. The specifier does not itself claim that every aspect of the autistic presentation is

caused by the genetic condition. It documents clinically relevant context while preserving the requirement to establish ASD on its own criteria.

In ICD-11, Rett is named as a boundary or differential condition rather than as an ASD category. Diseases of the nervous system with regression are distinguished from ASD with loss of skills by an early history of relatively normal social and language development and by characteristic neurological features. This is exactly the clinical distinction the psychiatrist should make at the bedside. The legacy codes are F84.2 in ICD-10 and 299.80 in DSM-IV or DSM-IV-TR; do not use them as current ASD subtype labels.

> A child with regression and autistic-appearing behaviour needs a syndromic neurological formulation: in Rett syndrome, social interest may later improve, whereas the hand, head-growth, motor and autonomic phenotype remains diagnostically central.

27.5 Childhood disintegrative disorder: the historical construct

Childhood disintegrative disorder is also known as Heller syndrome or Heller's disease and was originally termed dementia infantilis. It described a child with a substantial period of apparently normal development followed by marked loss of skills and the emergence of an autistic clinical picture. The eponym is worth recognising because older examinations, case records and ICD-10 terminology may use it, but current clinical reasoning should not mistake a legacy label for a separate contemporary ASD category.

The historical criteria required at least **2 years** of apparently normal development before regression. Regression begins no earlier than 2 years and no later than 10 years; the most typical onset is between 2 and 4 years. These constructs are easy to confuse. The first specifies the preceding normal developmental interval, the second the permitted boundaries for regression, and the third the modal onset. In a vignette, establish the timing before deciding that the term fits.

Under the legacy DSM-IV-TR criteria, loss had to occur in two or more domains. The domains were expressive or receptive language; social skills or adaptive behaviour; bowel or bladder control; play; and motor skills. This breadth of loss is clinically important. A history that includes toileting and motor loss cannot be reduced to a narrow social-communication account. It should trigger medical evaluation alongside developmental and psychiatric assessment.

- Ask what the child could actually do before decline, using concrete examples rather than global parental labels.
- Establish whether language, social or adaptive abilities, continence, play and motor skills were all affected, and in what sequence.
- Seek neurological and systemic symptoms alongside the behavioural change, then pursue the regression differential described in the differential diagnosis of ASD section.

Following the regression, **autistic symptomatology** emerges, with impairments in socialisation and communication together with restricted, repetitive patterns of behaviour. The clinical

features are otherwise quite similar to autistic disorder, and age at onset is the principal distinguishing feature. This is why a prolonged normal developmental period makes the distinction easier, while a shorter apparently normal period makes it less secure.

27.6 Rett syndrome and childhood disintegrative disorder compared

CLINICAL AXIS	RETT SYNDROME	CHILDHOOD DISINTEGRATIVE DISORDER
Core framing	MECP2-related neurological syndrome	Historical syndrome of regression after apparently normal development
Typical developmental timing	Apparently normal early period of 6-18 months	Regression most typically between 2 and 4 years
High-yield clues	Loss of purposeful hand use with hand stereotypies; acquired microcephaly and motor signs	Loss across language, social or adaptive, continence, play and motor domains
Current DSM position	Not an ASD subtype	Subsumed within ASD
Expected outcome	Long-term neurological disability with changing motor and medical needs	Generally more severe intellectual disability and poorer outcomes than autism

The table should be used as a reasoning aid, not as a substitute for a history. Rett syndrome has a biologically distinctive phenotype, while childhood disintegrative disorder is defined by its temporal pattern of broad regression. In both cases, apparent autism is a clinical presentation that must be interpreted in context. The most hazardous mistake is diagnostic closure after observing social withdrawal or repetitive behaviour.

27.7 Current status, intervention and outcome in childhood disintegrative disorder

Childhood disintegrative disorder is not a separate diagnosis in DSM-5 or DSM-5-TR; it is encompassed within ASD and referred to as a disorder previously described as childhood disintegrative disorder. There is no CDD category in ICD-11 CDDR. Its legacy codes are F84.3 in ICD-10 and 299.10 in DSM-IV or DSM-IV-TR. These codes are useful for interpreting older records, but they should not be presented as current parallel diagnostic categories.

DSM-5 absorption should not make the regression clinically bland. A child with the pattern formerly called CDD requires extensive medical investigation, because regression warrants a search for neurological and medical explanations. The detailed regression differential belongs in the differential diagnosis of ASD section, and the broader ASD medical workup is addressed in the medical comorbidities and workup section. The psychiatrist's immediate contribution is to preserve the chronology, document the breadth of lost skills and ensure that an autistic formulation does not truncate investigation.

Intervention methods for CDD are identical to those used for autism. This statement does not mean the clinical task is identical: marked functional loss often changes the family's practical needs and the level of support required. The named behavioural programmes, speech and

occupational interventions, and family support are dealt with in their respective sections of this chapter. Here, the key point is that a historical CDD label does not exclude access to ASD-focused intervention.

Outcome is generally more severe than in autism. CDD has been described as having a much worse prognosis, with more severe intellectual disability and poorer outcomes than autism. This is why it is important to be honest with families without becoming deterministic: outcome language should describe present support needs, continuing medical assessment and the need to revise the formulation if the course changes. It should never be used to imply that rehabilitation, communication support or family partnership are futile.

27.8 Practical clinical synthesis

GOOD TO REMEMBER

When a child with possible ASD has lost skills, establish the developmental baseline and order of loss. Ask about purposeful hand use, continence, play and motor ability; then examine for acquired microcephaly, stereotypies, gait or trunk abnormalities, breathing changes and other neurological signs. Decide whether this is ASD with loss of skills, a known syndrome such as Rett syndrome, or a regressive disorder requiring a broader medical differential.

- For Rett syndrome, preserve the distinction between a clinical diagnosis and a supportive molecular result.
- For any psychiatric prescription in Rett syndrome, incorporate the arrhythmia and QT-risk context into the medical review.
- For historical CDD, document the prolonged normal period and the domains of loss before applying a legacy label; in either presentation, communicate that regression deserves explanation, not merely classification.

These two conditions are therefore important not because they add obscure eponyms to ASD, but because they teach a general clinical discipline. Developmental regression is a temporal event with neurological and functional meaning. Recognising its pattern protects the child from premature diagnostic closure and gives the family a more coherent account of why assessment, treatment and support must extend beyond a behavioural description.

28 · Behavioural interventions (ABA, TEACCH, PECS, ESDM)

Behavioural intervention is a clinical method, not a branded package. In autism spectrum disorder, the psychiatrist's task is to translate a broad concern - communication, participation, learning, distress around change, or a behaviour that has become difficult to manage - into an observable target and a coherent programme. The useful question is not which acronym is fashionable. It is which learning opportunity, environmental support, communication route and review process are most likely to help this person participate more effectively in ordinary life.

Behavioural intervention means planned alteration of the conditions in which a skill is taught or a behaviour occurs, with observation of whether the planned change helps. It is not

synonymous with making a child compliant, and it must never be framed as a contest of wills. Good work begins with strengths, interests, communication needs and the burden placed on the person and family. It then turns a vague wish such as “behave better” into a target that can be observed, practised and reviewed. This fragment concerns named behavioural and educational programmes; the service model of early intervention belongs in the management of intellectual disability, while speech and occupational therapy and family support are addressed separately.

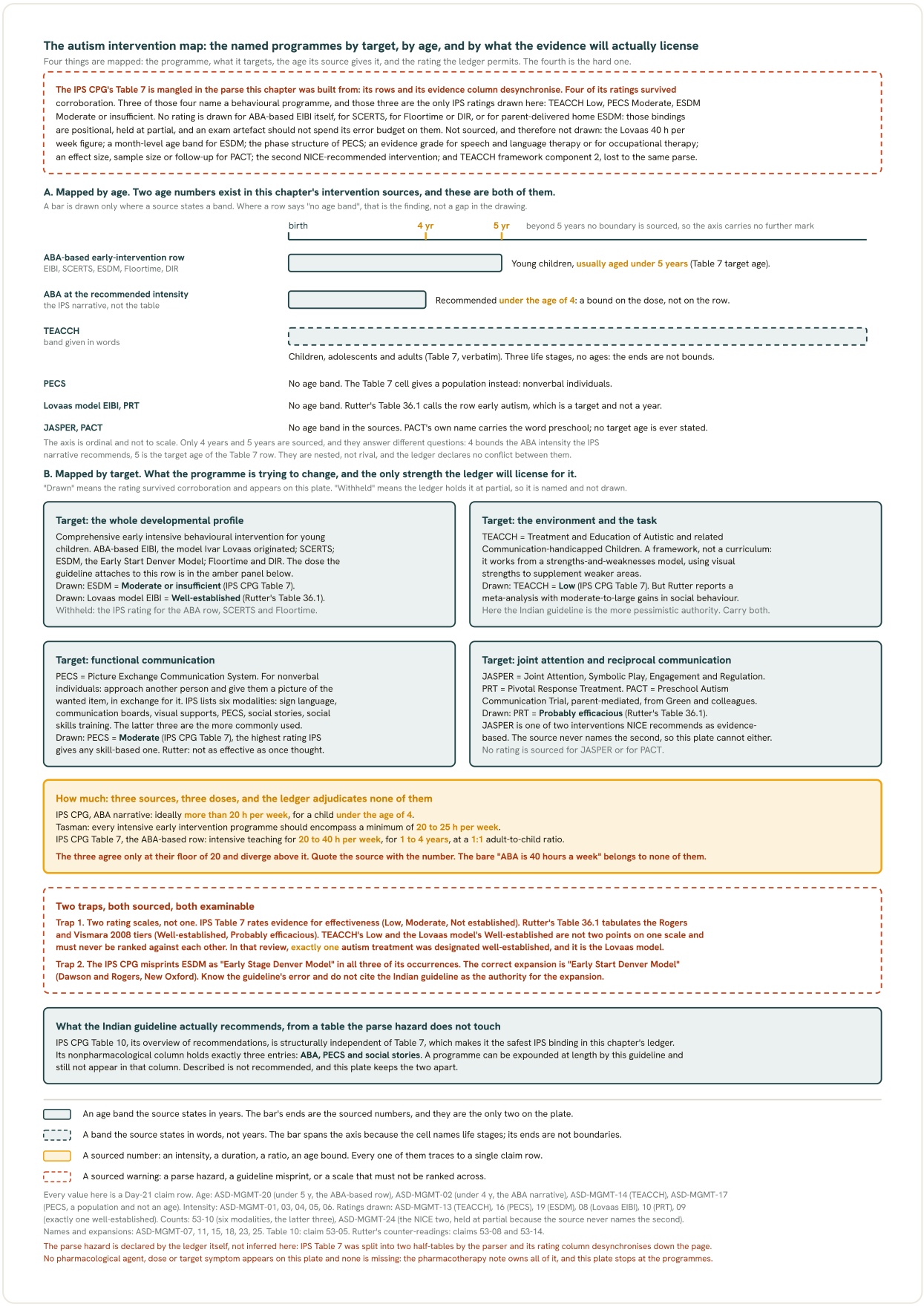


Fig 21.17 **The autism intervention map.** The named behavioural programmes placed by age and by target, carrying only the evidence ratings that survived corroboration: the IPS guideline's Table 7 is mangled in the parse behind this chapter, so TEACCH Low, PECS Moderate and ESDM Moderate-or-insufficient are drawn while the ratings for ABA-based EIBI itself, SCERTS, Floortime and parent-delivered home ESDM are named and withheld.

28.1 Clinical logic before programme selection

A programme label does not replace formulation. The clinician should first establish what is happening, where it happens, what tends to precede it, what follows it, and what the person may be communicating through it. The same outward behaviour can call for quite different changes. This is why copying a technique from another child's plan is poor practice.

Functional formulation links a target behaviour to its context and probable purpose. It makes the intervention humane and testable. Behavioural reasoning should reduce avoidable distress, not explain it away.

GOOD TO REMEMBER

When a child leaves a task, ask what the behaviour may be communicating before choosing a response. It may reflect an incomprehensible demand, a need for a break, an attempt to obtain an item, sensory overload or distress needing medical or psychiatric assessment. The plan may need a communication response, a better-matched task, clearer cues, a smaller step or a predictable way to ask for help.

RULE **Treat the function of a behaviour and the person's communication need, not merely its visible form.**

When the target and its context are explicit, family members, teachers and therapists can work towards the same practical outcome. When they are not explicit, adults often respond inconsistently and the plan becomes a series of reactions rather than treatment.

Targets should be meaningful to the person's daily life. They may concern requesting, tolerating a transition, joining a routine, sharing attention, completing a manageable activity, or reducing a barrier to participation. Progress must be interpreted in context: a newly learned response that occurs only in a training room has not yet become a dependable everyday skill. The clinician should ask where a skill is expected to occur, who can prompt it, what support makes it possible, and how the support will be reduced or adapted when appropriate. This keeps the programme connected to participation rather than to worksheet performance.

- State the target in observable language and identify why it matters in daily life.
- Record the setting, likely antecedents, consequences and communication context.
- Choose a teaching route that matches the person's current access to communication and learning.
- Review benefit, burden, generalisation and acceptability with the family and educational team.

28.2 Applied behavioural analysis and intensive behavioural teaching

Applied behavioural analysis is a framework for using behavioural principles to define targets, arrange teaching opportunities, record response and alter the plan in the light of observed learning. Its strength is not a rigid script. It is disciplined attention to what is being taught, what cues are available, what response is expected, what consequence makes a response worth repeating, and whether the skill transfers outside the teaching situation. A plan can be systematic without being mechanical, and it can be individualised without becoming unmeasurable.

Teaching may be organised into brief, clearly cued learning opportunities or embedded in meaningful activity and routine. The clinical distinction is useful: a more structured format can help when a new discrimination needs clarity, whereas opportunities in ordinary activity are important when a response needs to become useful beyond a formal session. In either case, prompting should help the person succeed rather than repeatedly expose failure. The team should watch for prompt dependence, because an apparently learned response may actually be a response to an adult's familiar cue.

Early intensive behavioural intervention is an ABA-based programme organised around a broad developmental teaching plan rather than an isolated target. The model is associated with **Ivar Lovaas**. A historical evidence review reported that only one autism treatment met its well-established category, namely that EIBI model, classified as well-established. That classification is useful for understanding the evidence history; it should not be turned into a promise about any individual child or into a reason to disregard individual goals and feasibility.

Intensity is often discussed as though it alone determines quality. It does not. The relevant question is whether there is enough repeated, purposeful teaching for a child to learn and use skills, while preserving play, rest, family life, education and responsiveness to distress.

IN INDIA

The IPS guidance recommends, ideally, more than **20** hours per week of ABA for children below 4 years. A broader intensive-programming statement sets a minimum of **20-25** hours per week. These are programme-intensity statements, not a prescription to fill every waking hour with adult-led drills.

20-40	1-4	1:1	<5
ABA-BASED EIBI TEACHING, WEEKLY HOURS	ABA-BASED EIBI PROGRAMME, YEARS	ADULT-TO-CHILD RATIO STATED FOR THE EIBI ROW	USUAL TARGET AGE IN THE EARLY-INTERVENTION ROW, YEARS

More specifically, the ABA-based EIBI row describes teaching between **20-40** hours per week over between **1-4** years, with an exactly 1:1 adult-to-child ratio for that EIBI row. The same early-intervention row is directed at young children, usually below 5 years. These details describe an intensive programme architecture. They do not mean that every component, or every teaching moment, must be delivered in an identical format.

The psychiatrist should make the architecture clinically workable. Ask who delivers each part, how parents understand the aim, whether the educational setting can reinforce the same communication route, and what data will trigger a change in plan. A programme can be intensive on paper but ineffective if targets are poorly chosen, staff turnover is high, the child is distressed, or skills never leave the treatment room. Conversely, a lower-resource setting can still use a disciplined behavioural formulation when it selects priorities carefully and trains the adults who share the person's routine.

TRAP **Examination trap.** Do not equate ABA with a single teaching technique, or assume that a stated adult-to-child ratio applies only to one teaching format. The cited ratio belongs to the ABA-based EIBI row as a whole.

28.3 Structured TEACCHing

TEACCH expands to **Treatment and Education of Autistic and related Communication-handicapped Children**. Its clinical contribution is the deliberate use of structure so that expectations are legible. Rather than assuming that verbal explanation will organise behaviour, the environment, task presentation and sequence are made easier to interpret. This can reduce ambiguity for a person who finds open-ended social or practical demands difficult to process.

Structured TEACCHing is best understood as a way of making the environment do some of the organising work. The framework is described as having exactly four components. In clinical use, the central question is whether the arrangement helps the person see what is happening now, what is expected next, how much is involved, and when an activity is complete. A visual or physical cue is not therapeutic merely because it has been produced; it is useful only if it reduces uncertainty and supports a meaningful response.

Visual structure can be incorporated into home, school or adult settings without reducing the person to a timetable. It may clarify transitions, make materials easier to locate, distinguish work from leisure, or show the endpoint of an activity. The clinician should avoid turning structure into inflexibility. If a plan is imposed without attending to preference and communication, it can become another demand. The better aim is predictable access to activity and choice, with room for the person to signal refusal, need for help, or a wish to change course.

The evidence-for-effectiveness rating listed for TEACCH is low. This should prompt honest discussion rather than dismissal. A low rating does not prove that a structured environment is useless for an individual; it says that confidence in an effect from the cited evidence base is limited. TEACCH is listed for children, adolescents, and adults, which is clinically important because environmental clarity can remain relevant across settings and stages of life. The decision should be based on a specific participation problem, a clearly described support, and observable review.

28.4 PECS and functional communication

PECS means **Picture Exchange Communication System**. It is a communication intervention in which pictures or symbols can provide an accessible route for an individual to initiate an exchange. Its relevance is not that pictures are inherently superior to speech. Its relevance is that a person who cannot reliably use speech may still have a clear reason to communicate, and should have a practical means of doing so.

Functional communication is the clinical aim: a response that changes the person's access to something meaningful in a socially understandable way. When an individual can request, refuse, seek help or indicate a preference, adults need not infer every need from distress or behaviour. This is why communication planning belongs in behavioural formulation. It may reduce conflict

by replacing an ineffective route with a more reliable one, but it should never treat communication as a device for suppressing autonomy.

PECS is listed for nonverbal individuals, with a moderate evidence-for-effectiveness rating. In assessment, establish whether the person notices the visual representation, can physically manage the exchange, understands what will follow, and has communication partners who will respond consistently. The picture, book or display is not the intervention by itself. The intervention is the repeated, successful experience that an intentional act reaches another person and has a meaningful consequence.

Clinical review should include whether the communication route is available where it is needed, not just during planned teaching. A system that stays in a cupboard or depends on one trained adult can produce a misleading appearance of non-response. Equally, the clinician should not make a symbolic system a compulsory endpoint. The right question is whether it expands the person's agency and participation. The separate discussion of speech and language therapy addresses broader communication assessment and therapy; the present focus is the behavioural logic of an accessible communicative exchange.

MNEMONIC

MAP the communication plan

Meaningful need, **A**ccessible response, **P**artner who responds. If any part is missing, the plan risks becoming a display rather than communication.

28.5 ESDM within early behavioural programmes

ESDM is the **Early Start Denver Model**. In a clinical plan it should be named accurately and then linked to a specified developmental and behavioural target, rather than invoked as a generic synonym for good early care. It appears in the same ABA-based early-intervention row as EIBI and other named programmes. That placement helps the candidate remember that programme labels are not competing diagnoses: they are organised approaches whose usefulness depends on the individual's needs, the quality of delivery and the fit with family and educational contexts.

The evidence rating listed for ESDM is **moderate or insufficient**. This wording requires restraint. It should not be simplified into a claim of established benefit for every outcome, nor used to imply absence of any possible benefit. When discussing it with a family, be precise about the proposed targets, the opportunity cost, the support required of caregivers and the plan for reviewing whether the programme is helping. In a fragmented service system, a recognisable model may be less important than a coherent, respectful plan that can actually be delivered and monitored.

The early-intervention row is directed to young children usually below 5 years. This is a description of the target age for that programme row, not a rule that support ceases after that threshold. Across the lifespan, the psychiatrist still identifies barriers to participation and coordinates a plan appropriate to the person's current setting. The form of teaching, the

communication route and the environmental adaptation may change, but the principle of setting meaningful goals and reviewing their effect remains constant.

28.6 Comparing programmes without false equivalence

Named programmes overlap in their concern with learning and participation, but they do not answer precisely the same clinical question. The accompanying table is a way to hold the distinction in mind. It is not a menu from which a family should be asked to choose unaided. The psychiatrist's role is to identify the main barrier, explain what a proposed approach is intended to change, and ensure that claims about evidence are communicated with their stated limits.

APPROACH	CLINICAL ORGANISING IDEA	EVIDENCE OR SCOPE STATED IN THE SOURCE	QUESTION FOR REVIEW
ABA-based EIBI	Planned behavioural teaching across meaningful targets	Between 20-40 hours per week over between 1-4 years	Are skills becoming useful outside teaching?
TEACCH	Environmental and task structure that makes expectations clearer	Low evidence rating; listed for children, adolescents, and adults	Does the support reduce ambiguity and improve participation?
PECS	Accessible symbolic exchange for communication	Moderate evidence rating; listed for nonverbal individuals	Can the person initiate a meaningful exchange in daily settings?
ESDM	Named early behavioural programme linked to individual goals	Moderate or insufficient evidence rating	Are proposed goals, burden and observed benefit explicit?

The comparison also guards against a familiar clinical error: treating an evidence rating as a property of the child. It is not. Ratings describe a body of evidence for an intervention as studied and categorised. The individual question remains whether a carefully specified, feasible intervention is improving a valued outcome. A programme with a stronger evidence classification may still be poorly delivered or poorly matched; a supportive adaptation with limited evidence may be reasonable when it has a clear rationale, low burden and observable benefit. Neither statement licenses therapeutic drift. Every plan needs a target, a responsible team, a way to observe change and a point at which it will be reconsidered.

28.7 Monitoring, ethics and the psychiatrist's coordinating role

Treatment fidelity means that the agreed intervention is actually being delivered in a recognisable and consistent way. It is an ethical as well as a methodological issue. Families are often asked to commit time, effort and money. If the target is vague, adults use incompatible prompts, or records are collected without changing the plan, the family carries the burden while nobody can say what is being tested. Clear documentation protects both the person and the team.

Monitoring should be practical. It can include the frequency or context of a target response, the level of assistance needed, the settings in which it occurs, the person's affect and signs of distress, and the views of family and staff. The record is not an end in itself. It should answer whether the

intervention is helping, whether the goal remains meaningful, and whether a different explanation of the difficulty is now more plausible. A plateau is not a moral failure by the child or caregiver. It is a prompt to re-examine the formulation, access to learning opportunities and competing sources of distress.

The psychiatrist also has a boundary-setting role. Behavioural work must preserve dignity, assent where it can be expressed, choice, and access to communication. It should not make the suppression of harmless difference its central aim. Concerns about pain, sleep, seizures, anxiety, mood change, trauma, bullying, family stress or adverse effects of medication require appropriate assessment rather than escalation of a behavioural plan. The wider assessment and care system is covered in the Child psychiatry: assessment, treatment, services and protection notes.

What the candidate should be able to defend. A sound answer does more than expand acronyms. It explains why a target was chosen, how the proposed method fits the observed barrier, what the evidence statement does and does not justify, and how progress will be judged in ordinary settings. The best intervention plan is not the most elaborate one. It is the one that gives the autistic person a more understandable environment, a more usable route to communication and participation, and a team capable of learning from the result.

> **Remember:** Select ABA, TEACCH, PECS or ESDM by the clinical problem and delivery context, then judge the programme by meaningful everyday change rather than by its acronym.

29 · Pharmacotherapy in ASD (target symptoms, risperidone, aripiprazole)

Medication in autism is symptom-directed. It is not a treatment for the autistic condition as a whole. No medicine is established to alleviate the **core features of autism spectrum disorder**. This distinction is the conceptual anchor for a safe prescription: the clinician is treating a disabling, observable target and its consequences, not attempting to medicate autistic identity or to substitute a drug for developmental, environmental, or family work.

The marks in this area come from clinical precision. A good answer begins by naming the target behaviour, judging its severity and impact, and stating why medication is justified now. It then selects a medicine whose evidence fits that target, starts conservatively, monitors benefit and harm with the same discipline, and plans review from the outset. The prescription is therefore a time-limited clinical experiment with explicit outcomes, rather than an automatic response to a diagnosis.

29.1 The target-symptom framework

Irritability in autism is a clinical cluster, not merely a description of a child who is difficult to manage. In the regulatory construct for risperidone, it includes aggression towards others, deliberate self-injury, tantrums, and rapidly changing moods. These behaviours may cause injury, exclude the person from ordinary settings, exhaust carers, or make education and communication

impossible. They deserve active treatment, but the treatment target must be stated in behavioural language that can later be reviewed.

RULE **Prescribe for a defined target behaviour and measurable impairment, never for autism itself.** A medication trial is defensible when the target is severe, persistent, and associated with meaningful risk or loss of function, after the immediate context has been understood. It is not defensible simply because a person is autistic, has repetitive behaviour, or causes concern to others.

Before calling a presentation psychiatric, reconstruct its function. Consider what precedes the behaviour, what follows it, whether pain, fear, sensory overload, communication failure, a demand that exceeds capacity, or a change in routine is maintaining it, and whether the behaviour has become a reliable route out of an intolerable situation. This is not an argument against medication. It is what prevents a medicine from being used to silence the only available signal of distress. The wider assessment of co-occurring medical and psychiatric conditions belongs with the medical-comorbidity discussion; here the practical point is that an unexplained new target should be assessed before it is medicated.

Agree in advance what improvement would look like: fewer episodes, less intensity, less injury, better participation, or greater ability to use supports. Also agree what would make the trial unsuccessful: no clinically meaningful change, unacceptable adverse effects, or a target that turns out to have been incorrectly formulated. Carer observations are valuable, but must be joined to direct clinical enquiry and the person's own communication wherever possible.

TRAP **Examination trap.** Do not write that antipsychotics treat the social-communication differences or restricted interests of autism. Their established role in this section is management of severe irritability and associated dangerous behaviour.

29.2 Deciding whether medicine is needed

A target-symptom formulation protects against diagnostic overshadowing. The question is not whether the behaviour appears dramatic; it is whether there is a sufficiently clear syndrome of irritability, a serious consequence, and a realistic opportunity to judge change. It also protects the person from prolonged exposure to a drug when its intended benefit was never specified.

- Describe the target in observable terms and identify the harm, distress, or loss of access that makes it clinically important.
- Seek a working explanation for the behaviour, including communication and environmental contributors, before interpreting it as a medication-responsive syndrome.
- Record a baseline and choose a small set of outcomes that matter to the person and carer.
- Discuss the expected trade-off openly: reduction of dangerous behaviour may be valuable, but sedation, appetite change, movement effects, endocrine effects, and weight change may alter quality of life.

This approach also clarifies the place of non-drug intervention. Environmental adaptation, communication support, behavioural work, and carer support remain necessary even when

medication is appropriate. A drug may lower the intensity of a crisis enough for those measures to become usable; it does not replace them. The detailed programmes and therapies are addressed elsewhere in this chapter.

Shared decision-making is particularly important where the observable benefit is reported mainly by adults around the child. Ask what has changed for the person: comfort, participation, sleepiness, appetite, mobility, alertness, and ability to engage. A quiet child is not necessarily a better child. The therapeutic objective is improved safety and access to life, not convenience for a system that has failed to adapt.

29.3 Evidence base and place of risperidone

Risperidone and aripiprazole are the **two FDA-approved atypical antipsychotics** for irritability in children and adolescents with autism. For risperidone, the approved wording is irritability associated with autistic disorder. The listed age range is **5-16 years**. These regulatory facts should not be converted into an indiscriminate rule that every distressed autistic young person requires an antipsychotic. They identify a well-studied target, not a substitute for formulation.

Risperidone has the clearest short-term evidence when severe irritability is the dominant target. In a controlled trial, **69%** of participants receiving risperidone showed substantial reduction in irritability, compared with 12% receiving placebo. Across additional trials, short-term response generally lay between 57% and 72%. The clinical reading is not that response is guaranteed. It is that a clearly observed trial can be worthwhile where the presenting problem is dangerous, severe, and recognisably within this target cluster.

69%	12%	2.7 kg	5.6 kg
SUBSTANTIAL IRRITABILITY REDUCTION WITH RISPERIDONE	CORRESPONDING PLACEBO RESPONSE	MEAN WEIGHT GAIN IN SHORT-TERM LARGE TRIALS	MEAN WEIGHT GAIN AT FOLLOW-UP IN THE CITED STUDY

Evidence must be interpreted alongside burden. In the discontinuation phase of a risperidone study, 62.5% relapsed when switched to placebo. This makes abrupt, reflexive withdrawal after a response unwise. It does not mean that treatment must continue indefinitely. It means that the earlier formulation and the conditions that supported improvement should be revisited before tapering, and that the family needs a plan for recognising recurrence.

A meta-analysis of 46 randomised controlled trials found risperidone and aripiprazole the most effective agents for irritability, with moderate to large effects relative to placebo. This supports their priority over speculative polypharmacy. It also explains why an answer that simply lists psychotropics loses the clinical point: the evidence concerns a particular target, not a general drug treatment of autism.

29.4 Risperidone: practical dosing and titration

Start low and make each change interpretable. Weight-sensitive paediatric dosing is designed to reduce avoidable adverse effects while allowing time to judge the direction of change. The smaller-weight schedule begins at **0.25 mg/day** and then moves to 0.5 mg/day. Subsequent

increases are 0.25 mg at intervals of 2 weeks or more, within a licensed range of 0.5-3 mg/day whose upper end is not a target for a small child: therapeutic benefit plateaus at **1 mg/day**, and only patients over **45** kg may require more. Read the range as the outer bound of the licence and the plateau as the clinical ceiling. That plateau applies to both weight schedules rather than to the lighter one alone; escalation beyond a useful response should therefore not be routine.

For the higher-weight schedule, the initial dose is 0.5 mg/day, followed by 1.0 mg/day. Further increases are 0.5 mg at intervals of 2 weeks or more, with a total range of 1.0-3 mg/day. The 1 mg/day plateau applies to this schedule too. Only those weighing more than **45** kg may require higher doses, and it is in that subgroup alone that the effect plateaus at 3 mg/day. Reading the higher plateau as licence for every child on this schedule is the error the weight bands exist to prevent. Below **15 kg** there is no paediatric dosing data at all, and the source's instruction is caution for that reason rather than caution about data that exist. The distinction is the whole point: an absent evidence base is not a cautious one.

CLINICAL SITUATION	STARTING AND EARLY DOSE	LATER ADJUSTMENT	INTERPRETIVE POINT
Lower body-weight schedule	0.25 mg/day, then 0.5 mg/day	Increase by 0.25 mg at intervals of 2 weeks or more	Licence 0.5-3 mg/day, but benefit plateaus at 1 mg/day: the top of the range is not a target at this weight
Higher body-weight schedule	0.5 mg/day, then 1.0 mg/day	Increase by 0.5 mg at intervals of 2 weeks or more	Licence 1.0-3 mg/day, but benefit plateaus at 1 mg/day: the 3 mg/day ceiling belongs to patients over 45 kg alone, so a lighter child on this schedule is not titrated toward it

GOOD TO REMEMBER

The table is a dosing aid, not permission to chase the top of a range. At each step, ask whether the pre-agreed target has changed enough to justify further exposure. A partial response may be clinically important if it prevents injury or restores access to school and family life. Conversely, an apparently calmer presentation accompanied by marked sedation may represent harm rather than success. Avoid simultaneous medication changes where possible, obtain information from the settings in which the target occurs, and resist adjunctive psychotropics before the initial trial has become interpretable.

29.5 Risperidone: adverse effects, monitoring, and review

Weight gain is the adverse effect that most often changes the risk-benefit balance over time. In the larger short-term trials, mean gain was 2.7 kg. In a follow-up study it was 5.6 kg, an increase of 16.7% from baseline body weight. A paediatric estimate over a year was 7.5 kg, compared with expected annual gain of 3-3.5 kg/year. These figures make routine measurement clinically necessary rather than an administrative formality.

Prolactin elevation is another important adverse effect. Reported rates in paediatric risperidone users range from 49% to 87%, compared with 2% to 7% on placebo. Galactorrhoea has been reported in 0.8%, and gynaecomastia in 2.3%, of paediatric users. These are not simply laboratory concerns: enquire sensitively about symptoms, pubertal concerns, and distress, and interpret any abnormality in the whole clinical context.

Movement effects require active questioning and observation, especially when altered posture, restlessness, or behavioural change might otherwise be attributed to autism. Tardive dyskinesia was reported in 0.1% of paediatric participants in industry-sponsored risperidone trials, while an annual estimate for young people receiving antipsychotics was 0.3% per year. Transient somnolence after initiation had a median duration of 16 days. The numbers are low enough to tempt omission and important enough to make that unsafe: movement phenomena and sedation may be difficult to elicit from a person with limited speech.

Monitoring must therefore connect directly to the indication. At every review, revisit the original behaviour, weight trajectory, appetite, alertness, movement symptoms, endocrine symptoms, and the person's capacity to participate. If benefit is marginal, adverse effects should carry more weight in the decision. If benefit is substantial, monitoring is still not optional, because an initially acceptable trade-off can become unacceptable as exposure continues.

29.6 Aripiprazole: alternative evidence and dosing

Aripiprazole is the other approved antipsychotic for autism-related irritability in children aged 6-17 years; the FDA approval was granted in 2009. It is an alternative when the same target syndrome is present and risperidone is unsuitable, not a drug for core autism. Its initial dose is 2 mg/day. The usual range is 5-15 mg/day, the maximum is 15 mg/day, and titration proceeds by 5 mg/day at intervals of no less than 1 week.

Across the pivotal trials, active-arm responder rates were 52.2% and 55.8%, while placebo rates were 34.7% and 14.2%. The trial-to-value pairing is not reliable and should not be asserted. The defensible conclusion is that the medicine has evidence of benefit for irritability, but the size of individual benefit and the placebo contribution need careful reading rather than slogan-level certainty.

At short-term follow-up, aripiprazole-associated weight gain was 1.3-2 kg, somewhat lower than that reported with risperidone in the cited trials. It was not associated with prolactin elevation. These differences may matter when choosing between agents, especially where weight or endocrine effects have already shaped the discussion. They do not make aripiprazole benign: treatment-emergent extrapyramidal symptoms and vomiting were reported more often than in the risperidone studies. Moreover, aripiprazole may be ineffective for self-injurious behaviours. The target behaviour, rather than an imagined universally preferable drug, should drive selection.

The comparative choice is therefore a conversation about the individual target and anticipated trade-offs. Risperidone offers a strong evidence base with substantial concerns about weight and prolactin. Aripiprazole offers a distinct adverse-effect pattern, including movement effects and vomiting, and may have limitations where self-injury is the central target. Both require the same

disciplined baseline, follow-up, and willingness to stop if the promised benefit does not materialise.

29.7 Restricted and repetitive behaviour and the limits of extrapolation

Restricted and repetitive behaviours and interests are often distressing, but they should not be treated as a single undifferentiated drug target. A Cochrane review found no evidence of effect and emerging evidence of harm for selective serotonin reuptake inhibitors in children with autism. This is an important corrective to the tempting, but unsound, analogy that repetitive behaviour in autism should automatically be treated as though it were an obsessive-compulsive symptom.

The negative citalopram trial illustrates the point: responder rates were 33% with citalopram versus 34% with placebo. Evidence should be allowed to overrule pharmacological intuition. If an SSRI is being considered for another clearly formulated indication, that decision requires its own assessment; it should not be presented as treatment of core repetitive behaviour in a child with autism.

Where fluoxetine is used in the paediatric regimen cited for repetitive behaviours, the starting schedule is 2.5 mg/day for 1 week and the maximum is 0.8 mg/kg/day. These dosing details do not overturn the negative child evidence. In an examination answer, that distinction earns more credit than listing a drug without stating the weakness of its indication.

29.8 Duration, discontinuation, and the longitudinal plan

Planned discontinuation belongs in the initial prescription, not only after adverse effects emerge. For irritability in autism, antipsychotic treatment appears beneficial for **6-12 months**, after which discontinuation should be considered. This is a prompt to review, not a rigid deadline. The clinician should ask whether the original syndrome persists, whether the environment and communication supports have changed, whether adverse effects have accumulated, and whether a carefully observed reduction is now feasible.

> The success of medication in autism is not a quieter person. It is a safer, less distressed person with greater access to ordinary life, at an acceptable cost in adverse effects.

When a person has improved, explain that withdrawal is also a clinical test. A gradual, monitored reduction allows the team to distinguish continuing need from inertia. If the target returns, reconsider the formulation rather than simply restoring medication without thought: relapse may identify a genuine medication-responsive syndrome, an environmental trigger, loss of support, or an insufficiently prepared taper. If the target does not return, the successful outcome is not merely drug cessation but a person spared unnecessary exposure.

For the MD or DNB candidate, the mature answer is consequently structured rather than encyclopaedic: state that core autism is not medicated, define the target, justify either risperidone or aripiprazole for severe irritability, give cautious dosing, name the adverse-effect trade-off, and

make review and discontinuation part of treatment. That sequence shows pharmacological knowledge and, more importantly, clinical judgement.

30 · Speech and occupational therapy and family support in ASD

The practical aim is participation, not performance. Speech and language work, occupational therapy and family support translate an autism formulation into a plan that can be used during ordinary communication, play, self-care, learning and community life. They are not interchangeable therapies, and they should not be presented to families as a menu of equally established options. The useful clinical question is: which barrier is limiting this person's participation now, and what change can the child, family and communication partners realistically carry across settings?

A good plan makes communication more available, adapts demands and environments, and gives carers a role that is sustainable. It also makes uncertainty explicit. This matters especially where an intervention has an appealing explanatory story or a memorable label: plausibility, popularity and individual preference do not by themselves establish effectiveness. The psychiatrist's contribution is to hold the formulation together, protect the family from exaggerated promises, and ensure that therapy goals remain linked to daily function rather than to a fashionable technique.

30.1 Speech and language therapy: from language form to functional communication

Speech and language therapy begins with an account of how the person currently communicates, what communication partners understand, and where breakdown occurs. The assessment is broader than spoken output. It asks what the person can initiate, how they show choice or refusal, whether they can repair a misunderstanding, and which routines make communication easier or harder. The resulting targets should be concrete enough for a caregiver and teacher to recognise in ordinary life. A target that cannot be seen outside the therapy room is difficult to generalise and difficult to judge honestly.

RULE **Treat communication as access to agency.** The person needs a reliable way to request, refuse, share attention and participate, whether or not speech is the route.

This framing prevents a common drift towards making speech itself the sole outcome. Spoken language may be an important goal for some people, but clinical work should not postpone access to communication while waiting for it. The therapist therefore works with the person's existing signals and with the adults who respond to them. The psychiatrist should ask whether the proposed therapy has identified a meaningful communication function, a communication partner, and a setting in which the new skill will be practised.

A **communication partner** is not merely a helper standing nearby. The partner notices an attempt, allows time for a response, interprets it cautiously, and creates repeated opportunities for the person to communicate. This is why family and classroom work cannot be an afterthought to individual sessions. A technically correct target can fail if the adults around the person repeatedly anticipate needs, change the routine without warning, or respond inconsistently.

Conversely, a modest target can become powerful when it is recognised in the home, school and clinic.

Clinical review should distinguish a communication goal from a behavioural label. For example, a difficult moment may represent an unmet need, an inaccessible instruction, an intolerable demand, or an ineffective means of protest. The response is not to assume intent from the behaviour alone, but to test whether a clearer route for communication and a changed interaction pattern reduce the impasse. This keeps speech and language work tied to formulation without turning every difficulty into a speech problem.

- Specify the everyday situations in which communication fails.
- Agree on the response expected from communication partners.
- Choose a visible marker of change that families can recognise without specialist scoring.
- Review whether the skill appears outside the session, not only during prompted practice.

30.2 Augmentative and alternative communication in the therapy plan

Augmentative and alternative communication covers every route that supplements or replaces speech: gesture, signing, objects of reference, picture and symbol systems, and speech-generating devices. The named manualised picture-exchange programme is described with the other manualised interventions in the behavioural-interventions section of this chapter; the question here is the one the speech and language therapist actually answers, which is what the person needs a communicative route *for*, and how that route is chosen, taught and kept available.

The clinical decision is not which system is best in the abstract. It is which communicative acts are currently impossible for this person, and what would make them possible. A route is worth introducing when there is a real need to request, refuse, seek help, comment or indicate a preference; when the person can physically and perceptually manage it; and when the people around them will respond to it consistently. Assessment therefore examines comprehension as carefully as expression, because a system chosen for output alone will be abandoned by a person who cannot follow what it is being used to negotiate.

The practical discipline is to decide what communicative act the route is meant to make possible and whether the family can keep it available in the settings that matter. A picture system that remains in a folder at school is not a functional communication system at home. Equally, the introduction of a symbol system should not lead adults to stop attending to gesture, vocalisation, movement toward an item, or any other communicative attempt. The method should widen the person's options, not narrow adults' attention.

TRAP **Examination trap.** Do not present augmentative communication as a last resort that follows failed speech therapy, and do not describe it as a substitute for therapeutic formulation. It is offered alongside speech and language work from the outset, because an accessible route now does not foreclose speech later; the evidence does not show that a symbol route suppresses talking.

Review asks whether the selected communicative functions have become more available in daily routines, whether the person initiates them with multiple partners, and whether the route itself has become a barrier through poor access, excessive prompting or inconsistent adult response. A system that only works in the therapy room has not yet done its job.

30.3 Occupational therapy: participation, adaptation and sensory questions

Occupational therapy addresses participation in the occupations of ordinary life. Its clinical value lies in observing the fit between a person, a task and an environment, then changing the part of that fit that can be changed. This may involve simplifying a routine, preparing a transition, altering materials, building a practicable self-care sequence, or helping adults recognise what makes an activity inaccessible. The emphasis is not on a universal profile of autism; it is on the particular activity that has become difficult for this person and family.

Sensory integration therapy, associated with Jean Ayres, requires sharper evidence literacy. The IPS narrative frames sensory integration as challenge within a purpose-designed sensory environment, intended to support adaptation in daily activity. It assumes that a child may be receiving too much or too little environmental stimulation. That description explains why the intervention can sound intuitively compelling to families: it connects a visible response to a tangible environment. It is a rationale, however, not an outcome demonstration.

CLINICAL QUESTION	CONSTRUCTIVE RESPONSE	BOUNDARY THAT PROTECTS THE FAMILY
What activity is difficult?	Describe the task, setting, demand and response in functional terms.	Do not infer a single cause from the label “sensory”.
What can change now?	Adapt the routine, materials or adult response and observe participation.	Keep goals visible outside the therapy room.
Is sensory integration proposed?	Discuss its rationale and agree a time-limited, functional review.	Explain the uncertain evidence before commitment.
Are sound-based treatments offered?	Ask what is delivered and what safety safeguards are in place.	Do not treat a sensory rationale as evidence of benefit.

The IPS evidence rating for sensory integration is **Not established, but potentially effective**; its overview also places sensory integration outside the guideline’s recommended nonpharmacological group. This is not a contradiction to be concealed from parents. It is a reason to state the uncertainty plainly, define a functional outcome in advance, and avoid allowing an expensive or time-consuming programme to displace communication, education, family support or medical review.

30.4 Sensory integration and auditory integration: explaining contested evidence safely

A sympathetic explanation is not an endorsement. Sensory distress can be real and can interfere substantially with participation. The clinical response should be respectful observation, environmental problem-solving and a careful search for what the person is communicating

through their response. It should not convert an observation into a certainty that a particular branded intervention will correct an assumed underlying deficit.

Systematic reviews find the evidence base for sensory integration inadequate for recommendation as an evidence-based ASD treatment, with poor study design a central concern. A different textbook account reports **an absence of controlled efficacy studies**. Rather than pretending these summaries are identical, the clinician should communicate their common practical implication: controlled evidence does not establish sensory integration as a treatment for autism. The source-level tension reinforces, rather than weakens, the need for transparent uncertainty.

Auditory integration training needs an even firmer boundary. Reviews do not support a conclusion that it is effective in ASD, and they identify a potential hearing risk from loud sound exposure. The distinction between occupational adaptation and a sound-based treatment matters. A clinician who hears "sensory therapy" should ask what is actually being proposed, what outcome is promised, and whether a potential harm is being minimised in the sales language.

For sensory interventions, validate the difficulty, adapt the environment, define a functional outcome, and state the evidence limit before recommending a programme.

30.5 Family support and parent-mediated work

Family support is clinical work, not a polite adjunct after referral. Families need a coherent formulation, clear goals, practical help in negotiating settings, and space to describe what a proposed programme costs them. A plan that depends on constant availability, specialised materials or flawless delivery may be technically elegant but clinically unsustainable. The psychiatrist should make the burden visible early, especially when multiple services each prescribe home tasks without coordination.

GOOD TO REMEMBER

Before adding a parent-mediated task, ask whether it can be carried across the family's routines. Engaging parents as **co-therapists** can help maintain treatment effects and generalise learned responses across settings, but the same therapeutic tasks can increase parental stress when they become an added burden. Protect capacity: the parent is a partner whose knowledge of routines is indispensable, not an untrained therapist to be judged for poor adherence.

Preschool parent-mediated social-communication interventions teach parents, carers or teachers strategies that increase **joint attention** and reciprocal communication, often through interactive play and action routines. Their evidence should be read with precision. Improvements are usually found in proximal measures closely related to what was trained or in broader developmental measures; autism behaviours are rarely assessed and longer-term outcomes remain unstudied. This does not make the work pointless. It tells the clinician what result may reasonably be expected and what result should not be promised.

JASPER expands to Joint Attention, Symbolic Play, Engagement and Regulation. It is one of **two** evidence-based ASD interventions recommended by NICE in the cited account. The remaining intervention is not named in that source, so it should not be guessed. **PACT** means Preschool Autism Communication Trial and illustrates the same broad clinical principle: the adult-child interaction is a treatment context that needs skilled support, realistic goals and review of burden.

2
NICE-RECOMMENDED EVIDENCE-BASED INTERVENTIONS IN THE CITED ACCOUNT

NICE found too little evidence to recommend a programme for parental stress, mental health or quality of life, and directs practical intervention towards behaviours that challenge. This is a statement about the evidence base, not permission to ignore carers. Clinicians should still listen to strain, coordinate demands, make plans understandable, and reassess whether the family can live with the regimen. Where family distress requires assessment or treatment in its own right, that work belongs within the broader child psychiatric care framework, including the Child psychiatry: assessment, treatment, services and protection notes.

30.6 Matching expectations to developmental stage and setting

RULE **Match the intervention to the functional target.** The target must survive in the setting where it actually matters, not only in the therapy room.

Home, clinic and educational settings each reveal different demands. The point is not to prescribe identical practice everywhere, but to make sure that the adults in those settings understand the same goal and respond in compatible ways. This is particularly important when a child is taught a new communication act in a setting but encounters a different routine, material or response elsewhere.

Social-communication programmes can have a role where social isolation is the therapeutic target. Their evidence does not justify comparable claims for restricted, repetitive behaviours or sensory sensitivities. Candidates should avoid claiming that a communication programme treats every domain of autism. It may improve an interactional pathway while leaving another source of difficulty unchanged. That is not a failure of the child or family; it is an instruction to keep the formulation plural and to revise the plan.

For older school-aged children and young people, the evidence for social-skills and peer-mediated work is methodologically limited. A possible signal in favour of more intensive and longer work is not enough for firm conclusions. In clinical practice, this calls for proportionate claims, collaboratively chosen goals and regular review of whether a group or peer-based format is producing meaningful participation rather than merely attendance.

- Write a shared functional aim in language that family and school can use.
- Identify the adult responses that will support that aim in each setting.
- Ask what new burden the plan creates before adding another home task.
- Review benefit, feasibility and unintended effects together.

30.7 The psychiatrist's synthesis and review

The psychiatrist need not deliver every therapy to lead the treatment plan. The task is to ensure that referrals answer a clinical question, that different professionals are not pursuing incompatible goals, and that the family is not asked to carry an impossible programme. At review, ask what has changed in the specified activity, who has observed the change, where it is seen, and what has become harder. A vague report that therapy is "going well" should be translated into observable participation and weighed against burden.

This approach also guards against false choices. It is not speech therapy versus occupational therapy versus family work. Communication access, environmental fit and caregiver capacity are linked, but each needs a distinct assessment and an honest account of evidence. When an intervention is uncertain, the ethical response is neither contempt nor uncritical referral: explain the rationale, disclose the limitation, agree a meaningful review point and retain interventions with clearer functional value. Document the family's own priorities and the practical barriers that may prevent follow-through. At the next review, revise the plan rather than interpreting an unworkable intervention as a failure of motivation.

31 · Prognosis and outcome in ASD

Prognosis is not a single endpoint. In autism spectrum disorder (ASD), outcome has to be described across the abilities and circumstances that make adult life workable: communication, adaptive functioning, education and employment, relationships, autonomy, mental health, safety, and the fit between a person's needs and the support actually available. A child may make striking developmental gains yet continue to need help in daily life; another may meet fewer diagnostic features over time but remain vulnerable at transitions. The useful clinical question is therefore not whether autism will "go away", but what trajectory is emerging, what risks can be anticipated, and what support will allow the person to participate as fully and safely as possible.

For examination answers, separate the course of autistic features from functional outcome and from service outcome. They overlap but are not interchangeable. A reduction in visible autistic symptoms does not itself prove independence, and a constrained adult outcome may reflect a poor transition environment as much as the person's intrinsic abilities. Prognosis should be communicated as an individual formulation: it recognises uncertainty while identifying practical levers for surveillance, preparation and support.

31.1 The longitudinal frame

Autism is developmental, not a progressive neurological decline. ASD is **not a degenerative disorder**. Learning, adaptation and compensation can continue across the life course, which is why adult outcome cannot be inferred simply from the severity of a child's presentation at one assessment. Development may be uneven: a person can gain language, practical routines or self-awareness while continuing to have substantial difficulty with reciprocity, flexibility, sensory demands or social judgement. Clinical review must therefore look for new demands as well as changes in core features.

At the population level, many people show a **gradual reduction in autistic symptoms** alongside improved adaptive abilities. The most important qualification is the breadth of individual variation. The same diagnostic label includes people whose adult lives are relatively autonomous and people who require continuing intensive support. That variation is not a reason to abandon prognosis. It is a reason to make prognosis descriptive, revisable and grounded in function rather than in a label alone.

RULE **State prognosis as a trajectory of function and support needs, not as a promise of cure or a verdict based on diagnosis alone.**

A minority have a more difficult behavioural course in adolescence, whereas improvement is the more usual direction overall across the long run. This distinction is clinically useful. It prevents the erroneous reassurance that developmental progress eliminates later risk, while avoiding the equally damaging assumption that autism inevitably worsens. Behavioural deterioration requires assessment of change in context, physical health, seizures, psychiatric symptoms, environmental demands and the adequacy of support. It should not be dismissed as an unavoidable feature of ASD.

31.2 Diagnostic stability and its meaning

Diagnostic stability is one strand of outcome research, not a complete outcome measure. A systematic review brought together 23 longitudinal studies of children diagnosed using earlier diagnostic categories. Such work asks whether a later profile still crosses a diagnostic threshold. It does not by itself answer whether the person is communicating well, participating in education, safe from exploitation, or living with a satisfying degree of autonomy. A resident should explicitly make this distinction in a long-answer question.

Within the earlier category of **DSM-IV autistic disorder**, the diagnosis was generally stable. At follow-up, however, approximately **15%** no longer met criteria for that earlier category. This should not be translated into a claim that a corresponding proportion of people with present-day ASD lose the diagnosis. The denominator, diagnostic system and question are different. It is better understood as evidence that developmental profiles can change sufficiently for some individuals to cross a categorical boundary.

Earlier labels of Asperger syndrome and pervasive developmental disorder not otherwise specified showed a **more variable diagnostic course**. Follow-up could move in either direction: some profiles no longer met criteria for a pervasive developmental disorder, whereas others became more consistent with autistic disorder. That finding illustrates a broader teaching point. Categories are useful clinical shorthand, but development is dimensional and context-sensitive. A later reassessment should be based on the current developmental history and functional picture, rather than treated as an administrative confirmation of a childhood label.

TRAP **Do not equate a change in diagnostic threshold with normalisation.** Diagnostic status, symptoms, adaptive functioning and participation must be reported separately.

31.3 What adult outcome studies actually show

Adult outcome studies are difficult to compare because they use different samples, eras of diagnosis, follow-up methods and definitions of a good outcome. Some use a common classification ranging from very good to very poor, while others emphasise schooling, work, living arrangement or social inclusion. These are related outcomes, but they are not synonyms. The evidence base also contains relatively few prospective studies that follow children from early childhood into adult life. That limitation matters when counselling families: precise-sounding estimates may conceal a very particular cohort and a very particular endpoint.

23	approximately 15%	between <5% and 25%
LONGITUDINAL STUDIES IN ONE STABILITY REVIEW	LATER BELOW AN EARLIER AUTISTIC-DISORDER THRESHOLD	VERY-GOOD RATING ACROSS PROSPECTIVE COHORTS

Using that common outcome classification, the proportion rated as having a **very good outcome** lay **between <5% and 25%** across prospective childhood-to-adulthood studies. This is not an employment rate, a school-attendance rate or a prevalence estimate. It is a rating on a particular outcome tool. The correct interpretation is that very favourable adult outcomes occur, but their frequency varies substantially by cohort and definition.

Another older, narrower set of estimates concerns children with **childhood autism**, not the contemporary whole ASD spectrum. Around **10% to 20%** improved from early childhood and subsequently attended an ordinary school and obtained work. A further 10% to 20% could live at home but could not work and required special schooling or a training setting. At least **60%** improved little and could not lead an independent life. These figures are worth knowing only with their boundaries intact: they are historical, category-specific outcome tiers, not a universal prediction for all persons now diagnosed with ASD.

OUTCOME QUESTION	WHAT IT CAPTURES	WHY IT MUST NOT BE SUBSTITUTED FOR ANOTHER OUTCOME
Diagnostic status	Whether current features meet a specified threshold	It does not establish independence or quality of life.
Symptom trajectory	Change in autistic features over time	Improvement can coexist with persistent support needs.
Functional outcome	Communication, practical autonomy, participation and work	It depends on both personal skills and opportunity.
Living arrangement	Where and with what support a person lives	Family resources and service availability influence it greatly.
Outcome classification	A study-defined global rating	It should not be relabelled as a single attainment figure.

Improvement is also not synonymous with the disappearance of autistic traits. People with childhood autism who improve may continue to have language problems, emotionally detached interpersonal style, and unusual behaviour. The clinical implication is respectful realism:

celebrate gains without withdrawing supports prematurely, and assess the person's own goals rather than judging success only against a conventional adult template.

31.4 Predictors: useful, but never deterministic

The strongest recurring predictors are markers, not destiny. Better long-term outcome is associated with **higher intellectual ability** and the presence of useful speech before **5 years of age**. A second account places useful speech at age 5 years. The slight difference in wording should not be forced into a false precision. Both formulations identify the early emergence of functional language as a favourable marker, while preserving the fact that people with similar apparent strengths can have very different adult outcomes.

Why are these predictors clinically plausible? Intellectual ability and functional speech can broaden the routes through which a child learns, signals distress, understands rules, uses education and later negotiates environments. They may also make it easier for carers and teachers to recognise strengths and tailor demands. Yet this is interpretation, not a reason to reduce prognosis to a score or a language milestone. Social understanding, emotional regulation, daily living skills, family circumstances, educational fit, co-occurring conditions and adult services can each alter what those strengths mean in real life.

There is also an **optimal outcome** literature describing a minority whose later presentation no longer meets diagnostic criteria. In observational comparisons, this group had four distinguishing features four:

- milder social impairment at baseline, despite comparable early communication and restricted or repetitive difficulties;
- earlier access to intervention;
- more intensive behavioural intervention services; and
- a greater likelihood of receiving applied behaviour analysis rather than pharmacological intervention.

These observations are important but do not prove that any one service caused the outcome. Baseline social differences may themselves influence both eventual outcome and access to an intervention. Service intensity may also be a proxy for family resources, local provision or a clinician's perception of need. This is the distinction a mature answer should make: observational association can guide hypotheses and service planning, but causal claims require a design able to separate treatment effects from pre-existing differences.

RULE **Use prognostic markers to individualise support and follow-up, never to ration intervention or forecast an individual's ceiling.**

That caution is especially necessary when discussing earlier identification. There has been **no randomised combined screening-and-treatment trial** demonstrating that screening and an earlier age at diagnosis itself lead to better long-term outcome. This does not make timely recognition unimportant. It means that the causal chain should be stated honestly: earlier

recognition may allow access to help sooner, but the long-term benefit of a screening-and-treatment pathway has not been established by that trial design. The detailed approach to early intervention belongs with the management sections of this chapter.

31.5 Adolescence: where trajectories can diverge

Adolescence increases both opportunities and strain. New educational settings, changing social expectations, bodily changes, growing self-management demands and the approach to adult roles can expose a mismatch between a person's coping strategies and their environment. A previously manageable pattern may become more visible when routines change or supports recede. It is therefore good practice to establish a functional baseline and to ask what has changed before assuming a deterioration reflects the autism itself.

Some studies describe a **deterioration in functioning during adolescence** that may coincide with the development of epilepsy or with the onset of psychiatric disorders. The occurrence of epilepsy in ASD, its assessment and medical management are covered in the medical-comorbidity section. In this prognosis section, its relevance is as a warning that a loss of functioning needs active medical review rather than passive observation.

Similarly, the onset of **co-occurring anxiety or mood disorder** may accompany adolescent deterioration. A changed presentation may be behavioural rather than verbal: withdrawal, irritability, sleep change, avoidance, increased rigidity, loss of skills in familiar routines or escalating distress can all be clinically meaningful. Do not presume that every change is a core autistic feature. The map of psychiatric and medical comorbidity, including how to detect it, is addressed in the medical-comorbidity section; pharmacological treatment of target symptoms is addressed separately.

Regression should also be handled carefully in prognostic discussion. A developmental history of regression may shape a family's fear about future loss, but it is not a licence to make a numerical prediction. When there is new loss of function, the clinician must revisit the differential diagnosis and investigate the current change. The differential of regression is considered in the differential-diagnosis section of this chapter.

31.6 Mortality and preventable risk

Mortality in ASD appears **slightly increased** relative to the general population, but the available evidence is limited and no numerical rate should be implied where one has not been established. Prognostic counselling should avoid alarmist language. The clinical value of raising this issue is not to frighten families; it is to focus attention on preventable hazards and on continuity of medical care.

One proposed pathway concerns medical events such as status epilepticus or accidental death associated with epilepsy. This makes seizure monitoring during adolescence a potentially preventable part of risk reduction when seizures emerge or change. The point is not that every deterioration is epileptic, nor that epilepsy explains all excess mortality. It is that a new seizure history, impaired recovery after an event, or a change in functioning deserves prompt clinical attention.

A change in function in an adolescent or adult with ASD is a clinical event: reassess health, mental state, environment and support before attributing it to autism.

31.7 Transition to adulthood is a prognostic intervention point

Services shape outcome. The exit from school can be profoundly stressful and may slow overall progress. The move into adulthood often reveals gaps in adult mental-health care, social care and developmental-disability provision. This is a service problem as well as a developmental challenge. A young person's competence can appear to fall when familiar structures disappear, travel demands increase, family carers become exhausted, or no one coordinates education, work, benefits, healthcare and daily support.

Transition planning should begin with an account of the person's present strengths, communication preferences, sensory and behavioural needs, health risks, decision-making support, family capacity and preferred adult roles. It should identify what the young person can increasingly manage independently and what assistance remains necessary.

GOOD TO REMEMBER

Make the transition plan concrete: who will notice deterioration, who will coordinate appointments, what happens when routine breaks down, and how meaningful daytime activity will be maintained. This protects dignity while avoiding a sudden and unrealistic withdrawal of care.

The evidence for specific transition-planning interventions remains limited, so clinical reasoning matters. Build continuity wherever possible, prepare the receiving service, and make the handover understandable to the person and family. The broader assessment battery, service organisation and safeguarding framework are set out in the Child psychiatry: assessment, treatment, services and protection notes. The detailed discussion of adult ADHD belongs in the ADHD and specific learning/communication disorders notes.

31.8 A practical way to communicate prognosis

A useful prognostic conversation starts with what is already known and names uncertainty without retreating into vagueness. Describe the person's trajectory in domains: communication, adaptive skills, learning, participation, mental health, physical health, safety and support network. Then identify favourable capacities, current barriers and foreseeable transition points. Families often need help distinguishing a realistic plan from a deterministic prediction. The clinician can say that development may continue, that outcomes vary markedly, and that progress is strengthened when demands and support are matched over time.

In a written formulation, avoid a bare phrase such as "poor prognosis". Instead state the active issues and the plan for each. This gives the team a framework for review and gives the family an intelligible account of why monitoring continues even when the person is doing well.

FORMULATION DOMAIN	QUESTION FOR FOLLOW-UP	CLINICAL USE
Developmental trajectory	What has improved, plateaued or changed?	Separates expected variation from a clinically significant loss of function.
Functional participation	What can the person do with the available support?	Prevents symptom counts from standing in for real-world outcome.
Communication and cognition	How are strengths being used in education and daily life?	Uses prognostic markers without turning them into ceilings.
Health and mental state	Has there been a new seizure, distress pattern or behavioural change?	Prompts review of potentially modifiable contributors.
Transition and supports	What will change when current structures end?	Turns a predictable risk point into a planned intervention point.

The final examination-quality conclusion is balanced. ASD usually permits ongoing learning and adaptation, but outcome is highly heterogeneous. Better intellectual ability and early useful speech are favourable markers, not guarantees. Co-occurring epilepsy or psychiatric illness, and failures at major transitions, can worsen functioning and require active review. The best clinical response is sustained, person-centred planning that measures meaningful function and keeps support available as needs change.

Discriminate, apply and consolidate

32 · Differentiation of intellectual disability from autism, GDD and borderline intellectual functioning

The discriminative task. A developmental presentation is not solved by attaching the most visible label to the child. Intellectual disability, autism spectrum disorder, global developmental delay and borderline intellectual functioning may all come to attention through slow learning, school difficulty, communication concerns or behaviour that unsettles adults. They answer different clinical questions. The useful formulation asks what the person can do in everyday life, how development has unfolded, whether social communication is qualitatively atypical for that developmental level, and whether the current assessment is a valid account of ability.

The marks are usually in the reasoning, not in reciting a checklist. Start by separating a broad developmental profile from a distinctive pattern of social-communication and behavioural difference. Then decide whether the profile has reached the threshold for intellectual disability or is better recorded as **borderline intellectual functioning**. This avoids opposite errors: calling every child with delayed language autistic, and using a reassuring test score to overlook everyday limitations that require support.

32.1 Begin with a developmental formulation, not a label

A **developmental formulation** is a map of strengths, difficulties and their context. It is more useful than an early diagnostic label because the same presenting complaint can arise from very different pathways. Slow acquisition of school skills may reflect a generally low level of functioning, a sensory or language barrier, missed learning opportunities, emotional distress, a neurodevelopmental condition, or an assessment that did not capture the child's usual capacity. The clinician therefore compares domains rather than treating a single complaint as the whole phenotype.

History supplies the frame. Establish the tempo of acquisition and whether abilities have continued to accrue, plateaued, or been lost. Ask how the person communicates needs, joins shared activity, copes with ordinary demands, learns from modelling, and manages changes in routine. Observe the same questions in the interview. A score is evidence, but not an explanation. It must be interpreted with language, sensory, motor, educational and behavioural context in view. This is especially important when the interview setting is unfamiliar or the person is anxious, overactive, shut down, oppositional, exhausted, or unable to understand the task demands.

RULE **Rule.** Diagnose the pattern of functioning and its real-world consequences; never let a conspicuous developmental symptom decide the diagnosis by itself.

The comparison must be made against **developmental level**, not merely chronological expectation. This prevents a common false positive for autism. A person with broadly delayed development may use simpler language, play, social approaches and coping strategies than peers. The pivotal question is whether social communication is more impaired, qualitatively different, or oddly uneven even when compared with that person's overall developmental level. Conversely, apparently sociable behaviour does not by itself settle the issue: the clinician asks whether contact is reciprocal, flexible, shared and meaningful, rather than simply frequent.

- Define the referral problem in ordinary functional terms before translating it into a diagnostic question.
- Seek convergent information from the person, family, school and direct observation.
- Describe strengths as carefully as limitations, since unevenness often carries the diagnostic clue.
- Record factors that could have distorted the assessment before treating its result as trait-like ability.

32.2 Comparison at the bedside

The table is a reasoning aid, not a substitute for assessment. Intellectual disability concerns the established diagnostic construct and its functional consequences. Autism spectrum disorder concerns a characteristic social-communication and behavioural pattern. Global developmental delay is a developmental description used when a stable intellectual-disability formulation cannot yet be made with confidence. Borderline intellectual functioning identifies a clinically relevant

zone near the boundary, where careful assessment and context matter more than an automatic diagnostic conclusion.

CLINICAL QUESTION	INTELLECTUAL DISABILITY	AUTISM SPECTRUM DISORDER	GLOBAL DEVELOPMENTAL DELAY	BORDERLINE INTELLECTUAL FUNCTIONING
Central task	Establish the intellectual and adaptive profile	Identify a characteristic social-communication and behavioural pattern	Describe developmental delay while the profile is still evolving	Judge a boundary presentation and its clinical relevance
Pattern of difficulty	Broad functional limitations are central	Qualitative difference may coexist with any intellectual level	Delay is described across development rather than presumed permanent	Difficulties may be real without establishing intellectual disability
Key comparison	Everyday function alongside intellectual assessment	Social reciprocity and behaviour relative to developmental level	Current abilities, opportunities and developmental trajectory	Intellectual and adaptive findings, including their mismatch
Common error	Equating a test result with the whole diagnosis	Calling global delay autism without qualitative evidence	Forcing a permanent label before adequate assessment	Using an obsolete numerical rule as a current diagnosis

Use the table dynamically. A person can occupy several categories in a clinically meaningful sense. Autism can occur with intellectual disability, and developmental delay can be the initial description before the later formulation becomes clearer. The task is not to make the categories mutually exclusive. It is to avoid using a category as an explanation for every feature and to state clearly what each diagnosis contributes to care planning.

> The decisive comparison is qualitative pattern against developmental level, interpreted alongside everyday functioning, not the presence of delay alone.

32.3 Intellectual disability and autism spectrum disorder

When intellectual disability is being considered, autism must be established by a characteristic qualitative pattern beyond the effects of broad developmental limitation, rather than inferred from delayed performance alone.

GOOD TO REMEMBER

Observe how the person uses the abilities they do possess. Do they seek another person for comfort, enjoyment, help or shared attention? Can they respond to a familiar social bid when demands are low? Is there flexibility in the way people and objects are used? Interpret the answers in the context of communication ability and the environment, not as isolated signs.

In autism, the important finding is a disproportionate alteration in **relative social reciprocity** and in the behavioural pattern, rather than delayed performance alone. A person may have very limited speech yet show clear social interest and shared enjoyment that are consistent with their overall developmental level. Another may have fluent speech but persistently atypical reciprocity, difficulty using communication socially, inflexible interests or restricted patterns that cannot be explained by general developmental delay. The latter pattern warrants an autism assessment even when intellectual ability is not clearly impaired.

Do not set up a false contrast in which one diagnosis excludes the other. When autism and intellectual disability coexist, the formulation should state both and then show what observations support the autism diagnosis beyond global delay. That discipline protects against diagnostic overshadowing in either direction. It prevents clinicians from attributing all behaviour to intellectual disability, while also preventing an autism label from obscuring practical support needs, communication limitations or safeguarding concerns associated with intellectual disability.

TRAP **Examination trap.** Delayed language, poor eye contact in an unfamiliar interview, repetitive play at a low developmental level, or behaviour under stress is not by itself evidence of autism. The question is whether the social-communication and behavioural pattern is qualitatively atypical for that person's developmental level.

Clinical interviews should therefore include situations that lower performance pressure. Watch what happens when the examiner follows the person's lead, offers a familiar object, pauses for a response, or involves a caregiver. A narrow clinic observation can overread anxiety, sensory discomfort or communication mismatch as autism, especially when the person has intellectual disability. Equally, a warm greeting or brief eye contact cannot rule autism out. Longitudinal history and reports across settings are what convert an impression into a defensible formulation. Detailed diagnostic criteria and assessment methods for autism belong in the relevant sections of this chapter; here the practical point is to ask whether autism adds a distinctive explanatory pattern beyond general developmental difficulty.

32.4 Intellectual disability and global developmental delay

Global developmental delay is best understood as a developmental description, not as a shortcut to a lifelong conclusion. It communicates that delay is evident across the developmental profile while acknowledging that the available evidence may not yet support a stable intellectual-disability formulation. The distinction matters because a child's observed performance is shaped by opportunity, health, communication access and the feasibility of formal assessment. A label that is too final can close inquiry prematurely; a label that is too vague can fail to communicate support needs.

The key concept is **developmental trajectory**. Ask whether the profile is broadly delayed or strikingly uneven, whether new skills are being acquired, and whether the child's functioning is being assessed under conditions that permit a fair estimate. Repeat assessment may be more informative than forced precision when a child cannot engage with formal tasks, when communication access is poor, or when the environment has substantially limited experience.

The clinician should explain uncertainty plainly: uncertainty is not indecision when it accurately reflects the developmental state of the evidence.

This distinction also changes the conversation with families. The immediate work is to describe present strengths and needs, arrange appropriate developmental and functional assessment, and review change over time. It is not to promise a future intellectual level or to use a temporary descriptive term as a euphemism for a diagnosis that has not been established. The normal developmental framework should be read alongside the *the Normal child development and milestones notes*; assessment, services and protection are addressed in *the Child psychiatry: assessment, treatment, services and protection notes*.

- Use global developmental delay when it accurately describes the current developmental evidence and its limits.
- Do not assume that a broad early delay proves a fixed intellectual-disability formulation.
- Do not assume that later identification of intellectual disability was predictable from one early observation.
- State what must be clarified on follow-up: developmental course, everyday function, communication access and validity of assessment.

The distinction is therefore practical rather than semantic. It determines how confidently the clinician communicates prognosis, how the record represents uncertainty, and why follow-up must revisit the formulation rather than merely repeat a label. It also keeps the assessment open to alternative or coexisting explanations when the profile is not globally uniform.

32.5 Borderline intellectual functioning: category, boundary and limits

Borderline intellectual functioning is not a milder synonym for intellectual disability. In the DSM-5-TR, it is coded **R41.83** and appears in Section II among conditions that may warrant clinical attention. That location is clinically useful: it permits recognition of a relevant cognitive vulnerability without converting every such presentation into a formal mental disorder.

The nosological boundary must be stated accurately. Borderline intellectual functioning is a **condition**, rather than a formal DSM-5 disorder. It should not be treated as a clear-cut disability category or as proof of definite deficits across intellectual and adaptive domains. The category is appropriate when the cognitive profile is itself the focus of clinical attention, or when it materially shapes treatment or prognosis. In practical terms, it can alert the clinician to adjust explanations, consent processes, expectations of independent follow-through, and the format of psychological or pharmacological care, while continuing to assess the actual person rather than acting on a label.

A psychometric zone may be described as **about 70 to 85**, or as one to two standard deviations below the mean, but this is not a current definitional rule. The current DSM-5 approach does not use a numerical IQ criterion to define the category. The older DSM-IV-TR definition explicitly used **IQ 71-84**; importing that band into current diagnostic reasoning is a category error. A

numerical result can prompt a closer look. It cannot replace clinical assessment of function, context and discrepancy.

R41.83	about 70 to 85	71-84	no numerical criterion
CURRENT CODE	DESCRIBED PSYCHOMETRIC ZONE	OLDER DEFINITIONAL BAND	CURRENT DEFINITION

Why this boundary matters. A person can have modest cognitive efficiency yet function capably in familiar, well-supported settings. Another may appear competent until demands increase, language becomes technical, routines change, or treatment requires planning and self-monitoring. Neither observation alone settles whether the person has intellectual disability, borderline intellectual functioning, another condition affecting performance, or a mixture of these. The clinician must avoid both minimisation and overpathologising.

32.6 Separating borderline intellectual functioning from mild intellectual disability

This is the boundary at which disciplined assessment matters most. The current DSM-5 framing expressly compares borderline intellectual functioning with mild intellectual developmental disorder, rather than making a broad contrast with intellectual disability in general. The assessment requires careful appraisal of intellectual and **adaptive functions**, including the **discrepancies** between them. This is not a mechanical exercise. A discrepancy may signal uneven opportunity, a co-occurring disorder, an invalid test session, a skill that is context-bound, or a genuine pattern requiring further understanding.

Work in both directions. If a test result lies near the boundary but everyday functioning is more limited than expected, ask what accounts for the gap before deciding that it demonstrates intellectual disability. Consider communication barriers, educational experience, emotional state, psychiatric symptoms, physical illness, sensory difficulty, family scaffolding and the actual demands of the setting. If everyday functioning appears stronger than a test result, ask whether the person has learned routines exceptionally well, receives extensive unobtrusive support, or had a poor-quality testing experience. The aim is not to rescue a preferred diagnosis. It is to identify the most credible account of the data.

Test validity is central to this judgement. The DSM entry specifically cautions that co-occurring disorders can affect adherence to standardised testing, including schizophrenia and ADHD with severe impulsivity. A low score obtained when attention, motivation, comprehension or behavioural regulation has collapsed is not automatically evidence of stable intellectual ability. Nor should a clinician dismiss all low performance as psychiatric. Reassess the conditions of testing, use collateral evidence, and formulate whether the observed score is likely to represent the person's usual functioning.

The practical record should make the reasoning visible. State the referral question, the data sources, their limitations, the relation between assessed ability and everyday functioning, and what alternative explanation has been considered. Then state why the conclusion is intellectual disability, borderline intellectual functioning, or a provisional formulation. This is kinder to families and more defensible clinically than an unsupported categorical assertion. Intelligence

testing itself and the instruments used for it are addressed in the assessment section of this chapter; do not confuse such testing with a complete functional evaluation.

32.7 Coding, language and the historical trap

The current label has a complicated historical trail, which explains why outdated terminology still appears in records and teaching material. In DSM-II, the 1968 entry used the label "borderline mental retardation" and the code 310. In 1973, the American Association on Mental Deficiency, now AAIDD, eliminated that name in favour of "borderline intellectual functioning".

The coding location also shifted. DSM-III placed it on Axis I with V62.89 in 1980. DSM-III-R moved it to Axis II with V40.00 in 1987; DSM-IV later used Axis II V62.89 in 1994. Axes were removed in DSM-5 in 2013, which used R41.83.

This history should sharpen, not confuse, current practice. It shows why an old numerical band, an old axis placement, or stigmatising terminology is not a substitute for a contemporary formulation. Use language that describes the person's current functioning and support needs. When older records are encountered, translate their meaning cautiously and verify what assessment was actually done before carrying a label forward.

Finally, "borderline" in borderline intellectual functioning has no relationship with borderline personality disorder. Similar wording is not shared aetiology, shared diagnosis or a reason to infer one condition from the other. Keeping this distinction explicit is particularly important when a patient has emotional dysregulation or interpersonal difficulty, because such features can tempt clinicians into semantic rather than phenomenological reasoning.

32.8 A defensible conclusion and its clinical use

Clinical attention is the right frame for borderline intellectual functioning. The category may be recorded when it changes how treatment is delivered or how prognosis is understood, not as a casual descriptor of anyone who struggles academically. Explain information plainly, check comprehension through the person's own account, anticipate difficulties with abstract instructions, and involve supports appropriately while respecting autonomy. These are good clinical practices because they respond to observed needs, not because a code has replaced assessment.

In a written answer, a strong conclusion follows a consistent sequence. Identify whether the central issue is broad functional limitation, a distinctive autism pattern, an evolving developmental profile, or a boundary cognitive presentation. State the observations that support that judgement. Name any coexisting diagnosis separately. Finally, state what must be reassessed and how the formulation changes communication, support and follow-up. This makes the differential useful at the bedside rather than a set of mutually exclusive examination labels.

Diagnostic humility is not vagueness. It means using a provisional developmental description when the evidence is still developing, making an autism diagnosis only when its qualitative pattern is established, recognising intellectual disability through the established formulation rather than a score alone, and using borderline intellectual functioning accurately when it is

clinically relevant without overstating its status. For associated attention and learning questions, use the ADHD and specific learning/communication disorders notes.

33 · Four worked cases

Vignettes test clinical organisation, not label recognition. A short stem rarely gives a complete developmental history, a valid cognitive assessment, an adaptive profile and a collateral account. The candidate's task is to recognise what the available pattern supports, state what remains uncertain, and choose the next discriminating enquiry. The same discipline protects clinical practice from premature labelling.

Use each vignette as a **developmental formulation**: place the person's trajectory, everyday function, social-communication pattern, repetitive or restricted behaviour, medical context and environment alongside each other. A diagnostic label is the conclusion of that process, not a substitute for it.

Trajectory	Function	Pattern	Context
DEVELOPMENTAL COURSE	EVERYDAY INDEPENDENCE	SOCIAL COMMUNICATION AND BEHAVIOUR	MEDICAL, FAMILY AND EDUCATIONAL SETTING

33.1 Read the stem before naming the diagnosis

The useful first question is not “Which disorder is this?” but “What is impaired, in which settings, and since when?” Separate a person's learning and problem-solving capacity from **adaptive functioning**, and separate both from the quality of social reciprocity and the behavioural pattern. School difficulty, delayed language, disruptive behaviour and poor self-care are findings. None is diagnostic in isolation.

RULE **Rule.** Diagnose the pattern of impairment, then test whether the pattern is global, autism-specific, developmental but still evolving, or below expected ability without the pervasive functional deficits required for intellectual disability.

- **Trajectory:** establish whether development has been delayed, uneven, static or marked by loss of previously acquired abilities.
- **Current function:** ask how the person manages communication, self-direction, daily routines, safety and participation in ordinary settings.
- **Pattern and context:** seek social-communication differences, restricted or repetitive behaviour, sensory concerns, physical clues, medical symptoms and the accounts of people who know the person well.

33.2 The central discrimination

Intellectual disability remains an assessment question when a stem describes broad difficulty with learning and everyday function. An autism-spectrum formulation depends on its characteristic social-communication and behavioural pattern. These formulations may coexist. The presence of autism does not answer the question of intellectual functioning, and global learning difficulty does not by itself establish autism.

FINDING IN A STEM	WHAT IT SUGGESTS	WHAT PREVENTS AN OVERCALL
Broad difficulty learning and carrying out everyday tasks	Consider a global impairment formulation	Clarify adaptive functioning and the developmental course
Qualitative difference in reciprocal social communication with a restricted or repetitive pattern	Consider an autism-spectrum formulation	Do not infer intellectual disability from the autism label alone
Developmental concerns in an early developmental presentation without a stable assessment picture	Use **global developmental delay** as a working developmental formulation where appropriate	Plan review as developmental information becomes clearer
Academic or occupational struggle with relative preservation of everyday adaptive competence	Consider **borderline intellectual functioning** and competing explanations	Do not convert a low score, poor scholastic performance or social disadvantage directly into intellectual disability

TRAP **Exam trap.** "Delayed speech" is not a shortcut to autism, and "poor school performance" is not a shortcut to intellectual disability. The discriminating evidence is the whole developmental and functional pattern.

33.3 Vignette: broad developmental difficulty without an autistic pattern

A child is brought because progress across learning, communication and self-care has been slow. The family describes a steady developmental course and needs help across ordinary routines. In the consultation, the child seeks shared engagement in a manner that is proportionate to developmental level, with no clear history of a restricted or repetitive behavioural pattern.

The defensible formulation is that the stem raises concern for intellectual disability or, when developmental assessment remains incomplete, a developmental-delay formulation. The point is not to exclude autism because the child is friendly, nor to diagnose autism because language is delayed. Ask for formal assessment of ability and adaptive behaviour, collateral accounts across settings, developmental history, physical and neurological review, and an aetiological evaluation guided by the wider presentation.

Why the distinction matters. The intervention plan must address the actual pattern of support needs. If autism is added without its defining pattern, families may be diverted from the assessment and supports that explain the person's everyday difficulty.

33.4 Vignette: autism with an uneven profile

An adolescent has intense, narrow interests, inflexible routines and marked difficulty interpreting reciprocal social interaction. Academic achievement is strong in selected areas, while practical independence and social judgement are variable. The stem offers no valid basis to conclude that intellectual functioning is globally impaired.

The appropriate formulation is autism spectrum disorder with a separate assessment of intellectual and adaptive functioning. Uneven strengths do not cancel clinically important disability, but neither do social difficulty and inflexibility prove intellectual disability. Document

the areas of competence as carefully as the areas of difficulty, because support planning follows function rather than assumptions about a diagnostic label.

- Ask which difficulties are intrinsic to social communication and behavioural flexibility.
- Ask whether practical dependence reflects limited capacity, limited opportunity, anxiety, environmental mismatch or a combination.
- Describe co-occurring attention, mood, anxiety, sleep, neurological and medical concerns without attributing all distress to autism.

33.5 Vignette: the apparent boundary case

A young person has struggled academically for years and has been described as "slow." At home and in the community, the person manages familiar routines, relationships and responsibilities with little extra help. An isolated test result is quoted in the referral note, but its validity, context and relation to everyday functioning are not described.

This stem is designed to expose **score determinism**. Consider borderline intellectual functioning, specific learning or communication disorders, attention problems, educational deprivation, sensory impairment, emotional disorder and contextual barriers. For the detailed assessment battery, see the Child psychiatry: assessment, treatment, services and protection notes; for attention and learning differentials, see the ADHD and specific learning/communication disorders notes.

GOOD TO REMEMBER

Do not diagnose from a number or a pejorative description. Interpret formal assessment alongside adaptive function and developmental history.

33.6 Vignette: change after an established developmental course

A caregiver reports that a child who had acquired skills is no longer using them in the same way. The current presentation includes communication difficulty and repetitive behaviour. The temptation is to treat the current surface pattern as sufficient for an autism diagnosis.

Here the clinical priority is a careful account of **regression**, its tempo, the affected skills, associated neurological or medical symptoms, and the possibility of an alternative developmental or neurological condition. Autism may remain part of the formulation, but loss of skills changes the workup and makes a broad differential essential. Do not reassure, diagnose or narrow the enquiry before establishing the trajectory.

MNEMONIC

MAPS

Map the developmental trajectory. **A**ssess everyday adaptive function. **P**attern-match social communication and restricted or repetitive behaviour. **S**earch for syndromic, neurological, psychiatric and contextual signals. This is a retrieval map, not a substitute for assessment.

33.7 Turning a vignette into a clinical answer

A strong answer starts with a provisional formulation, gives the positive findings that support it, then names the information required to distinguish nearby alternatives. It states comorbidities and safety or medical concerns as possibilities to assess, rather than presenting them as conclusions not contained in the stem. A developmental diagnosis does not make new distress, behaviour change or physical symptoms self-explanatory: assess each reported change on its merits.

When asked for management, link recommendations to the person's identified functional needs and family priorities. Do not leap from a label to a named programme, medication or legal entitlement. The relevant chapters provide the detailed intervention, medical-workup, disability-certification and family-support pathways.

> In every developmental vignette, the diagnosis must explain the whole pattern of trajectory, function and behaviour better than its nearest alternative.

34 · Consolidate: the master mnemonic, the retrieval prompts and the synthesis map

Consolidation is a clinical discipline, not a list of labels. Intellectual disability, autism spectrum disorder, developmental delay and borderline intellectual functioning can overlap in a referral, yet each answers a different clinical question. The task is to preserve that distinction while building a single account of the person's abilities, difficulties, context and needs.

A high-quality answer therefore moves from the presenting concern to a **developmental formulation**. It does not let a test result, a screening result, a certificate or a diagnostic label substitute for the history, observation and functional account. The chapter's component sections supply the detail; this closing section shows how to hold them together without flattening them.

MAP	READ	CHECK	PLAN
THE DEVELOPMENTAL COURSE	THE FUNCTIONAL PROFILE	CO-OCCURRENCE AND CONTEXT	MEANINGFUL SUPPORT

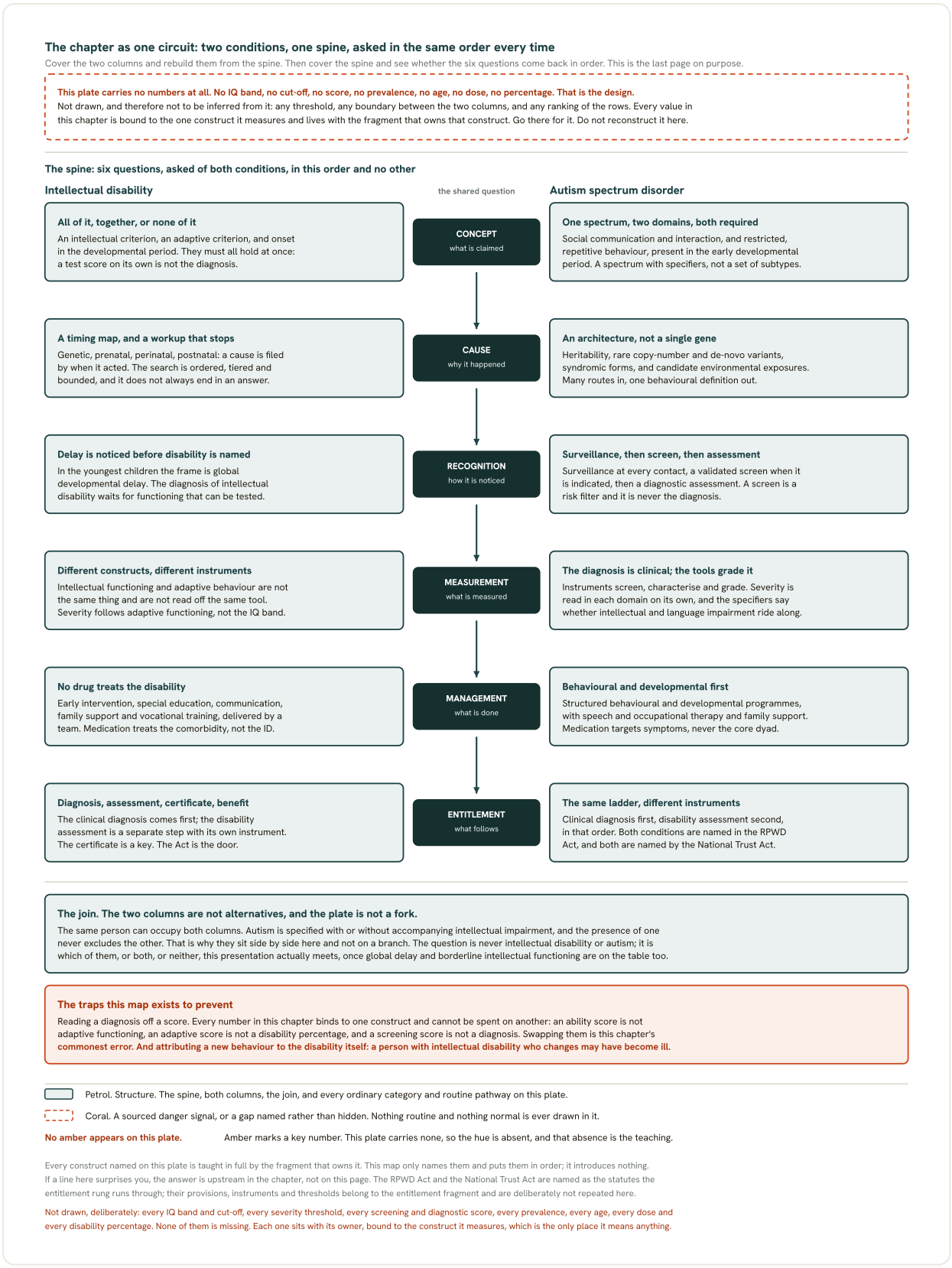


Fig 21.18 The chapter as one circuit. Intellectual disability and autism spectrum disorder set side by side across a shared spine of six questions, concept to cause to recognition to measurement to management to entitlement, asked of both conditions in the same order. The plate carries the shape of the chapter and no numbers at all, because every number in this chapter binds to the one construct it measures and lives with the fragment that owns that construct. The join band states the thing the side-by-side layout is there to teach, that the two columns are not alternatives and the same person can occupy both.

34.1 Start with the developmental question

The starting task is to establish the **developmental trajectory**: what was acquired, what is currently difficult, whether skills have been lost, and how the pattern affects everyday life. This makes the assessment developmental rather than merely behavioural. It also keeps current symptoms in relation to the person's communication, cognitive and social capacities.

Developmental history should be interpreted alongside the account in the Normal child development and milestones notes. If the presentation includes regression, unusual episodic events, altered sleep, pain, sensory difficulty or a change from baseline, the formulation must remain open to medical and psychiatric contributors rather than assuming that all change belongs to the neurodevelopmental condition.

RULE **Rule.** Begin with the person's developmental course and present functioning; use diagnostic categories to clarify the pattern, not to replace the formulation.

34.2 Separate constructs before combining them

Intellectual functioning, **adaptive functioning**, developmental level and the autism phenotype are related but not interchangeable. A cognitive assessment addresses an aspect of ability. Adaptive functioning asks what the person manages in ordinary settings. Autism assessment asks whether a characteristic pattern of **social communication** differences and **restricted and repetitive behaviour** is present. Each finding changes the meaning of the others, but none can be inferred from another alone.

CLINICAL TASK	QUESTION TO KEEP SEPARATE	WHERE TO RETURN FOR FULL TEACHING
Describe ability	What can the person understand, learn and do?	Intellectual disability definition, clinical assessment and IQ testing
Describe everyday function	What support is needed in usual environments?	Adaptive behaviour assessment and intellectual disability severity
Describe autism features	Is the social-communication and behavioural pattern characteristic of autism?	Autism diagnostic criteria, clinical features and diagnostic tools
Describe developmental concern	Is the available profile sufficient for a stable diagnostic conclusion?	Global developmental delay and differentiation of intellectual disability from autism, GDD and borderline intellectual functioning
Describe participation	What changes at home, in education, in work and in the community are required?	Management, family support, disability certification and benefits

TRAP **Examination trap.** Do not use an intelligence score as a synonym for adaptive functioning, or use either as proof for or against autism. They are different clinical constructs.

34.3 Read the profile, not only the label

A useful formulation gives a profile of strengths and vulnerabilities. It asks how communication is used, how social reciprocity is experienced, how routines and sensory demands affect participation, and how demands change behaviour. It records what helps the person engage, learn and regulate. This profile is often more useful for care planning than a diagnostic word alone.

- Describe the person's effective means of communication and the situations in which communication breaks down.
- Identify abilities that can be recruited for learning, self-care, relationships and participation.
- Notice environmental triggers, including mismatch between demands and capacity.
- Ask whether behaviour is communicating distress, pain, fear, overload, confusion or an unmet need.

This approach protects against **diagnostic overshadowing**: the error of attributing every new symptom or behaviour to an established developmental diagnosis. A change from baseline still deserves ordinary clinical curiosity. The full child psychiatric assessment battery, services and protection are set out in the Child psychiatry: assessment, treatment, services and protection notes.

34.4 Look actively for co-occurrence

Co-occurrence is clinically important because it can alter presentation, risk, family burden and the practical plan. Medical conditions, sensory problems, sleep disturbance, emotional symptoms, behavioural disturbance and psychiatric syndromes may make a familiar developmental profile look suddenly more difficult. The right response is not diagnostic proliferation; it is a disciplined search for treatable contributors and an account of how they interact.

When autism and intellectual disability coexist, describe each profile explicitly. When attentional symptoms or social difficulties raise the possibility of ADHD, use the ADHD and specific learning/communication disorders notes for the full assessment framework. Do not treat social difficulty as automatically explained by either condition.

MNEMONIC

FIND

is a practical closing mnemonic: **F**unction in daily life; **I**nteraction and communication profile; **N**eurodevelopmental course; **D**rivers of change, including co-occurring conditions and context.

34.5 Convert formulation into support

The endpoint of assessment is a plan that is intelligible to the person and family and usable across settings. State the priority difficulty, the strengths that can be used, the support required, the contributors that need treatment or further assessment, and the follow-up question. A plan framed around **support needs** is more clinically honest than one framed around a label alone.

- Link recommendations to a named functional difficulty rather than to diagnosis alone.

- Make family partnership part of the formulation, not an afterthought.
- Coordinate educational, communication, occupational, behavioural and mental-health input around shared goals.
- Review change over time, especially after a new stressor, treatment or transition.

Intervention models, speech and occupational support, family support, pharmacotherapy and prognosis are considered in their respective sections.

GOOD TO REMEMBER

Keep statutory processes separate from clinical formulation: certification may help access entitlements, but it does not replace an individualised account of function and care.

34.6 The final answer in practice and in examination

For a case discussion, write the problem representation before writing the diagnosis: developmental course, current profile, functional impact, possible co-occurrence, contextual modifiers and priority supports. This structure prevents the common drift from a complex clinical presentation into a label-led answer.

The must-remember synthesis is simple: diagnose carefully, describe function clearly, search for change and co-occurrence, then plan support around the person rather than the label.

Previously asked

Genuine past questions from this chapter's territory, with their board of origin. Only two examination questions in the catalogue fall inside intellectual disability and autism, and both are printed here. No question is reconstructed, paraphrased into a new one, or invented to pad the list.

QUESTION	TYPE	ASKED
Describe the etiology, clinical features and management of Fragile X syndrome	Long essay	MUHS 2013
Asperger's syndrome	Short note	MUHS 2012

TRAP **Asperger's syndrome no longer exists as a diagnosis.** A question asked under the older nomenclature still expects you to know where the construct went and why it was absorbed, not to defend it as a current category. The historical topic in this chapter carries that story.

The quick review

One table, read down the left column, cover the right. Every line is taught in full somewhere above; nothing here is new. Where the sources disagree, the disagreement is the answer.

ASK	HOLD
WHAT MAKES INTELLECTUAL DISABILITY	All three criteria: intellectual deficits, adaptive deficits, onset in the developmental period. Not an IQ score on its own.
WHAT GRADES ITS SEVERITY	Adaptive functioning across the conceptual, social and practical domains. IQ is supporting evidence, not the grade.
WHAT GRADES AUTISM SEVERITY	Support needs, rated separately for the two domains. Never IQ. Grading autism by IQ is the classic error.
GLOBAL DEVELOPMENTAL DELAY	The under-fives placeholder for a child too young or too impaired for a valid standardised assessment. It is a holding position, not a diagnosis to keep.
THE FOUR INSTRUMENTS ROUTINELY CONFUSED	They measure four different constructs: intelligence, development, social maturity, adaptive behaviour. Being asked for adaptive scales and listing all four is the exam error this chapter exists to prevent.
THE STATUTORY INSTRUMENT FOR INTELLECTUAL DISABILITY IN INDIA	Not the mental-illness disability scale. The routes are separate, and the chapter shows which is which.
THE AUTISM DYAD	Social communication, all of its sub-criteria; restricted and repetitive behaviour, the stated minimum of its four. Language is a specifier, not a criterion.
WHERE ASPERGER'S WENT	Absorbed into the spectrum. A past question under the old name still expects you to know why.
THE COMMONEST PSYCHIATRIC CO-OCCURRENCE IN AUTISM	ADHD. DSM-IV forbade the joint diagnosis; DSM-5 permits it. The frequency is contested and the chapter gives both figures.
DIAGNOSTIC OVERSHADOWING	A new behaviour in intellectual disability gets blamed on the disability and the treatable illness is missed. It is a reasoning failure, not a knowledge failure.
WHAT NO DRUG DOES IN AUTISM	Treat the core features. The licensed agents target irritability, and the monitoring burden is the price.
WHAT THE SOURCES REFUSE TO TELL YOU	Several cut-offs, several band tables and several Indian figures are simply not in the literature consulted. The chapter says so where it happens rather than inventing them.

References

Every specific in this chapter is bound to one of the sources below, and the count is the number of claims each carried. Books and guidelines are cited author, title and year in the house convention; no PMID is asserted for a book. Where two sources disagreed, the chapter shows both rather than choosing, and those disagreements are named in the text where they fall.

SOURCE	CLAIMS CARRIED
IPS CPG CAP CHILD	165

SOURCE	CLAIMS CARRIED
Tasman Psychiatry	119
Kaplan & Saddock's CTP 2024	118
Companion to Psychiatric Studies	107
DSM-5-TR-2022	98
New Oxford Textbook of Psychiatry	85
Shorter Oxford Textbook of Psychiatry	69
RC Jiloha - Forensic Psychiatry - An Indian Perspective	59
ICD-11 CDDR	58
Rutters Child and Adolescent Psychiatry 6E	47
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Dulcan's Textbook of Child and Adolescent Psychiatry	33
Maudsley 15th edition	33
Shorter Oxford Textbook of Psychiatry, ch 17	28
Kaplan & Sadock's CTP 2024	26
Kaufman's Clinical Neurology for Psychiatrists	23
61:254-69)	17
Kaplan & Sadock's CTP 2024, intellectual disability chapter	17
Tasman Psychiatry, ch 42	17
Clinical Methods in Psychiatry 1st Ed 2024, AIIMS Publication	16
IPS CPG CAP CHILD - Kishore et al., Indian J Psychiatry 2019	15
Geriatric Psychiatry Study Guide	12
New Oxford Textbook of Psychiatry, Chapter 24	12
IPS CPG CAP CHILD (Kishore et al.)	12
Developmental Psychopathology (Cicchetti)	11
Dulcan's Textbook	11

On what is not here. This chapter declares its gaps rather than filling them. Where a value a reader might expect is absent, the absence is deliberate and stated at the point of use: the source consulted did not carry it, and nothing was

supplied from memory to cover the silence.

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