Chapter 4 Assessment Principles and Diagnostic Criteria

"I didn't label my child. My child got a diagnosis so that he can get the help that he needs."

BIOLOGICAL MOTHER AND ADVISORY GROUP MEMBER

"Diagnosis has allowed me to shift the blame and sadness of my perceived shortcomings and redefine them with a new appreciation of what I have overcome and what I have managed to achieve despite them."

ADULT WITH FASD AND ADVISORY GROUP MEMBER

Chapter 4: Assessment Principles and Diagnostic Criteria

4.1 Assessment Principles to Support Application of the Diagnostic Criteria.

The following *Assessment Principles* are provided to support practitioners in applying the diagnostic criteria in practice:

- For those already diagnosed with FASD under previous criteria, re-assessment is only needed if clinically indicated.
- PAE can result in a wide range of whole-body outcomes from subtle to severe. In diagnosing FASD, the aim is to identify individuals who are experiencing pervasive, persistent, and clinically significant impairments that impact daily functioning.
- Assessment should include input from health professionals across multiple disciplines and be guided by value-based and person-centred care principles. This approach places the individual and their support network at the centre of care, fostering trust, mutual respect, and active engagement in decision-making.
- There is no formally agreed definition of impairment within, or between, health disciplines. As such, differences in functional performance and/or physical features evidenced by indices such as percentile ranks, should not be used in isolation. Clinical judgement informed by the available information is essential to determine the best explanations for an individual's presentation.
- Assessment should follow a 'developmentally informed approach'; whereby different assessment approaches are applied across developmental stages to provide the most appropriate assessment, given an individual's presentation.
- Assessment and diagnosis of FASD can and should take place across the lifespan. Individual attributes that may manifest as barriers to equitable inclusion may only become evident with age. Periodic Review should occur when clinically indicated, considering the supports in place, and the potential impacts of major life transitions on functioning.
- In providing a diagnosis of FASD, practitioners determining that an individual is impacted by a lifelong condition. This means impairments are not transient, due to changes in current circumstances or enduring environmental adversity. However, practitioners also need to consider how an individual may change over time due to life experiences and opportunities, formal supports, or the lack thereof, as well as changing expectations across life stages and contexts.
- Practitioners are encouraged to seek relevant discipline-specific professional development and clinical supervision, preferably from those with specific FASD expertise to support them in undertaking assessment and diagnosis in their specific settings, whilst also being mindful of professional and ethical guidelines.

4.2 Diagnostic Criteria

Diagnostic criteria aim to inform practitioners of the symptoms and signs usually required to ensure accurate diagnosis of a health condition, while also allowing a degree of flexibility to accommodate natural variances in presentation and clinical decision-making (WHO, 2004). Therefore, the following criteria do not form strict rules for diagnosis but provide evidence-based guidance to inform assessment, diagnostic reasoning, and case formulation.

Please note that <u>additional information</u> is provided in the sections following the diagnostic criteria to support implementation.

Fetal alcohol spectrum disorder (also termed neurodevelopmental disorder associated with prenatal alcohol exposure).

All criteria (A-E) must be considered, and all relevant specifiers applied for diagnosis.

- A. Evidence of prenatal alcohol exposure (confirmed by point 1 or 2)
 - 1. Prenatal alcohol exposure (PAE) above a low risk level at any time during gestation, including prior to pregnancy recognition. *See the additional information for further details to support assessment of PAE risk.* Confirmation of PAE may be obtained from any of the following sources: self-report of alcohol use in pregnancy, and/or collateral reports from individuals who directly observed the prenatal alcohol use, and/or information obtained from medical or other records.
 - 2. In the absence of a confirmed history of PAE, following the exclusion of other causes, the presence of the three sentinel facial features (i.e., short palpebral fissures, thin upper lip, and smooth philtrum) may be considered sufficient to meet Criterion A.
- B. Presence of pervasive neurodevelopmental impairments.

This is evidenced by clinically significant impairments in three or more neurodevelopmental domains (intellectual abilities, communication, motor skills, literacy and/or numeracy skills, memory, attention, executive functioning, emotional and/or behavioural regulation, adaptive/social functioning).

Clinically significant impairment is defined by points 1 and 2:

- Reports indicative of clinically significant developmental and/or behavioural problems as described by the individual undergoing assessment and/or multiple informants across different settings; and
- 2. Direct evidence of clinically significant impairments. Practitioners should use standardised tests where appropriate, but not rely solely on these tests in assessing the significance of impairments and functional impacts. *See further information below on defining clinically significant impairments*.

Note: In infants and young children, in the absence of direct evidence of clinically significant impairments, following exclusion of other causes, microcephaly ($\leq 3^{rd}$ percentile) may be used as an indicator of neurodevelopmental impairment, meeting criterion B.

C. The neurodevelopmental impairments result in functional impacts that necessitate significant supports across multiple areas of functioning, relative to an individual's developmental stage and cultural context.

D. The onset of neurodevelopmental impairments is evident during the developmental period

Note:

- Intellectual, behavioural, and functional capabilities emerge variably as individuals grow and mature, and some delays in development may represent age or developmentally appropriate diversity, rather than impairments.
- Neurodevelopmental impairments may not become apparent or fully manifest until the demands of life and context exceed developmental capabilities. Repeat assessments may therefore be required.
- E. An individual's presentation is not better attributed to another condition or exposure.

Diagnosis requires consideration of other conditions or exposures, which could better explain the person's presentation. However, some conditions and exposures can co-exist with FASD. This includes consideration of other neurodevelopmental risk factors such as, but not limited to:

- *Predisposing/familial* (e.g., family history of learning disorders, cognitive impairments, mental ill-health, intergenerational trauma).
- *Genetic conditions* (e.g., Fragile X, chromosomal variants including microdeletion or duplication syndromes, or single gene disorders that are known to be associated with neurodevelopmental impairment).
- *Prenatal* (e.g., exposure to other teratogens, including prescription medications [e.g., sodium valproate] and/or other drugs [e.g., nicotine, cannabis, amphetamines, opioids], pregnancy complications, congenital infections, premature birth, other environmental factors [e.g., nutritional deficiencies during pregnancy]).
- *Postnatal* (e.g., hypoxic ischaemic encephalopathy, adverse childhood, adolescent, or adult experiences, acquired or traumatic brain injury, central nervous system infections, or cranial malformation).
- Other neurological conditions (e.g., delirium, dementia, seizure disorders [e.g., genetic seizure syndromes [e.g., genetic epilepsy syndromes, developmental and epileptic encephalopathies], metabolic [e.g., mucopolysaccharidoses] or other neurocognitive conditions).
- *Current medications or substances* (i.e., the direct physiological effects associated with the use of medications or substances by the individual being assessed).

Specify the following physical features:

1, 2 or 3 or no sentinel facial features (include the specific measurements for palpebral fissure length (e.g., 10th [1.28 SD], 5th [1.65 SD], ≤ 3rd percentile [≤ 2 SD]).

- Head circumference restriction at birth and/or postnatally (e.g., at the 10th [1.28 SD], 5th [1.65 SD], ≤ 3rd percentile [≤ 2 SD]; include the specific measurements for head circumference at birth and postnatally).
- Physical size restriction at birth and/or postnatally (weight and/or length/height at the 10th [1.28 SD], 5th [1.65 SD], ≤ 3rd percentile [≤ 2 SD]; include specific measurements at birth and postnatally).

Note: These physical features provide clinically meaningful information and are an important part of the assessment. These features are not provided as specifiers to diminish their importance but because not all individuals will present with these physical features. This approach encourages practitioners to document these physical features along a continuum, informing both current and future clinical care and research.

Associated features: Record all the associated features including structural brain abnormalities, neurological conditions (e.g., seizures of unknown origin, cerebral palsy, hearing, or vision impairments), congenital anomalies (e.g., cardiac, renal, or other organ defects, ptosis, strabismus), musculoskeletal conditions, (e.g., flexion contractures), other health problems (e.g., sleep disorders, eating/feeding or toileting concerns), sensory processing challenges, social cognition impairments, social communication/pragmatics, motor speech or speech-sound impairments.

Co-occurring conditions: FASD can co-occur with a wide range of conditions. This includes but is not limited to other neurodevelopmental conditions (e.g., ADHD, ASD, language disorder, specific learning disorder) and mental health conditions (e.g., anxiety, depression, trauma and other stressor-related conditions, substance use conditions). Assessment should consider relevant co-occurring conditions to enable appropriate conceptualisation of an individual's treatment and support needs. When an individual is found to meet criteria for multiple diagnoses, care should be taken to consider the possible overlap of symptoms and whether multiple diagnoses assist in understanding the individual's needs.

At risk of FASD: In situations where PAE above a low risk level is confirmed and developmental concerns are identified, but available assessment is insufficient to determine if pervasive and clinically significant impairments exist, or assessment could not be completed due to a young child's capacity to engage in assessment, individuals may be considered 'at risk of FASD' with follow-up and reassessment recommended. Practitioners should specify why the 'at risk' designation has been used. This designation should not be used when neurodevelopmental impairments are present, and PAE is suspected, but has not been confirmed (see alternate diagnostic terminology below); or when an assessment and diagnosis are not possible due to limited resources.

Diagnostic terminology: There are different diagnostic terminologies available for the diagnosis of FASD and associated presentations. DSM-5-TR terminologies and codes include:

DSM-5-TR: Other Specified Neurodevelopmental Disorder (F88)

• Neurodevelopmental disorder associated with prenatal alcohol exposure. This is equivalent to a diagnosis of FASD and may be applied interchangeably.

DSM-5-TR: Unspecified Neurodevelopmental Disorder (F89)

This terminology could be applied for individuals who have clinically significant neurodevelopmental impairments, where PAE was not confirmed, and/or when an individual does not meet full criteria for any of the conditions in the neurodevelopmental disorders diagnostic class. This terminology could also be applied where individuals and families do not want to specify the prenatal alcohol exposure.

There are also terminologies included in the **ICD-10** (other congenital malformations - fetal alcohol syndrome [Q86.0] and **ICD-11** (fetal alcohol syndrome [LD2F.00]; other specified neurodevelopmental disorder [6A0Y] - neurodevelopmental syndrome due to prenatal alcohol exposure) that may be relevant for public health system coding requirements.

Individuals and families may have a preference to use these or other non-medical self-identifying terms (e.g., neurodivergent) that support their autonomy in defining their own identity.

Recognising the diverse perspectives on diagnostic terminology in Australia, and in alignment with the foundational considerations of these guidelines, it should be considered a right of an individual and their family to have choice and control over the terminology that is applied.

Α	Evidence of Prenatal Alcohol Exposure (PAE)	
	\downarrow \downarrow	
+	Confirmed PAE above low risk levels at any tine during gestation or Presence of 3 sentinel facial features	*Neurodevelopmental domains
В	Presence of pervasive and clinically significant neurodevelopmental impairments in 3 or more domains*	2. Motor skills3. Intellectual abilities4. Attention
+	↓ ↓ Direct evidence + H Reports from informants in different settings	 5. Memory 6. Executive function 7. Emotional and/or behavioural regulation 8. Literacy and/or Numeracy skills 9. Adaptive/social functioning
С	Neurodevelopmental impairments result in functional impacts that require significant supports	Note. In infants and young children, in the absence of direct evidence of impairments,
+		following exclusion of other causes,
D	Onset of impairments is during the developmental period	microcephaly may be used as an indicator of neurodevelopmental impairment.
+		
E	Presentation not better explained by other factors	
+		
Specifiers	Any sentinel facial features, head circumference and/or physical size restriction	



4.3 Additional Information

4.3.1 Structure of the diagnostic features, diagnostic specifiers, and associated features.

A diagnostic framework aligned with other neurodevelopmental conditions included in the DSM-5-TR was used to integrate the findings from the evidence review. Clinical features with sufficient evidence that must be present were included as diagnostic features. Clinical features with sufficient evidence that may or may not be present, were included as diagnostic specifiers. Other features without sufficient evidence but that may be present at higher rates in individuals with FASD were included as associated features. This structure reflects the heterogeneity of FASD presentations and provides an evidence-based framework adaptable to new evidence.

4.3.2 Criterion A: Prenatal alcohol exposure (PAE)

PAE is a key factor in differentiating FASD from other conditions. Practitioners need reliable evidence of PAE at levels that could lead to adverse outcomes.

- Risk and protective factors for harm need to be considered at all PAE levels.
- Increased risk for FASD is observed with increased exposure. However, no safe level of PAE has been established.
- The PAE standard drink levels from the evidence review were included to compare diagnostic outcomes at different exposure levels but should not be used as clinical cut-offs for diagnosis.
 - In the absence of quantifiable PAE, practitioners should consider available information to inform the assessment of risk. For example, biological parents may not be available to interview, or the biological parents may not recall precise details. However, other information, such as self-reported information, witness reports, or available records that document episodes of intoxication during the pregnancy, can inform risk assessment.
 - In such instances, after considering the reliability of the information (i.e., including the nature of the relationship between biological parent/s and witness reports), practitioners may exercise informed clinical reasoning about the PAE risk based on the best available information.
 - $\circ\,$ Practitioners are encouraged to engage in case discussion to support clinical decision making.
- Figure 9 provides additional information to support the assessment of FASD risk.

See the prenatal alcohol exposure assessment section of the <u>main guidelines document</u> for good practice statements and implementation considerations.

Also see the additional information <u>section below</u> on facial features and the medical assessment section of the <u>main guidelines document</u> to support implementation of Criterion A2.

Diagnostic Risk of FASD	No to Low Risk	Medium Risk	Medium to High Risk	High	Risk
AUDIT-C Scores	0-2	3-5	-	È	5
Evidence Review PAE Levels	"Light" (Up to 2 standard drinks/week; 20 grams of alcohol)	"Moderate" (>2-10 standard drinks/week; 21- 100 grams of alcohol)	Confirmed Unquantifiable	"Heavy" (>10-20 standard drinks/week; 101-200 grams of alcohol)	"Very Heavy" (>20 standard drinks/week; > 200 grams of alcohol)
Key Evidence Review Considerations	While there is evidence for potential adverse outcomes from PAE, there is a low likelihood of FASD diagnosis at this level.	There were mixed findings in the evidence review. There may be the potential for increasing levels of risk across this PAE level.	Increased risk of adverse FASD diagnostic outcomes at this PAE level, with most studies reporting 'heavy' exposure. However, lack of quantifiable PAE information limits conclusions.	Increased risk of adverse FASD diagnostic outcomes demonstrated in the evidence review.	Increased risk of adverse FASD diagnostic outcomes demonstrated in the evidence review.

Risk and protective factors need to be taken into consideration at all PAE levels. Increasing levels of risk for FASD are observed with increasing levels of exposure. There is no established safe level of PAE. The PAE levels from the evidence review were created to allow appropriate comparison of diagnostic outcomes between exposure levels and are not intended for use as clinical cut-offs for diagnosis. In the absence of quantifiable PAE clinicians should consider all available information to inform their assessment of risk.

Figure 9. Visual to support the assessment of risk for FASD.

Note. PAE = prenatal alcohol exposure. 1 standard drink = 10g ethanol. "Light" exposure level was determined based on clinical situations where people report having consumed no more than 1 to 2 standard drinks (SD) per week. The distinction between "moderate" and "heavy" exposure was based on the NHMRC Alcohol Guidelines (2020) determination of risky drinking (i.e., no more than 10 standard drinks per week). A pragmatic distinction was made to separate out the two higher levels of PAE to provide the opportunity to differentiate between "heavy" and "very heavy" exposure. Exposure may be **one or more** occasions during a week. A binge exposure pattern was included in the evidence review and may fall into "moderate", "heavy", or "very heavy" exposure categories depending on how many drinks were consumed on the **one or more** binge occasions per week.

4.3.2.1 Further details regarding the evidence review

To support assessment and diagnosis across a wide range of clinical contexts in Australia, including outside of specialist settings, feedback from the Clinical Advisory Groups indicated that practitioners would benefit from further guidance interpreting PAE risk. Consequently, an extensive evidence review was undertaken. To facilitate appropriate comparisons across the diagnostic outcomes, available evidence was quantified based on the grams of ethanol exposure per week and grouped into different exposure levels (as per Figure 6). However, several key limitations must be considered when applying this evidence in practice at an individual level:

- The review could not control for, or compare, different timings or patterns of exposure (e.g., chronic exposure, exposure only prior to pregnancy recognition, first trimester only exposure, or binge exposure). This was due to the variability in definitions, reporting, and the limited number of studies available assessing the same outcomes at the same PAE level.
- PAE assessment is typically based on self-report, which remains the most accurate method to assess PAE, due to lack of accuracy of currently available biomarkers and screening tools (e.g., for recent review see Kable and Jones, 2023). However, self-reported PAE information can have limitations, such as memory recall issues and under-reporting due to stigma.
- It is possible that a lower level of PAE at a critical period of gestation could result in adverse outcomes and practitioners need to use clinical judgement when assessing PAE risk.
- Although adjusted outcomes were used where possible, the review often could not control for, or compare, various individual, prenatal, parental, and child factors that may exacerbate or ameliorate the impacts of PAE (e.g., prenatal nutrition, metabolic rates, genetic factors, biochemical and inflammatory responses to alcohol).
- Similarly, although adjusted outcomes were used where possible, the review was often unable to control for, or compare, different individual postnatal, parental, and child factors, which may exacerbate or ameliorate the impacts of PAE (e.g., postnatal environments and traumatic events, postnatal nutrition).

For the full results, see the <u>Association between Prenatal Alcohol Exposure Physical size</u>, <u>Dysmorphology and Neurodevelopment: Systematic Review Report</u>

Refer to the prenatal alcohol exposure assessment *section of the main guidelines document for good practice statements and implementation considerations to further support applying Criterion A in practice.*

4.3.3 Criterion B: Presence of pervasive neurodevelopmental impairments

The evidence review indicated that PAE exposure increases the potential for adverse outcomes across all neurodevelopmental areas included in the diagnostic criteria, wither high levels of PAE associated with increased risk for adverse outcomes.

To demonstrate the pervasive nature and clinical significance of these impairments, there must be evidence that an individual's daily functioning across contexts is negatively impacted in multiple domains. As such, the Guidelines Development Group have retained the *three or more neurodevelopmental domains criterion*.

Importantly, as discussed in the risk and disease section of the <u>main guidelines document</u>, while PAE is a risk factor for neurodevelopmental impairments, it is not a predetermined outcome. Practitioners must recognise that having three or more neurodevelopmental domains with clinically significant impairments is neither specific to, nor discriminatory for, FASD, and a wide range of neurodevelopmental conditions must be considered. As such, practitioners will need to consider other possible factors that could explain or contribute to the observed neurodevelopmental impairments (Criterion E) and may need to apply a higher threshold for pervasive impairments in the presence of multiple comorbidities.

The Guidelines Development Group acknowledges that further research is needed to empirically validate criterion B.

4.3.3.1 Applying standardised tests in the assessment

Consistent with the 2016 Guide, Criterion B recommends using standardised tests as part of the assessment. While some of the tests listed in the previous Guide were included in the available evidence contributing to the evidence-to-decision framework outcomes, no studies focused on comparing the clinical utility of specific tests over others within the diagnostic process.

Feedback from the Clinical Advisory Groups indicated that the list of example standardised tests included in the 2016 Guide was potentially being applied rigidly, resulting in assessments that were not person-centred and culturally responsive.

It is widely recognised across professions that there may be circumstances where standardised tests are not appropriate. Some examples include (*note – non limiting list*):

- Individuals who are extremely low functioning, where standardised tests would not likely produce valid results, and may negatively impact well-being.
- Situations where practitioners in consultation with the individual or their family decide that the use of standardised tests are not culturally and linguistically appropriate.
- When assessment of a domain or use of a tool is not appropriate given the person's history, such as academic testing of a child who has not been in the education context for many years.

In such circumstances, practitioners are encouraged to exercise their professional judgement in the assessment process (including determining to not assess a domain) and to note any limitations to assessment and formulation that may result.

It is also important to reiterate that most normative studies of standardised tests do not include representatives from Australia's culturally diverse population. Therefore, caution must be exercised when using normative data to determine the presence of clinically significant impairments for individuals from different cultures to the population on whom the tests were developed and normed.

Therefore, based on the acknowledged limits to the broad application of tests and their normative data, the expert input from the Clinical Advisory Group, and the lack of evidence found in the current review, the Guidelines Development Group determined that specifying examples of standardised tests was not appropriate. This position is broadly supported by professional representative bodies both in Australia and internationally through their respective Codes of Conduct, Codes of Ethics, and ethical or practice guidelines on the use of psychometric tests, which in summary direct practitioners

to understand the theoretical basis, psychometric properties, and other influences on utility when selecting and using tests and measures in their clinical practice.

The Guidelines Development Group recommends that practitioners apply their discipline specific knowledge, professional expertise, and clinical judgement to determine the most appropriate approaches for examining the individual within the context of the assessment.

4.3.3.2 Determining the clinical significance of neurodevelopmental impairments

There is no universally agreed formal definition of "impairment" (see Assessment Principles section for discussion), and no test, or score can unequivocally determine the presence of an impairment. As such, to decide if clinically significant impairments are present and whether they should contribute to a diagnosis, practitioners are required to consider all the information collected during the assessment. A percentile range is provided to support diagnostic decision-making (i.e., scores Below Average – Exceptionally Low Scores may be indicative of clinically significant impairments; Table 3), but practitioners should be mindful of the following aspects:

Interpreting Standardised Tests

When considering the results of standardised tests, practitioner are reminded that:

- "Scores cannot be impaired; only a function can be impaired" (Guilmette et al., 2020, p. 442); therefore, single test scores do not equal impairment and should not be used in isolation to define impairment, but rather in combination with functional correlates; and
- While tests may contribute to multiple domains due to the connection with various aspects of functioning, a single test score or construct (e.g., attention, working memory, communication) should not be used to establish impairments in multiple neurodevelopmental domains.
- It is the responsibility of the practitioner to understand the theoretical basis of the tests and apply an individualised formulation process to interpret test results and decide how particular test scores and constructs are counted across the neurodevelopmental domains.

Percentiles

Percentiles are a simple and popular metric for interpreting and conveying assessment outcomes. However, practitioners should be familiar with the relevant considerations and challenges in relation to interpreting percentiles in clinical practice (Crawford et al., 2009). Appendix C of the <u>main</u> <u>guidelines document</u> provides a brief overview of some key considerations for using percentiles.

Cut Scores

The *Standards for Educational and Psychological Testing* (American Educational Research Association et al., 2014) lay the foundational requirements for the development of many widely applied standardised tests used in clinical work across the professionals who may contribute to the FASD diagnostic process. Standards 5.21 through 5.23 specifically address the nuances of developing and applying test cut scores. Readers are directed to this resource to further their understanding.

Beyond the requirements of the above Standards, several other authoritative professional groups have addressed the use of cut scores and the interpretation of test scores more generally (non-exhaustive example list below).

- American Psychological Association Task Force on Psychological Assessment and Evaluation Guidelines: Guidelines 5 through 8 (American Psychological Association, 2020).
- International Guidelines for Test Use: Guideline 2.7, particularly sub-point 2.7.9 (International Test Commission, 2011).
- CATALISE: A multinational and multidisciplinary Delphi consensus study: Identifying language impairments in children: Consensus statement 12 and associated supplemental material (Bishop et al., 2016).
- Ethical guidelines for psychological assessment and use of psychological tests: Guideline 10 (Australian Psychological Society, 2014).
- International clinical practice recommendations on the definition, diagnosis, assessment, and intervention of developmental coordination disorder: Recommendations 11, 12 and 13 (Blank et al., 2019).

Practitioners are encouraged to review and consider their discipline specific and relevant other discipline and interprofessional guiding principles in the application of cut scores and exercise their informed professional judgement in the application of these to the FASD diagnostic process.

The process for determining cut scores, particularly in high stakes decisions (i.e., determining the presence or absence of a diagnosis) relies on applying at least one of several processes, all of which are well informed clinically, technically, empirically, and statistically (for thorough review of the various processes options for developing cut scores see Cizek & Bunch, 2007). While the 2016 Australian FASD Guide specified that equal to or less than the 3rd percentile or 2 standard deviations below the mean was a suitable cut-off for designating severe impairment in a neurodevelopmental domain; explanation of the rationale and process used to establish that cut-off in the diagnosis of FASD was not provided.

Demonstrating the diagnostic meaningfulness for clinical cut-offs requires evidence that there are differences in important life outcomes between people above and below that cut-off. The body of evidence investigating associations between PAE and neurodevelopmental outcomes considered in the current GRADE process provided no evidence to support the clinical validity of specific percentiles or standard deviation cut-offs. Until such evidence becomes available, the Guidelines Development Group determined that the interpretation of test scores to characterise impaired functioning is better informed by:

- 1. The practitioner exercising their clinical reasoning anchored in consensual expert guidance and/or best practices that apply to test interpretation in their specific professional field.
- 2. An integrative analysis of the whole person, conducted by practitioners who exercise their professional expertise in synthesising relevant historical, cultural, medical, and allied health, behavioural and other information into evidence-based clinical formulations.

Note. Points 1 and 2 are drawn from Guilmette et al (2020).

As per Table 3, test scores in the Below Average and Exceptionally Low Score Ranges could be considered significantly below the normative level and may be indicative of impairment.

Standard score	Percentile	Score label
<u>></u> 130	<u>></u> 98	Exceptionally high score
120–129	91–97	Above average score
110–119	75–90	High average score
90–109	25–74	Average score
80–89	9–24	Low average score
70–79	2–8	Below average score
<70	<2	Exceptionally low score

Table 3. Test score labels based on standard scores and percentiles for tests with normal distributionstaken from Guilmette et. al (2020)

The Guidelines Development Group considered this to be a reasonable guide but noted that the table likely does not apply for tests that have non-normal score distributions. These categories may vary by a few or several standard scores or percentiles depending on the specific nature of a test's score distribution.

Given the complexity in interpreting test scores, it is recommended that practitioners consult the manuals and relevant psychometric research for all tests used in the diagnostic process to ensure that the characterisation of an individual's performance aligns with established best practices and naming conventions for interpreting test results.

Confidence Intervals

All standardised tests, produce scores that contain both the individual's true ability, plus measurement error. To account for the uncertainty introduced by measurement error, most tests provide confidence intervals for subtests/domains, index, and full-scale/general scores. Some also provide confidence intervals for percentiles. Where confidence intervals are available or can be calculated, practitioners should use them together with the suggestions in Appendix C of the <u>main</u> <u>guidelines document</u> to support interpretation.

4.3.3.3 Assessing neurodevelopmental domains in practice

FASD is a complex and multifaceted condition best assessed and diagnosed via an interprofessional framework. Practitioners in multidisciplinary settings should not contribute isolated assessment findings, but contribute to all domains, bringing their relevant scope of practice to the assessment process and collaborating in case formulation.

Ideally, specific disciplines will bring their unique expertise to the assessment of certain domains (e.g., speech pathology assessing communication, occupational therapy or physiotherapy assessing motor skills). However, in settings where all disciplines are not available, practitioners can still work within their qualifications, training, and experience to provide assessment and formulation within their scope of practice. Upskilling to develop interdisciplinary skills can also be beneficial. Practitioners working in isolation or in limited multidisciplinary contexts are reminded that external

consultation and supervision are helpful approaches to supporting sound diagnostic assessment and formulation.

While a comprehensive assessment likely provides the greatest support to the individual, practitioners are reminded that assessment of all domains is not always required to consider a diagnosis of FASD. For further discussion see the Holistic Developmental, Functional and Wellbeing Assessment section of the main guidelines document.

An overview of the neurodevelopmental domains and specific considerations for assessment are provided in Table 4. Descriptions and assessment considerations for the domains are provided based on the results of the evidence review, discipline specific guidance from the Clinical Advisory Groups, and consultation with the Guidelines Development Group.

Assessment of infants and young children

Consistent with the principles underpinning these guidelines and good clinical practice, practitioners should consider the appropriateness of all assessment components to the individual infant or young child and their family. Given the limited availability of standardised tests for this age group, young children with microcephaly and three sentinel facial features may meet criteria for FASD, provided other causes are excluded. While standardised tests may not be available across all domains, practitioners can still have access to a range of clinical information regarding current development to consider alongside microcephaly in infants and young children to inform diagnostic decision-making. There is also the option of assigning 'at risk of FASD' in sufficient information is not available. See the <u>at risk of FASD</u> section below for further information.

Consideration of co-occurring conditions

Diagnoses of co-occurring conditions (e.g., ADHD, ASD, anxiety, depression) have not been included in the neurodevelopmental domain table (Table 4). Feedback from the Clinical Advisory Group indicated that including these as part of the domain table may unintentionally lead to a 'tick box' approach to diagnosis. Pre-existing diagnoses can provide helpful information regarding current functioning and should be considered when reviewing the available evidence. Practitioners are encouraged to evaluate an individual's functioning in each of the neurodevelopmental domains based on all the available information and determine if there are clinically significant impairments.

See the co-occurring and differential diagnosis section of the <u>full guidelines document</u> for further information.

Domain	Definition	Specific assessment considerations
Communication (Language skills)	Communication involves receiving and convey ideas, thoughts, and feelings to others. Language skills refer to the words, syntax, morphology, and pragmatics we use understand and communicate in oral, sign, and written forms. The domain focuses on language as a developmental process that can be disrupted by PAE. Although language skill development is sensitive to a range of factors (including other exposures, absence of modelling, hearing difficulties) it can also be disrupted idiopathically. Currently there is no clear phenotype for disordered language skills in the presence of PAE. Therefore, the domain should be assessed according to best practice recommendations. There is limited evidence that other communication disorders (e.g., motor-speech, speech sound, pragmatic/social communication, and voice disorders) are associated with or attributable to PAE. Therefore, such communication disorders will not solely contribute to a FASD diagnosis but are important to the overall clinical profile and treatment of a client and should be characterised and	 Impairment is present in this domain if the individual's language skills are found to be <i>disordered</i>. Assessment should follow best practice principles (Bishop et al., 2016; Bishop et al., 2017), specifically: Consider that disordered language skills are heterogenous and a thorough assessment should examine the principal dimensions of language: Syntax/morphosyntax Word finding and semantic knowledge Discourse/narrative Phonology (where indicated and considered linguistic in origin, though phonology should not solely contribute to meeting the criteria) Verbal learning/memory (if best attributable to communication skills rather than memory abilities). Consider functional language skills as part of the assessment (e.g., how the person performs in everyday meaningful tasks). For assessment involving Aboriginal and Torres Strait Islander peoples and other culturally and linguistically diverse individuals, use relevant Practice Guidelines produced by Speech Pathology Australia to guide practice.

Table 4. Overview of neurodevelopmental domains, definitions, and specific assessment considerations.

	documented in reports, with recommendations made as appropriate.	•	Evaluate the prognostic indicators for poor outcomes resulting from disordered language skills. If an individual meets criteria for FASD and disordered language is identified, the appropriate diagnosis relating to language disorder is 'Language Disorder associated with FASD' (as per Statement 6; Bishop et al., 2017). Diagnostic terminology should not distinguish between 'expressive' and 'receptive' diagnostic subtypes, as these categories are not considered stable over time (Bishop et al., 2017).
Motor skills	Motor skills include general motor abilities, areas of fine motor, gross motor, graphomotor (handwriting) skills, and/or visual motor integration.	•	Assessing more than one aspect of motor skills is recommended to understand of strengths and challenges in this domain. Assessment could commence with understanding the area of functional motor concern. A dynamic performance analysis can be undertaken to understand where the breakdown in performance is occurring and help select the most appropriate standardised test or additional functional assessments required. Consider performance on standardised tests as well as within a functional context (e.g., handwriting within the classroom, gross motor skills moving around a playground). Gross motor impairment may not be detected without a comprehensive assessment of gross motor skills. Ensure that an impairment in visual motor integration is due to a motor deficit and not a visual spatial deficit. Graphomotor tasks require learned skills and need to be assessed in relation to opportunity and only after access to relevant intervention.

		 Consider other causes of motor challenges, such as dysfunction the vestibular system, executive function, musculoskeletal system or peripheral nervous system.
Intellectual abilities (Cognition)	Practitioners should apply generally accepted models of intelligence, which is often defined to include the capacity for abstraction, to solve problems, and acquire new skills. As there are multiple models and definitions in current usage, practitioners are recommended to consider the implications of the model they select and maintain their knowledge of this area.	 Impairment in this domain may be established through deficits an underlying general factor of intelligence ('g' e.g., full-scalintellectual quotient) or one or more major subdomains that load on this factor according to established models of intelligence Examples include Verbal Comprehension, Visual Spatial Inder (visual perception), Fluid Reasoning, Working Memory, ar Processing Speed constructs as defined in the Wechsler paradig or broad and narrow constructs as defined by the Cattell-Hor Carroll Model. Assessment may be limited to nonverbal measures, whe appropriate. Practitioners should consider the impact of any language impairments (or if English is not the dominant language) or measures that include verbal instructions or responses. Practitioners are advised that while discrepancy analysis forms critical part of interpreting test scores in co-normed test batteried discrepancies in test scores are not sufficient in and of themselve to demonstrate impairment. Working memory could be included in either this domain or that attention or executive functioning domains depending on wheth the scores are considered more strongly associated with the scores as the strongly associated with the scores as

	performance on tests of general intellectual functioning or with the individual's attention and executive functioning performance.
Attention Generally considered the cognitive skill that connects sensory activity with mental processing (Posner & Petersen, 1990), attention is a complex cognitive activity with strong influences both to and from other cognitive skills, particularly working memory, and executive function. As such, it affects every aspect of what we do and experience (McDowd, 2007). At an operational level, attention has been characterised as a filter (Wickens, 2021) or selection (Angelopoulou & Drigas, 2021) mechanism for information from the environment that when operating effectively admits only relevant information to the task at hand for further processing. Other theories have operationalised attention as consisting of alerting, orienting, and executive control functions (Posner & Petersen, 1990), or modality-specific, bottom-up modulation or top-down modulation functions (Mesulam, 2000). Practitioners should consider relevant models of attention when constructing and interpreting results.	 There are many models of attention, which may place differing degrees of emphasis on indirect (e.g., questionnaire) and direct measures of attention. Models derived from both sets of measures may be considered under this domain, although factors which also fall directly under the definition of intellectual or executive functioning should be considered within those domains instead. Depending on the individual's presentation during the assessment of attention and their performance on language skills, memory, and executive function assessment, more basic attentional processes (i.e., visual scanning, immediate attention span) could be considered as part of the attention domain, while more complex attention processes, which require coalition of multiple abilities including attention and executive functioning (e.g., inhibition, dividing, shifting/switching) could be considered as contributing to other domains (i.e., executive functioning, communication, memory, literacy/numeracy) as appropriate. Challenges with visual scanning could indicate problems with oculomotor control, which could be further explored if clinically indicated. Consider the potential impact of prescribed medications (e.g., stimulants), level of engagement/rapport, and whether formal testing was conducted in a quiet room without distractions.

over prolonged periods of time.
Attention switching: alternating focus and resources between different tasks or sources of information.

• Sustaining attention: maintaining focus to a task

Several sub-skills have been proposed across the various attention models and theories. The following

 Selective attention: focusing on one source of information for processing and not processing other sources of information available in the

may be useful characterisations of attention:

environment.

• Divided attention: processing more than one source of information at a time or performing more than one task at a time by sharing capacity between them.

Attention encompasses both auditory and visual modalities. The available evidence for the impact of PAE did not demonstrate differences between auditory and visual attention. Therefore, it is advisable to assess attention using the method most appropriate for the individual.

Memory	Memory includes the ability to encode, store and retrieve information. It is traditionally conceptualised as including declarative (explicit) and procedural memory. Explicit memory may be further subdivided by modality (verbal, visual) or by the type of information stored, including episodic memory (personal events and experiences) and semantic memory (factual information; Mujawar et al., 2021). The available evidence for the impact of PAE on memory did not include procedural/implicit memory tasks or separate the impact of PAE on different stages of memory (encoding, storage, retrieval). However, a comprehensive memory assessment should evaluate these capabilities to provide a thorough understanding of an individual's memory challenges, to identify memory disorders, and inform targeted supports.	•	Memory may be assessed through performance on free recall, cued recall (immediate, delayed), and recognition tasks. Consider the interplay between attention, language skills, intelligence, executive functioning, anxiety, and memory. Based on test performance determine the best explanation for impairments. Consider self or informant reported memory abilities across settings (including but not limited to home, education, work, and community), to accurately represent any deficits and their functional impacts. It may be appropriate to assess prospective memory (i.e., remembering to perform a specific action in the future, at a particular time, or in response to a specific event) to assist in understanding an individual's day-to-day functional memory problems. However, practitioners should consider the multi- dimensional nature of this ability, including the impacts of executive function (e.g., Ji et al., 2021; Martin et al., 2003).
Executive Function (EF)	There are multiple different definitions of EF, with no universally accepted conceptualisation. EFs are traditionally defined as a set of higher-order cognitive functions, including initiation, inhibition, mental flexibility, novel problem solving, planning, emotion regulation, and self-awareness, all of which are needed for adaptive goal-directed functioning (Sira & Mateer, 2014).	•	Capabilities and deficiencies in EF are best captured through a combination of standardised tests, domain specific questionnaires, and semi-structured interviews. Consider performance across settings (including but not limited to home, educational settings, work, and social engagement), to accurately represent any deficits and their functional impacts. Individuals with severely impaired EFs may have limited insight into their difficulties and may not be able to accurately report their level

of functioning. In such instances, convergent information from a reliable informant should be sought (e.g., via questionnaires). • For older children, adolescents, and adults, EFs are generally considered multi-factorial, including different inter-related and inter-dependent skills that act within an integrated top-down control system. • For young children, some research indicates that EFs could be considered as a unitary concept that differentiates as children age (i.e., distinct EF abilities have not developed yet). There is discrepancy in available research regarding the specific ages at which differentiated EF skills emerge (e.g., varying from 6 to 12 years). Clinical judgement is required to determine if multicomponent assessment of EF skills is beneficial, based on an individual's presentation. For assessment and formulation purposes, practitioners may find it helpful to distinguish between hot (i.e., reward or affect-related, high emotional arousal during decision-making) versus cold (i.e., purely cognitive, no affective component) domains of EFs. There are many abilities that fall under the *cold EF* umbrella; however, core skills are better assessed by formal tests and include (and are not limited to): response inhibition (e.g., inhibitory control), cognitive flexibility, updating (i.e., self-monitoring, working memory), shifting (i.e., switching flexibly between tasks or mental states), planning and problem-solving. Hot EFs, can include processing of information related to reward, emotion, and motivation, and can be better assessed via clinical history, questionnaires, or direct observation (Salehinejad et al., 2021).

		• Depending on assessment results, emotion driven (reward, arousal, affective based) behaviours may be considered under the behavioural regulation domain.
Emotional and/or behavioural regulation	 Emotional and/or behavioural dysregulation could include significant difficulties with any of the following: Mood: internalising symptoms such as depression or anxiety, negative affect, suicidal ideation) Emotional regulation: irritability, low frustration tolerance, mood lability, suicide threats, where this is not the direct impact of another aetiology). Behavioural regulation: externalising behaviours could include rule-breaking behaviour (e.g., confabulation, taking things that belong to others), oppositional/non-compliant, behavioural outbursts, and reactive aggression. 	 The frequency, intensity, severity, and duration of the behaviour must be disproportionate and/or inappropriate for the context and developmental age of the individual. The behaviour must be persistent over time and across contexts, though may present differently due to the nature of specific contexts. The behaviour must not only occur in response to specific life circumstances and/or current substance use. When required, re-assessment can be recommended to determine whether behaviours are persistent. Consider the individual's history to identify the best explanation for the current presentation (e.g., family history, postnatal exposures, and adverse childhood experiences). Parental substance use may be associated with an increased genetic and environmental risk for emotional and behavioural regulation problems. Consider whether the individual has had access to evidence-based treatments and how well they have responded. Involvement with the justice system should not be used as direct evidence of significant impairment in this domain as a variety of criminogenic factors could be involved that are not related to an individual's impairments. Emotional/behavioural regulation impairments should only be considered diagnostically when there is sound evidence to suggest

Literacy and/or Literacy refers to reading, writing, and spelling skills and numeracy refers to mathematics skills. • This domain should only be considered towards a diagnosis when individuals have had access to appropriate engagement in formal education and remediation in the learning environment, in a				they are due to the direct effects of PAE or secondary effects of the disabilities that have arisen from PAE.
 language in which the individual is fluent and when the person has not significantly benefitted from attempts at remediation. Consideration must also be given to an individual's educational placement (e.g., mainstream, educational support class, special school) and opportunities (e.g., remote location, multi-lingual setting, new immigrant) and the type and level of supports provided. It is possible that impairments in literacy and/or numeracy could be a direct consequence of PAE or a functional consequence of the combined impacts of impairments in other neurodevelopmental domains (e.g., intellectual abilities, communication, attention, memory, executive function). As such, practitioners must carefully consider whether literacy and/or numeracy deficits independently contribute to the person's neurodevelopmental profile when formulating against the diagnostic criteria. For example, if significant attention impairments are identified it is recommended, they are treated before retesting to determine if impairments in literacy and/or numeracy and as present. 	Literacy and/or Numeracy skills	Literacy refers to reading, writing, and spelling skills and numeracy refers to mathematics skills.	•	This domain should only be considered towards a diagnosis when individuals have had access to appropriate engagement in formal education and remediation in the learning environment, in a language in which the individual is fluent and when the person has not significantly benefitted from attempts at remediation. Consideration must also be given to an individual's educational placement (e.g., mainstream, educational support class, special school) and opportunities (e.g., remote location, multi-lingual setting, new immigrant) and the type and level of supports provided. It is possible that impairments in literacy and/or numeracy could be a direct consequence of PAE or a functional consequence of the combined impacts of impairments in other neurodevelopmental domains (e.g., intellectual abilities, communication, attention, memory, executive function). As such, practitioners must carefully consider whether literacy and/or numeracy deficits independently contribute to the person's neurodevelopmental profile when formulating against the diagnostic criteria. • For example, if significant attention impairments are identified it is recommended, they are treated before retesting to determine if impairments in literacy and/or numeracy are also present.

Adaptive/social functioning	Effective adaptive and social functioning requires a collection of learned skills that enable people to function in their daily lives according to cultural and societal expectations. This can include understanding concepts of money and time, activities of daily living (personal care), occupational skills, safety, health care, travel/transportation, schedules/routines, interpersonal skills (e.g., quality of peer relations and challenges in social interactions), social responsibility, gullibility, naivety, suggestibility, or social problem solving.	 Consider any formal and informal supports the person may be receiving and how this may influence ratings of their adaptive/social functioning. Take into account different expectations and skills required at different developmental stages. Consider the level of exposure to different adaptive and social opportunities and differences that can exist across different communities (e.g., urban vs rural and remote settings). Utilise direct functional assessments of adaptive and social skills, as well as informant rating scales. Evaluate the functional impacts of language skills and pragmatic language skills on social functioning and social problem-solving abilities.
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4.3.3.4 Neurodevelopmental domains: evidence for inclusion

Inclusion of domains was based on review of the best available evidence (see the <u>Association</u> <u>between Prenatal Alcohol Exposure Physical size, Dysmorphology and Neurodevelopment:</u> <u>Systematic Review Report</u> for further details). For inclusion, the available evidence had to demonstrate an association between PAE and the neurodevelopmental outcome. Areas not included in the neurodevelopmental domains following review of the evidence were: social cognition, social communication/pragmatics, motor speech impairments, speech-sound impairments, voice disorders, sensory processing, neurological conditions, and seizures. Whilst these areas can still be assessed to inform support needs and can be documented as 'associated conditions', they are not included as part of the diagnostic criteria as further research is needed.

Wherever possible, adjusted outcomes were used that incorporated consideration of confounding variables. However, the available neurodevelopmental evidence did not often include adjusted outcomes. As such, the available evidence often did not exclude the impact of other factors that may influence neurodevelopmental outcomes. To provide additional examination of the evidence, a summary of the studies that included regression analyses was undertaken (results provided in the *Association between Prenatal Alcohol Exposure Physical size, Dysmorphology and Neurodevelopment: Systematic Review Report*). Overall, the pattern of results was generally consistent, whereby after controlling for confounding variables, results remained significant only at higher levels of PAE.

Extensive feedback was received from the Clinical Advisory Groups and discussions were undertaken in the Guidelines Development Group regarding the conceptualisation of the neurodevelopmental domains. The complex interplay between neurodevelopmental domains was thoroughly discussed. Detailed information is provided in Table 4 to support practitioners in considering the complex interplay between neurodevelopmental domains in the formulation process.

Creating higher-order groupings of the domains (e.g., as per the proposed DSM-5 criteria) was considered and discussed. However, it was decided this would introduce another arbitrary element to the diagnostic criteria, which would not currently be evidence based and may lead to the exclusion of certain presentations from this type of grouping system. It was determined that it is better for practitioners to undertake these conceptualisations at the individual case formulation level. Additionally, the possibility of splitting the adaptive and social domain was discussed, however it was determined that further research is required to inform decision making in this area.

The conceptualisation of each of domain was reviewed and updated based on available evidence and discipline specific best practice recommendations. A notable change is the previously termed 'affect regulation domain,' which is now 'emotional and/or behavioural regulation.' The available evidence was based on self and informant reports, with the most commonly available measure being the ASEBA Child Behaviour Checklist and Teacher Report Form. Thus, the available evidence focused on symptomatology not presence of psychiatric conditions. Updates were also made in the Communication (Language) domain to align with

best practice recommendations produced by the CATALISE consortium (Bishop et al., 2016, 2017). This included, for example, discerning areas/dimensions of language difficulty and removal of references to subtypes of language disorder (i.e., expressive/receptive). The previously named 'academic achievement' domain is now termed 'literacy and/or numeracy' to more specifically communicate the impairments considered in this domain (i.e., to clarify that this is not related to general behaviour/functioning in educational settings).

4.3.4 Criterion C: The neurodevelopmental impairments result in functional impacts that necessitate significant supports.

It is important to demonstrate the connection between neurodevelopmental impairments, impacts on functioning, and the need for supports. As with other neurodevelopmental diagnoses, practitioners must use their clinical judgement to determine if a significant level of support is required, given the individual's level of impairment. As stated in the DSM-5-TR, assessing whether this criterion is met, is an inherently difficult clinical judgement. Information from the individual, family members, and other informants is necessary. Care should be taken to ensure that this determination is based on the level of impairment and not due to other contextual factors (e.g., family, school, or community factors that affect functioning).

4.3.5 Criterion D: Onset of neurodevelopmental impairments in the developmental period

Criterion D refers to the recognition that impairments are present during infancy, childhood, or adolescence. The Guidelines Development Group want to ensure that this criterion does not impact on adults accessing assessment and diagnosis. This criterion should not be interpreted to mean that specific assessment results are required from the early developmental period for diagnosis of adults. Rather, it means that the overall pattern of available evidence indicates impairments were present in early development. Impairments are, therefore, not a decline in abilities or due to specific life circumstances or events. Information from previous assessments can be used as support for Criterion D if available.

4.3.6 Diagnostic Specifier: Sentinel facial features

4.3.6.1 Inclusion of three sentinel facial features

The review of current diagnostic criteria (overview of findings included in the Administrative and Technical Report [hyperlink to be inserted once available online]) indicated that nearly all current diagnostic criteria only permit diagnosis without confirmed PAE in the presence of three sentinel facial features. The two diagnostic criteria that included two facial features (i.e., Revised IOM and CDC) stated that criteria had been changed to two facial features to improve the sensitivity of diagnosis. However, no evidence was cited to support this decision. No studies identified through the evidence review provided support for a change from three facial features to two facial features. Future research is required to further understand the potential diagnostic utility of such a change. The inclusion of facial features as a diagnostic specifier aims to support documentation of facial features along the full continuum, enabling detailed assessment, monitoring, and future evaluation.

4.3.6.2 Palpebral fissures

Short palpebral fissures are defined at $\leq 3^{rd}$ percentile (i.e., ≤ 2 SD). Due to limited evidence, comparison across different percentile cut-offs was not possible. The Guidelines Development Group also considered current implementation factors, noting that most practitioners in Australia currently use the University of Washington facial analysis software, which applies $\leq 3^{rd}$ percentile definition of short palpebral fissures. Thus, changing this definition without appropriate tools to support practice could create significant barriers. Importantly, as discussed in the assessment principles section, clinical cut-offs are arbitrary, as physical features occur on a continuum. The inclusion of facial features as specifiers aims to enable practitioners to document the continuum of the facial features.

Due to the small number of studies and lack of reporting on the normative charts used in the available research, the evidence review could not examine the impacts of different palpebral fissure reference values on diagnostic outcomes. Limited has compared available palpebral fissure normative charts. In a retrospective comparison of U.S FASD clinical data, Astley Hemmingway et al. (2019) observed that switching to the Clarren charts from 6 years of age resulted in an artificial decrease in short palpebral fissures. In the only Australian study to examine this, Tsang et al. (2017) found that the Strömland et al. (1999) norms were the best fit from the norms available for a sample of Aboriginal children from one Australian community. Overall, there is very limited research, particularly in the Australian context regarding the assessment of facial features. This is an area that needs to be addressed in future research. Based on the limited evidence available, the Strömland palpebral fissure length charts are recommended for use across the lifespan.

4.3.6.3 Lip and philtrum

The University of Washington lip/philtrum guides were most commonly used in the available research evidence and are recommended for continued use. Practitioners should use clinical judgement to decide which lip/philtrum guide is most applicable based on the individual's physical features (i.e., Guide 1 Caucasians or combination of ethnicities with features most similar to Caucasians, or Guide 2 African American or combination of ethnicities with features more similar to African Americans). As per the palpebral fissures section, there is a lack of locally developed lip/philtrum guides, and the appropriateness of these tools for the Australian context is an important consideration for future research.

See the medical assessment section of <u>the main quidelines document</u> for further good practice statements and implementation considerations to support facial features

assessment in practice, including hyperlinks to access the University of Washington diagnostic tools.

4.3.6.4 Assessment of facial features for individuals from culturally diverse backgrounds

Concerns were raised regarding the lack of local palpebral fissure norms and lip/philtrum guides for the assessment of people from diverse ethnic backgrounds, including Aboriginal and Torres Strait Islander peoples (e.g., see Hayes et al., 2022). Future research is urgently required to develop local norms and tools relevant to the Australian context to improve the assessment of facial features. The Cultural Advisory Group recommend practitioners use shared decision-making with individuals and families attending for assessment to provide information about the limitations of current approaches to facial features assessment available in Australia.

Individuals can still be assessed and diagnosed with FASD without assessment of facial features. The wording of Criterion A.2 that facial features "may be considered sufficient" is to reflect that inclusion of facial features in Criterion A is not a requirement for diagnosis if not deemed appropriate, following consultation with individuals and families.

4.3.7 Diagnostic Specifiers: Head circumference and physical size restrictions

Based on review of the best available evidence, physical size $\leq 10^{\text{th}}$ percentile (i.e., weight, height/length, and head circumference) is included as a diagnostic specifier. However, as noted in the diagnostic criteria it is recommended practitioners report specific measures, including the 5th and 3rd percentile ranges, to capture the full continuum of these physical features. As described in the good practice statements in the medical assessment section, it is important to consider measurement error, interpretation of norm charts in the context of ethnicity, and assessments over time (where available) to avoid applying rigid cut-offs.

As per the <u>assessment of infants and young children section</u>, when direct information about the clinical significance of neurodevelopmental impairments is not available, microcephaly ($\leq 3^{rd}$ percentile) may be used as an indicator. A more stringent definition of small head circumference is applied when it is used as a proxy for assessment of neurodevelopmental impairments.

For further good practice statements supporting physical size assessment in practice, refer to the medical assessment section of the <u>main guidelines document</u>.

4.3.8 Associated features

There was insufficient evidence for some physical, neurological, and neurodevelopmental outcomes to be included in the diagnostic criteria. However, collecting information on the presence of these features/conditions is useful as they can provide vital information to inform individualised referrals, treatment, and ongoing supports. Future research is needed to better understand the potential associations of these features/conditions with PAE.

4.3.8.1 Reasoning regarding structural brain abnormalities

Based on a review of the best available evidence, PAE can be associated with a range of structural brain abnormalities. However, research documenting these abnormalities is predominately based on advanced quantitative MRI findings. Currently, available data from routine clinical MRI (i.e., qualitative radiological MRI) do not currently provide diagnostic utility. Therefore, if abnormal imaging results are available, it is recommended these are recorded as associated features. This approach supports documentation and consideration of available results in the assessment but does not include these results as part of the neurodevelopmental domains, based on the available evidence.

4.3.8.2 Reasoning regarding other neurological conditions

A review of the best available evidence indicated insufficient evidence to understand the association between PAE and neurological conditions of hearing and vision impairment, seizures, and cerebral palsy. Therefore, it is recommended that these neurological conditions be recorded as associated features. Some members of the Clinical Advisory Group members also highlighted that the genetic basis of seizures is an emerging area of research. This approach supports recording and consideration of neurological conditions in the assessment process but does not include these conditions as part of the neurodevelopmental domains, based on currently available evidence.

4.3.9 At risk of FASD

Feedback from the Clinical Advisory Groups indicated that the 'at risk' designation has been a helpful option for practitioners. Specifically, it was discussed that this designation can facilitate access to early supports and encourage review when children are older to determine if a diagnosis is appropriate.

In Australia, access to early intervention does not require a diagnosis but rather presence of developmental delay. Therefore, an 'at-risk' designation in these cases should not impact access to supports, including the NDIS. Instead, it allows for more time and consideration of whether a lifelong diagnosis would be appropriate. However, it was noted that the decision to repeat testing should be made by an appropriately qualified practitioner, not an NDIS coordinator who may lack necessary qualifications to make these clinical decisions.

Concerns were raised by Advisory Group members that the 'at risk' designation can sometimes be inappropriately applied, leading to inequities for individuals and families, especially, across different settings where resources and clinical capacity differ. Practitioners are encouraged to use shared-care approaches to support additional assessment and diagnostic pathways in low-resource settings and access professional development and clinical supervision as required.

