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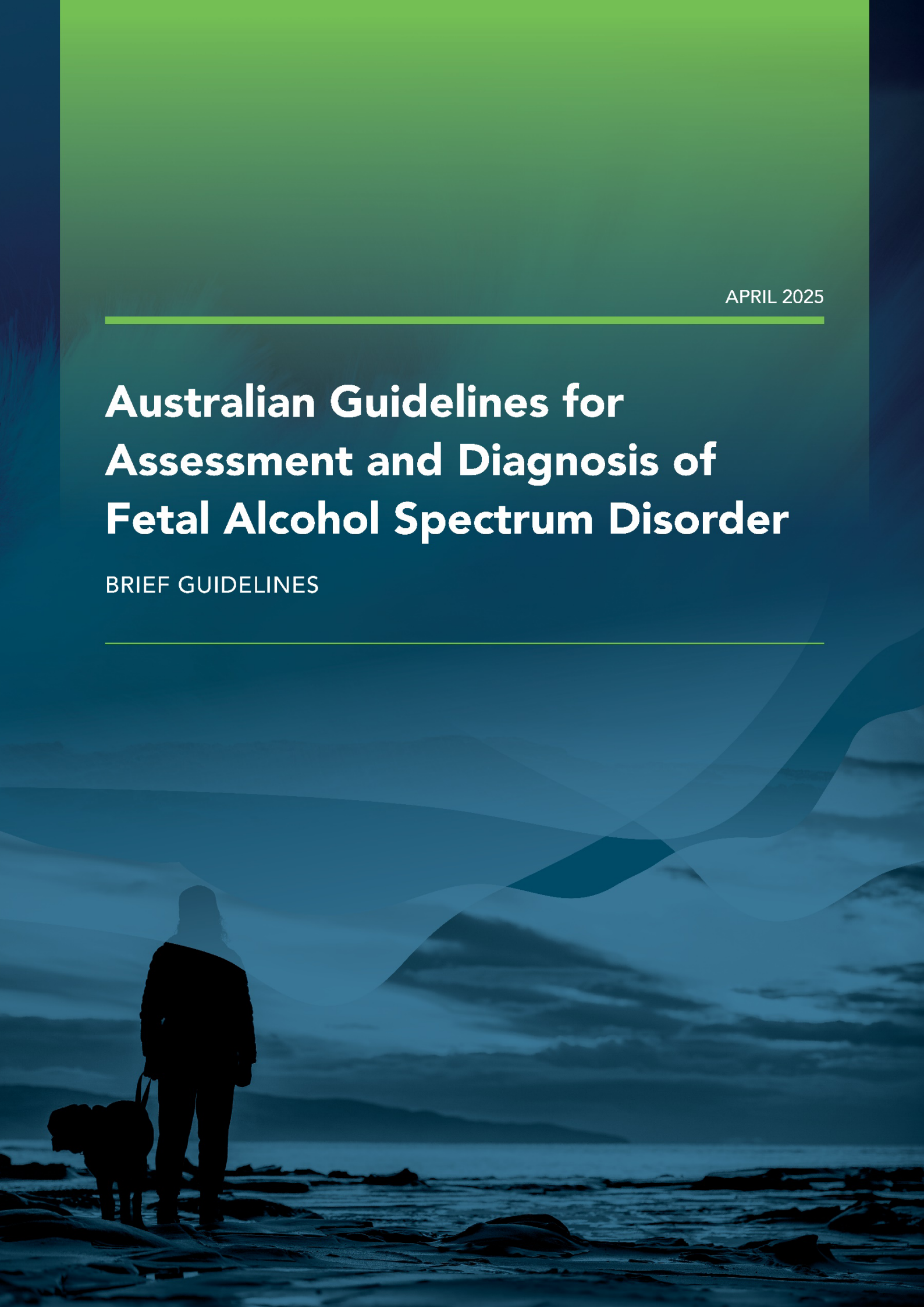
Australian Guidelines for Assessment and Diagnosis of Fetal Alcohol Spectrum Disorder

BRIEF GUIDELINES



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Disclaimer:	These guidelines are a general guide to best practice, to be applied subject to health professionals' judgement and values, and the circumstances and needs and preferences of the individual attending for assessment. These guidelines are designed to provide information to assist clinical decision making and the recommendations included are based on the best evidence available when they were developed. Practitioners can access appropriate professional development and supervision where required to support effective implementation.
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See the Administrative and Technical Report for member affiliations and details of group recruitment.

1. Overview

This abridged version of the *Australian Guidelines for Assessment and Diagnosis of Fetal Alcohol Spectrum Disorder (FASD)* was produced to support health practitioners in undertaking assessments that may result in a diagnosis of FASD. It outlines the recommended assessment principles and diagnostic criteria and provides links to the actionable statements, additional information, and practitioner resources to support application of the diagnostic criteria.

Practitioners are advised to familiarise themselves with the full guidelines document. This abridged version is provided as a practical support for day-to-day application in clinical settings and intended for use in conjunction with the full guidelines document.

The diagnostic criteria contained in this abridged version are intended for use as part of a holistic interprofessional assessment process, which incorporates shared decision-making with individuals and families, as detailed in the full guidelines document.

Summary tables with details of where to access more information and links to all the associated documents are provided to support implementation of the diagnostic criteria.

2. Assessment Principles

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 life-long 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.

3. 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 additional information is provided in the sections following the diagnostic criteria in the full guidelines document 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:

5. 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**

5. 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 (≤ 3rd 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.

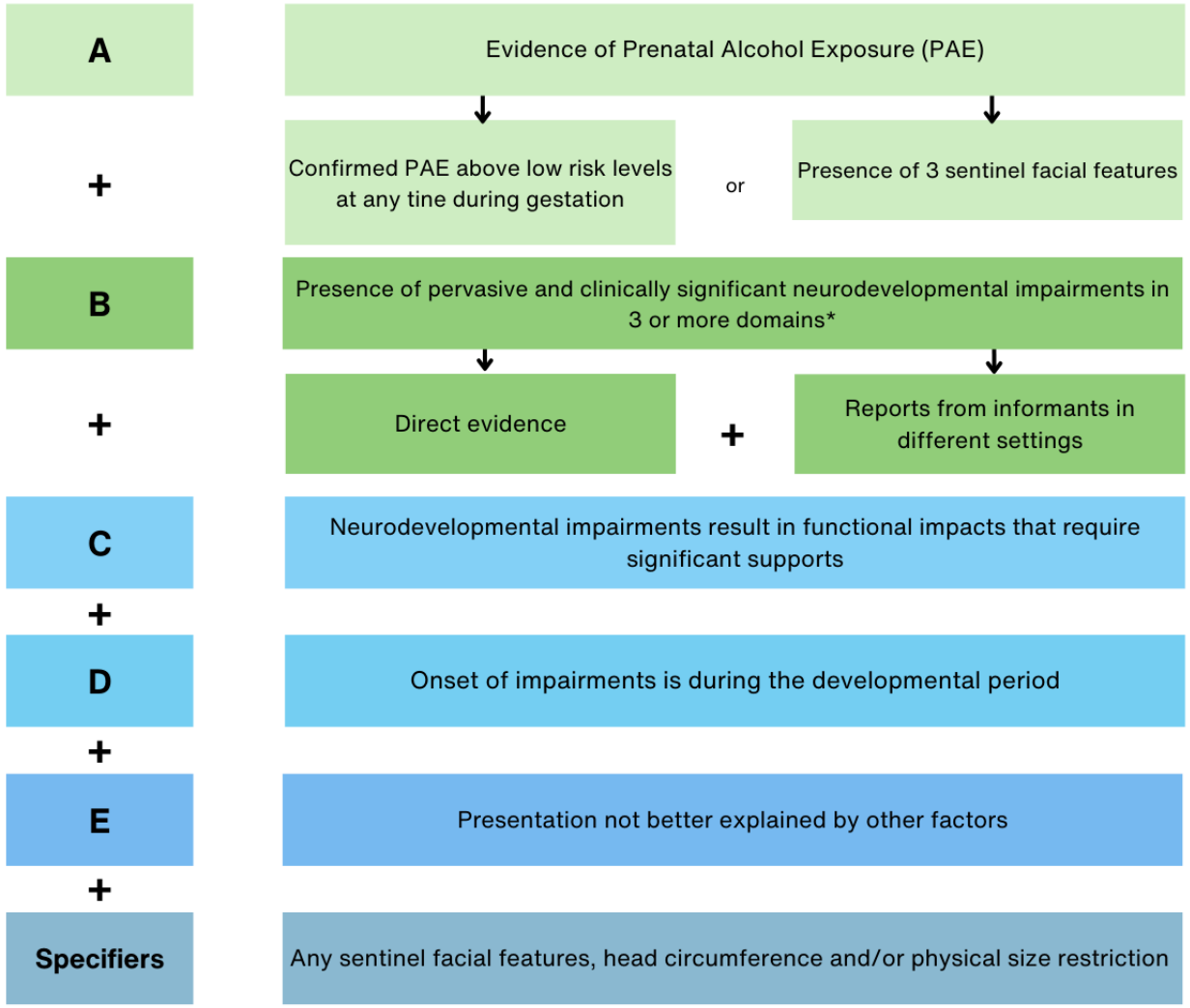
DSM-5-TR: Unspecified Neurodevelopmental Disorder (F89)

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.



***Neurodevelopmental domains**

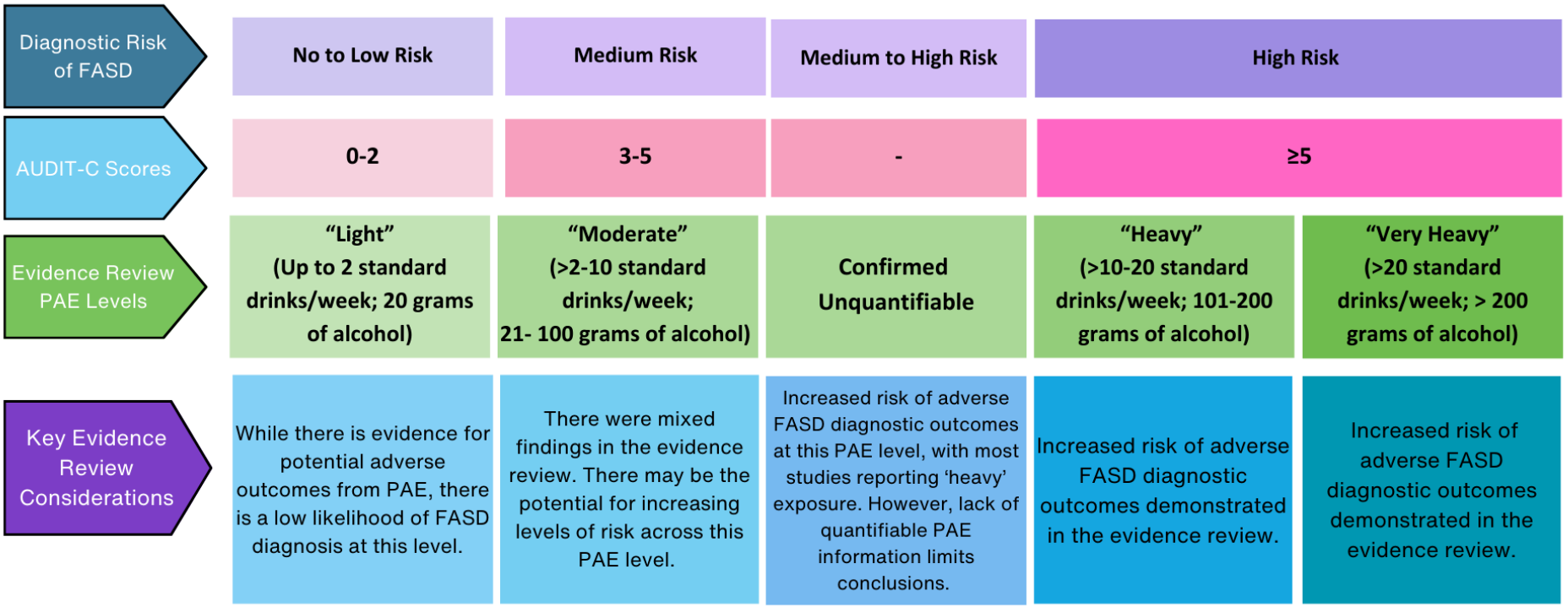
1. Communication
2. Motor skills
3. Intellectual abilities
4. Attention
5. Memory
6. Executive function
7. Emotional and/or behavioural regulation
8. Literacy and/or Numeracy skills
9. Adaptive/social functioning

Note. In infants and young children, in the absence of direct evidence of impairments, following exclusion of other causes, microcephaly may be used as an indicator of neurodevelopmental impairment.

5. Site of Additional Information to Support use of the Diagnostic Criteria

Criterion	Location of additional information in the full guidelines document
Criterion A: More than low risk exposure or presence of three sentinel facial features.	• Figure 2 below provides further details to support assessment of PAE risk. • Additional information section 4.3.2 – Criterion A: PAE • *Chapter 6: Prenatal alcohol exposure assessment.* Detailed good practice statements and implementation considerations are provided to support practitioners in assessing PAE. • *Appendix E: P*ractitioner support templates • Additional information *section 4.3.6 – diagnostic specifier: sentinel facial features* • *Chapter 7: Medical Assessment.* • *Appendix E: Practitioner support templates*
Criterion B: Presence of pervasive and clinically significant neurodevelopmental impairments	• Table 1 provides an overview of the neurodevelopmental domains and key assessment considerations. • Additional information *section 4.3.3 – Criterion B: Presence of pervasive neurodevelopmental impairments* • Additional information *section 4.3.3.1* - applying standardised tests in the assessment. • Additional information *section 4.3.3.2* - determining the clinical significance of neurodevelopmental impairments (includes sections on standardised tests, percentiles, cut scores, confidence intervals). • Additional information *section 4.3.3.3* - Assessing neurodevelopmental domains in practice (includes information on general assessment advice [e.g., interprofessional framework, what to do if working in contexts with limited multidisciplinary team. access], assessment of infants and young children and consideration of co-occurring conditions). • Additional information section 4*.3.3.4*-Neurodevelopmental domains: evidence for inclusion. • *Chapter 8: Holistic developmental, functional and wellbeing assessment.* • *Chapter 9: Holistic profile, formulation and strengths-based pathways.* • *Appendix E: Practitioner support templates*
Criterion C: The neurodevelopmental impairments result in functional impacts that necessitate significant supports.	• Additional information *section 4.3.4 – Criterion C* • *Chapter 8: Holistic developmental, functional and wellbeing assessment.* • *Chapter 9: Holistic profile, formulation, and strengths-based pathways.*

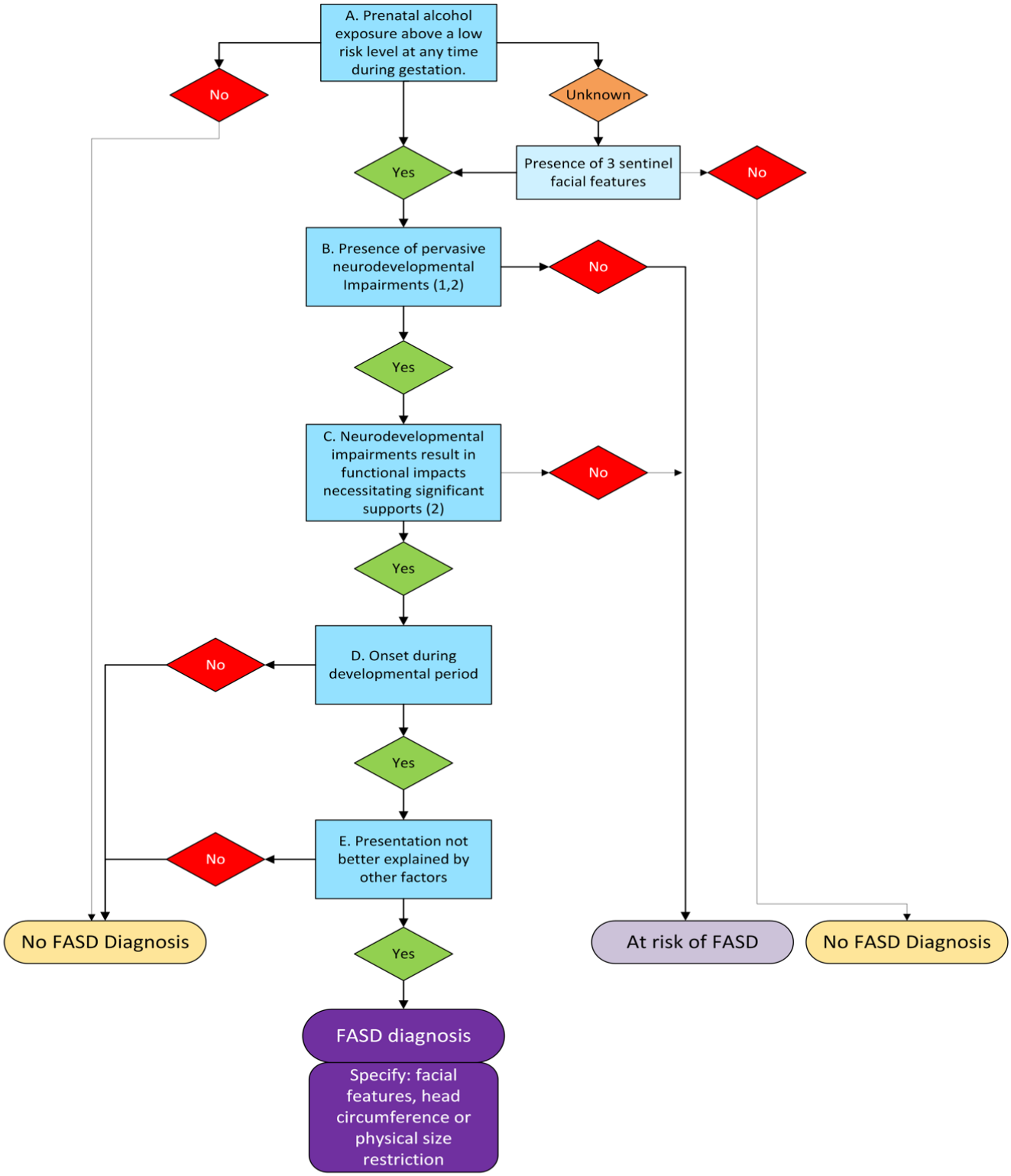
	• *Appendix E: Practitioner support templates.*
Criterion D: Onset of neurodevelopmental impairments is in developmental period.	• Additional information *section 4.3.5 – Criterion D* • *Chapter 8: Holistic developmental, functional and wellbeing assessment.* • *Chapter 9: Holistic profile, formulation, and strengths-based pathways.* • *Appendix E: Practitioner support templates.*
Criterion E: The symptoms are not better attributed to another condition or exposure.	• Detailed information provided in the diagnostic criteria box. • *Chapter 7: Medical Assessment.* • *Appendix E: Practitioner support templates.* • *Chapter 9: C*o-occurring and differential diagnosis and trauma and PAE sections
Diagnostic specifiers 1,2, 3 or no sentinel facial features Head circumference restriction at birth and/or postnatally. Physical size restriction at birth and/or postnatally.	• Additional information *section 4.3.6 (facial features), 4.3.7 (head circumference & physical size)* • *Chapter 7: Medical Assessment*. • *Appendix E: Practitioner support templates.*
Associated features	• Additional information section 4.3.8 – associated features.



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 2. 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.



(1) Presence of pervasive neurodevelopmental impairments meets Criterion B providing
- Clinically significant impairments in 3 or more neurodevelopmental domains.
- Documentation of impairments by multiple informants.
- Direct evidence of clinically significant impairments.

(2) In infants and young children
- Microcephaly (≤ 3rd percentile) could be used as an indicator of clinically significant neurodevelopmental impairment, providing the presentation is not better explained by another condition or exposure (Criterion E).

Figure 3. Diagnostic Algorithm

5. Summary of Some Key Further Information

Summary of Changes	• A summary of changes from the 2016 Australian Guide to the Diagnosis of FASD and the current Guidelines are provided in the main guidelines full document (Chapter 10) and as a separate document.
Evidence underpinning the guidelines	• A summary of the evidence is provided in the main guidelines full document (Chapter 11), an overview of findings is provided in the Administrative and Technical Report (including summarised evidence-to-decision frameworks), and detailed information is provided in each of the individual Technical Reports and Supplemental Files.
Foundational Considerations	• A summary of several key frameworks and principles are provided to support practitioners in Chapter 3 of the main guidelines document.
Assessment Process	• Chapter 5 provides an overview of the recommended assessment process. This process aims to encourage all practitioners, no matter the setting or discipline to contribute where they can to assessment and diagnosis of FASD.
Evidence gaps	• The main guidelines document (Chapter 11) provides a brief overview of some of the key evidence gaps identified through the guidelines development process.
Indigenous Framework	• Information is embedded throughout the full main guidelines document and an additional expanded resource is also provided that provides more details to support practitioners in providing culturally responsive assessment and diagnostic services.
Administrative and Technical Report	• Provides an overview of the project governance and process. • Appendices in this document include summaries of Advisory Group, public consultation, and NHMRC Methodological and Clinical review feedback.
Dissemination, Implementation and Evaluation Report	• Provides additional information to support uptake of the guidelines in practice, as well as monitoring and evaluation.

Note. Links to all the documents referred to in this Table are provided below.

6. Links to associated documents

- Full Guidelines Document
- Indigenous Framework
- Frequently Asked Questions
- A Plan English Guide to Reading the Guidelines
- Summary of Actionable Statements
- Summary of Changes
- Assessment Principles and Diagnostic Criteria
- Administrative and Technical Report
- Dissemination, Implementation, and Evaluation Report
- Lived experiences of the assessment and diagnostic process: Systematic review and qualitative synthesis report
- Factors to be considered as part of a holistic assessment: Scoping review report
- Exploring resource implications and models of care: Scoping review report
- Association between prenatal alcohol exposure, physical size, dysmorphology and neurodevelopment: Systematic review report
- Supplemental File A: Study exclusion list
- Supplemental File B: Risk of bias assessment
- Supplemental File C: Physical size GRADE ratings and forest plots
- Supplemental File D: Regression summaries
- Supplemental File E: Dysmorphology GRADE ratings and forest plots
- Supplemental File F: Functional neurodevelopmental GRADE ratings and forest plots
- Supplemental File G: Structural and neurological GRADE ratings and forest plots