BRIEF COMMUNICATION

Australian guide to the diagnosis of foetal alcohol spectrum disorder: A summary

Foetal alcohol spectrum disorder (FASD) is a complex neurodevelopmental disorder caused by prenatal alcohol exposure (PAE). In some individuals, characteristic facial features result from the teratogenic effect of first trimester PAE.

In response to limited training opportunities in FASD, lack of a nationally adopted diagnostic instrument and confusion about diagnostic criteria, *The Australian Guide to the Diagnosis of Fetal Alcohol Spectrum Disorder* has been developed, funded by the Commonwealth Department of Health (DoH).

Building on a literature review of diagnostic criteria and guidelines from Canada and the USA,[1–4] Delphi surveys[5–9] and a consensus workshop using the GRADE approach,[10,11] an Australian diagnostic instrument was developed in 2012. In 2015–2016, with additional funding from the DoH, a feasibility trial was conducted, and the instrument, guide and e-learning modules were finalised and harmonised with the new Canadian guideline for diagnosis of FASD.[12] The *Australian Guide to the Diagnosis of Fetal Alcohol Spectrum Disorder* and e-learning modules were released in mid-2016 and are freely available at http://alcoholpregnancy. telethonkids.org.au/australian-fasd-diagnostic-instrument/australian-guide-to-the-diagnosis-of-fasd/. This includes clinical forms that can be used during evaluation.

A key recommendation in the guide is the adoption of FASD as a diagnostic term, with two subcategories: FASD with three sentinel facial features and FASD with less than three sentinel facial features. Diagnostic criteria relate to PAE, severe neurodevelopmental impairment in 3 out of 10 domains and sentinel facial features (small palpebral fissures, smooth philtrum and thin upper lip) (Table 1; Fig. 1). FASD with three sentinel facial features replaces the diagnosis of foetal alcohol syndrome, but without a requirement for growth impairment.[11] FASD with less than three sentinel facial features encompasses the previous categories of partial foetal alcohol syndrome and neurodevelopmental disorder-alcohol exposed.[11] Co-existing or alternative diagnoses including genetic conditions (e.g. microdeletions or duplications), effects of other teratogens and prenatal exposures, as well as the effects of postnatal exposures such as early life trauma and brain injury should be considered.

The diagnosis of FASD requires multidisciplinary assessment including comprehensive physical and developmental assessment, as well as psychometric testing, preferably by a multidisciplinary diagnostic team, and typically led by a medical specialist such as a paediatrician, psychiatrist or geneticist.

The assessment process may be confronting for the individual or caregiver who should provide informed consent beforehand, and receive appropriate support as required. This is particularly salient when biological parents or family are involved. An assessment report outlining the individual's strengths and difficulties, and recommendations should be provided to the family and referring clinician or agency. The value of sharing this information with relevant service providers (including teachers) should be discussed. If FASD is diagnosed, written information about the condition and contact details for the National Organisation for FASD (http://www.nofasd.org/) should be provided.

Dissemination of the guide and the e-learning modules aims to standardise FASD diagnosis. We hope that it will provide clinicians with increased confidence to consider a diagnosis of FASD, the knowledge to refer for or make the diagnosis, and the information needed to manage and support individuals and families living with FASD. The guide will be updated as new evidence emerges, to ensure it reflects current knowledge and best practice in the evolving field of FASD. Having national, standardised criteria for diagnosis will also improve our ability to advocate for services, monitor FASD prevalence, and support efforts to reduce PAE and hence enable primary prevention of FASD.

Acknowledgements

Table 1 Diagnostic criteria and categories for foetal alcohol spectrum disorder (FASD)

Diagnostic criteria	Diagnostic categories	
	FASD with three sentinel facial features	FASD with less than three sentinel facial features
Prenatal alcohol exposure	Confirmed or unknown	Confirmed
Neurodevelopmental domains	Severe impairment† in at least three neurodevelopmental domains	Severe impairment† in at least three neurodevelopmental domains
• Brain structure/Neurology		
• Motor skills		
• Cognition		
• Language		
• Academic achievement		
• Memory		
• Attention		
• Executive function, including impulse control and hyperactivity		
• Affect regulation		
• Adaptive behaviour, social skills or social communication		
Sentinel facial features	Presence of three sentinel facial features	Presence of 0, 1 or 2 sentinel facial features
• Short palpebral fissure		
• Smooth philtrum		
• Thin upper lip		

†Severe impairment is defined as either a global score or a major subdomain score on a standardised validated neurodevelopmental scale that is ≤2 standard deviation below the mean or <3rd percentile.
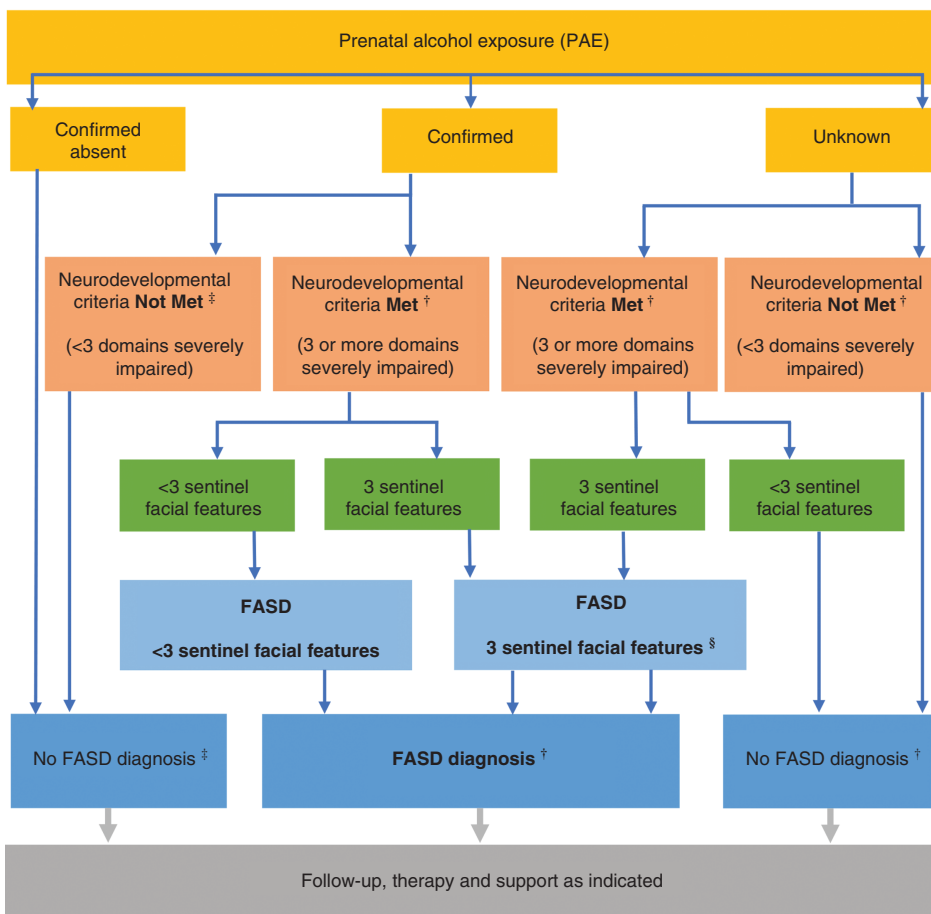
Fig. 1 Australian diagnostic algorithm for foetal alcohol spectrum disorder (FASD). †Assessment fully completed and other diagnosis have been considered. Currency of assessment is also assumed. For infants and children under 6 years of age, *severe global developmental delay* meets criteria for neurodevelopmental impairment (in three or more domains) if it is confirmed on a standardised assessment tool (e.g. Bayley or Griffiths). ‡In the presence of confirmed PAE, reassessment of neurodevelopmental domains can be considered as clinically indicated (e.g. if there is a decline in an individual's functional skills or adaptive behaviour over time). §In infants and young children under 6 years of age with *microcephaly and all three sentinel facial features*, a diagnosis of FASD with three sentinel facial features can be made, whether *PAE is confirmed or unknown*, even without evidence of severe neurodevelopmental impairment in three domains based on standardised assessment. (Modified from Cook et al.,[12] fig. 1, with permission.)

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Eagle by Dulan Kumal Acharya (age 10) from Operation Art 2016