



Fetal alcohol exposure



PAEDIATRICS

Fetal alcohol spectrum disorder in Australia: Practice guidelines for diagnosis and management

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ALCOHOL is a teratogen and causes damage to the fetal CNS and other organ systems. This may result in significant neurocognitive impairment, prenatal and postnatal growth impairment as well as a syndrome of mid-face abnormalities.¹ Disorders characterised by these features are collectively termed fetal alcohol spectrum disorders (FASD). However, it must be noted that some individuals with FASD do not have growth restriction or characteristic facial features.

While higher levels of alcohol consumption increase the likelihood of FASD, it is not known if there is a safe level of alcohol use in pregnancy. Animal studies have shown low-level exposure may lead to loss of fetal brain cells.² There is emerging evidence

that alcohol use during pregnancy can have epigenetic effects that may be transmitted to future generations.³

Alcohol consumption is common among Australian women. National survey data suggest that about 50-60% of Australian women drink in pregnancy.⁴ FASD occurs in all cross-sections of society and geographic regions, with a reported incidence of 0.6 per 1000 live births.⁵

Although alcohol use in pregnancy is ubiquitous across Australia, Aboriginal Australians are a highly researched population, and a lot of data come from their communities. FASD rates of up to 19% are documented.⁶ Alcohol consumption during pregnancy has a complex aetiology, including poor understanding of fetal consequences, stressful living circumstances, a

partner who consumes alcohol, addiction or underlying mental illness.

In 2009, the NHMRC produced national guidelines recommending no alcohol consumption was the safest option during pregnancy.

In 2016, the AMA

and for patients with FASD to be able to access the National Disability Insurance Scheme.

The AMA guidelines also encourage GPs to engage in non-judgemental conversations with their patients about alcohol consumption during pregnancy, and

experiencing developmental delay. The AUDIT-C screening questionnaire to assess exposure risk is a helpful tool (see box, next page). The AUDIT-C questions should be asked prior to pregnancy, and in each trimester. Any report of alcohol use in pregnancy should be responded to with a brief intervention or, where appropriate, referral to mental health or alcohol counselling services.

Diagnostic assessment

Australian diagnostic criteria were finalised in 2016, and updated to include two diagnostic categories — FASD with sentinel facial features and FASD without sentinel facial features.⁷

The new Australian FASD diagnostic guidelines and e-learning package were developed as part of the Commonwealth FASD Action Plan to help standardise FASD diagnoses

across Australia, in line with international frameworks.

Facial features indicative of FASD (figure 1) should be confirmed by a paediatrician, but may be assessed by a GP as part of the referral process.

The three sentinel facial features are:

1. Short palpebral fissure lengths (the distance between the inner and outer corners of one eye);
2. Smooth philtrum (the vertical groove between the nose and upper lip); and
3. Thin upper lip.

It is important to note that facial features are not necessary for a diagnosis of FASD, and may occur in only a small percentage of affected individuals.

Other dysmorphic features that may be seen in FASD include a flat nasal bridge, flat mid-face, *cont'd next page*

IT IS NOT KNOWN IF THERE IS A SAFE LEVEL OF ALCOHOL USE IN PREGNANCY.

released its position statement on FASD, with a number of recommendations. One recommendation suggested that the disorder be included in the Australian Government Department of Social Services' List of Recognised Disabilities,

provide advice consistent with the NHMRC *Australian Guidelines to Reduce Health Risks from Drinking Alcohol*.

It is advised to sensitively screen for alcohol use in every pregnancy and consider FASD if a child is

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upturned nose, clinodactyly (curving of the fifth finger towards the fourth finger), abnormalities of the palmar crease (figure 2), decreased elbow pronation/supination, railroad track ears, camptodactyly (most common on fourth and fifth fingers), as well as major birth defects (cardiac, renal, ocular, auditory and skeletal). Prenatal or postnatal growth retardation can result in height or weight percentiles in the low range. Microcephaly can also occur, as well as structural brain anomalies.

Ideally, accurate diagnosis requires a multidisciplinary team approach, and confirmation by a paediatrician or neurologist. Individuals over 18 may be referred to a neurologist or psychiatrist for further diagnostic assessment.

Generally, diagnosis is made when a patient has a history of prenatal alcohol exposure and, as per the Australian diagnostic guidelines, significant impairment (less than the third percentile) in at least three neurodevelopmental domains — including brain structure, motor skills, cognition (IQ), language, academic achievement, memory, attention, executive function, impulse control/ hyperactivity, affect regulation, adaptive behaviour, social skills or social communication.

Some neurodevelopmental impairments may not be evident in younger children as it is possible that they will 'grow into' their difficulties as school and social demands increase. A high proportion of children with FASD will have significant problems with attention and hyperactivity and it is important not to overlook a potential FASD diagnosis when considering ADHD.

A detailed history should



Figure 1. Facial features indicative of FASD may be assessed by a GP as part of the referral process.

include maternal use of alcohol or other substances during pregnancy, and personal and family history of mental health or other medical disorders. The developmental and psychosocial history should include a behavioural history including major life stressors, such as domestic violence and early life trauma/neglect and protective factors.

Additionally, an education history should explore any cognitive or learning difficulties.

Physical examination should include height, weight and measurement of head circumference in patients of all ages. Other tests that may be clinically indicated include audiology and visual testing, FBC, ferritin, vitamin B, thyroid function and creatinine kinase. A chromosomal microarray should also be

requested if there are dysmorphic features, intellectual disability or a CNS structural abnormality present.

FASD is a notifiable developmental anomaly in some states, and an Australian FASD registry has been established through the Australian

THERE HAS BEEN GROWING EVIDENCE FOR THE EFFECTIVENESS OF INTERVENTIONS FOR FASD THAT FOCUS ON PSYCHOEDUCATION FOR PARENTS OR CARERS.

Paediatric Surveillance Unit.

Referrals for further assessment

Referral for a comprehensive



Figure 2. Other dysmorphic features that may be seen include abnormalities of the palmar crease.

developmental assessment — clinical psychology, neuropsychology, occupational therapy, physiotherapy or speech pathology — may be necessary, depending on the patient's presenting complaints. This will tease out other potential contribut-

ing factors, such as cultural impact, past trauma/neglect, disrupted attachment, substance abuse or head injuries.⁸

Obtaining a profile of the child's strengths and weaknesses across cognitive, psychological, social and behavioural domains is important because of the heterogeneous nature of FASD, to provide a baseline to monitor progress and develop appropriate educational goals, treatment options, compensatory strategies and case management.

Early diagnosis to facilitate access to additional assessment and treatment helps to reduce the impact of the cognitive, social/emotional and educational difficulties that can occur in FASD.

Early therapy intervention enables carers, teachers, family members and the treatment team to better understand the child's difficulties and behaviour leading to more appropriate and tar-

geted interventions.

Individuals with a diagnosis of FASD are also more at risk of becoming involved in the justice system without adequate supports. Compensatory strategies, such as memory aids or simple visual tools to assist with planning and organisation may be helpful, as well as additional support if patients are involved in the justice system. Adult patients may wish to apply to the state administrative tribunal for a Public Trustee for financial management.

There has been growing evidence for the effectiveness of interventions for FASD that focus on psychoeducation for parents or carers and integrating individualised interventions into existing treatments.⁹

A recent Australian review of FASD interventions emphasised a range of key principles when planning services for individuals with FASD.¹⁰

These principles include the benefits of a multi-disciplinary, co-ordinated approach, building on the patient and caregiver strengths, cultural security for ethnically diverse patients and recognising that certain transition periods may be a vulnerable time in a patient's life (such as the transition from primary to high school, or from adolescence to adulthood). ●

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References on request.

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AUDIT-C Questions

Source of reported information on alcohol use: Birth Mother Other (please specify)

1. How often did the birth mother have a drink containing alcohol during this pregnancy?

Unknown	Never (skip Q2+Q3)	Monthly or less	2-4 times a month	2-3 times a week	4 or more times a week
<input type="checkbox"/>	<input type="checkbox"/> 0	<input type="checkbox"/> 1	<input type="checkbox"/>	<input type="checkbox"/> 3	<input type="checkbox"/> 4

2. How many standard drinks did the birth mother have on a typical day when she was drinking during this pregnancy?

Unknown	1 or 2	3 or 4	5 or 6	7 to 9	10 or more
<input type="checkbox"/>	<input type="checkbox"/> 0	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4

3. How often did the birth mother have five or more standard drinks on one occasion during this pregnancy?

Unknown	Never	Less than monthly	Monthly	Weekly	Daily or almost daily
<input type="checkbox"/>	<input type="checkbox"/> 0	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4

AUDIT-C score during this pregnancy: (Q1+Q2+Q3)= _____ Scores: 0 = no risk; 1-4 = confirmed; 5+ = confirmed high-risk

ADDITIONAL RESOURCES

NHMRC guidelines on reducing health risks from alcohol. See: bit.ly/1fd7Mvu

Telethon Kids Institute. Australian Guide to the diagnosis of FASD. See: bit.ly/2pm0B40

National Organisation for Fetal Alcohol Spectrum Disorders (NOFASD Australia) See: www.nofasd.org.au

Russell Family Fetal Alcohol Disorders Association See: www.rffada.org