

What is this research about?

Exposure to alcohol in pregnancy may cause irreversible damage to the brain of the unborn child, with devastating life-long consequences. Although avoiding alcohol will prevent fetal damage, alcohol is still frequently used by Australian women in pregnancy. Fetal Alcohol Spectrum Disorders (FASD) is a term used to refer collectively to a range of disorders caused by prenatal alcohol exposure.

The Australian FASD Collaboration set out to develop a diagnostic instrument for FASD in Australia.

What is a diagnostic instrument?

A diagnostic instrument is usually a series of documents (paper, on-line or apps) to ensure that health professionals know:

- what to measure
- how to measure it
- how to record the information
- how to rank the information to meet criteria for a particular condition, illness or disease

The project was funded by the Commonwealth Department of Health and Ageing.

What did we do?

Literature review: Reviewed Australian and international literature on screening and diagnosis of FASD.

Community conversations: Engaged women in Perth and Cairns in conversations to explore questions about alcohol and pregnancy.

Delphi survey: Conducted an on-line questionnaire (2 rounds) with Australian and international health professionals and researchers with expertise in FASD to identify key components for a diagnostic instrument for use in Australia.

Consensus development workshop: Held a workshop with members of the Australian FASD Collaboration to consider the evidence from the literature review, community conversations and the Delphi survey and made recommendations for the content of the diagnostic instrument and recommend methods to validate the instrument and support its use.

Who took part in this study?

19 members of the Australian FASD Collaboration (health professionals, researchers, epidemiologists and consumer and community representatives)

32 women in Perth and Cairns participated in the community conversations

103 health professionals (paediatricians, geneticists, obstetricians, addition medicine specialists, psychiatrists, nurses, psychologists, occupational therapists, speech pathologists, physiotherapists and social workers) and researchers participated in a Delphi survey

What did we find out?

Literature review

- Did not identify any standardised screening or diagnostic instruments developed in Australia
- No internationally recognised gold standard for screening and/or diagnosing disorders within the fetal alcohol spectrum

Community conversations

- Provided an important foundation for meaningful and inclusive consumer and community participation
- A standard set of questions (smoking, alcohol, diet) should be asked of all pregnant women
- Parents, guardians or kinship carers should be asked to provide informed consent before proceeding with screening and diagnosis
- Health professionals need education and training on FASD and how to speak to women about alcohol use in pregnancy
- Health professionals should provide a clear and consistent message to women that researchers don't know what level of alcohol if any, is safe in pregnancy. Therefore the best advice is not to drink any alcohol while pregnant

Delphi survey and workshop

Summary of consensus recommendations for the diagnosis of FASD in Australia

Population screening: Not recommended

Referral: Use of standard criteria for referral for specialist diagnostic assessment

Diagnostic categories: Fetal Alcohol Syndrome (FAS), Partial Fetal Alcohol Syndrome (pFAS) and Neurodevelopmental Disorder-Alcohol Exposed (ND-AE)

Diagnostic assessment methods: Interdisciplinary (paediatrician, psychologist, speech pathologist, occupational therapist) approach to assessment

Resources for implementation: Development of comprehensive resources to facilitate national implementation of standard diagnostic criteria and national case reporting

Consumer information and support: Information and support should be provided to individuals and their parents or carers during the diagnostic process

See Watkins et al 'Recommendations from a consensus development workshop on the diagnosis of fetal alcohol spectrum disorders in Australia' for full details of the recommendations and diagnostic categories

How will this research help?

The adoption and implementation of an instrument of standard diagnostic criteria for FASD in Australia will:

- enable earlier diagnosis and management of FASD and improve health outcomes
- enable us to estimate the number of children with FASD and improve reporting
- inform programs for the management and prevention of FASD
- improve training and awareness of FASD amongst health professionals and other service providers
- optimise service provision and quality of life for people with FASD and their families

Project researchers

Australian FASD Collaboration – health professionals, researchers and consumer and community representatives. The Collaboration was led by Professor Carol Bower, Telethon Institute for Child Health Research and Professor Elizabeth Elliott, University of Sydney

The full membership of the Australian FASD Collaboration can be found in 'Our Research' section of the website.

Recommended Australia FASD diagnostic categories and criteria			
Diagnostic criteria	Diagnostic category		
	Fetal Alcohol Syndrome (FAS)	Partial Fetal Alcohol Syndrome (PFAS)	Neurodevelopment Disorder-Alcohol Exposed (ND-AE)
Requirements for diagnosis	Requires all 4 of the following criteria to be met:	Requires confirmed prenatal alcohol exposure, the presence of 2 of the 3 characteristic FAS facial anomalies at any age, and CNS criteria to be met:	Requires confirmed prenatal alcohol exposure and CNS criteria to be met:
Prenatal alcohol exposure	Confirmed or unknown	Confirmed	Confirmed
Facial anomalies	Presence of all 3 of the following facial anomalies at any age: <ol style="list-style-type: none"> short palpebral fissure length (2 or more standard deviations below the mean) smooth philtrum (Rank 4 or 5 on the UW Lip-Philtrum Guide[†]) thin upper lip (Rank 4 or 5 on the UW Lip-Philtrum Guide[†]) 	Presence of any 2 of the following facial anomalies at any age: <ol style="list-style-type: none"> short palpebral fissure length (2 or more standard deviations below the mean) smooth philtrum (Rank 4 or 5 on the UW Lip-Philtrum Guide[†]) thin upper lip (Rank 4 or 5 on the UW Lip-Philtrum Guide[†]) 	No anomalies required*
Growth deficit	Prenatal or postnatal growth deficit indicated by birth length or weight \leq 10th percentile adjusted for gestational age, or postnatal height or weight \leq 10th percentile	No deficit required*	No deficit required*
Central Nervous System (CNS) abnormality	At least 1 of the following: <ol style="list-style-type: none"> clinically significant structural abnormality (e.g. OFC \leq 3rd percentile, abnormal brain structure), or neurological abnormality (seizure disorder or hard neurological signs); and/or severe dysfunction (impairment in 3 or more domains of function, 2 or more standard deviations below the mean)[‡] 		