THE DEVELOPMENT OF A DIAGNOSTIC INSTRUMENT FOR FETAL ALCOHOL SPECTRUM DISORDERS IN AUSTRALIA

Australian FASD Collaboration

Final Report: Volume 1

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The Development of a Diagnostic Instrument for Fetal Alcohol Spectrum Disorders in Australia

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EXECUTIVE SUMMARY

INTRODUCTION

Exposure to alcohol in pregnancy may cause irreversible damage to the brain of the unborn child, with devastating life-long consequences. Although alcohol avoidance will prevent fetal damage, alcohol is still frequently used by Australian women in pregnancy. Fetal Alcohol Spectrum Disorders (FASD) is a non-diagnostic descriptive term used to refer collectively to a range of disorders caused by prenatal alcohol exposure. The term FASD is commonly understood to refer to the following four diagnostic categories identified by the Institute of Medicine (Stratton, Howe & Battaglia 1996): Fetal Alcohol Syndrome (FAS), Partial Fetal Alcohol Syndrome (PFAS), Alcohol-Related Neurodevelopmental Disorders (ARND) and Alcohol-Related Birth Defects (ARBD). Early recognition of alcohol-exposed infants and early diagnosis of FASD are crucial to allow early intervention and improve long term outcomes for children, to help treat vulnerable women and families, and to prevent subsequent birth of affected children.

There is good evidence however that FASD are poorly recognised in Australia and that the diagnoses are often missed or delayed. There are no sustainable services specifically for the screening or diagnosis for FASD in Australia, no national guidelines for the diagnosis and management of FASD and no nationally accepted diagnostic or screening instrument. Furthermore, health professionals infrequently ask about alcohol use in pregnancy and most feel ill-equipped to advise women about alcohol use in pregnancy or its potential adverse effects. They do not know the diagnostic features of FASD, feel ill-prepared to diagnose or manage FASD, have concerns that diagnoses will stigmatise children and their families, and do not know where to refer affected children. These barriers to diagnosis are accentuated in rural, remote and Indigenous communities where access to health services is limited. Lack of identification of FASD means we are unable to quantify the problem and thus lack evidence to advocate for much needed health professional training and diagnostic services.

THE PROJECT

In response to a tender from the Telethon Institute for Child Health Research and The University of Sydney, the Commonwealth Department of Health and Ageing funded the Australian FASD Collaboration to develop an instrument that could be used to improve the identification and/or diagnosis of FASD in Australia. Early and accurate diagnosis will improve health outcomes and quality of life for individuals with FASD and their families. The Australian FASD Collaboration is a national collaboration of highly qualified and experienced health professionals, epidemiologists, researchers

and consumer and community representatives dedicated to the prevention, diagnosis and care of individuals with FASD.

The Australian FASD Collaboration sought to develop a diagnostic instrument for FASD for use in Australia by collecting and evaluating evidence from the following four sources:

1. A systematic review of the international literature on FASD screening and diagnosis to critically evaluate the evidence base for existing screening and diagnostic instruments.
2. Community Conversations held in Perth and Cairns with a total of 32 women to identify consumer and community members' perceptions relevant to the design and implementation of screening and diagnostic instruments for FASD in Australia.
3. A consultation process using a Delphi survey with input from 103 health professionals with expertise and/or experience in the screening or diagnosis of FASD. Using this process consensus was sought on the key components of screening and diagnostic instruments for FASD for use in Australia. This included screening and diagnostic methods, diagnostic criteria, criteria for referral and models of service delivery. A wide range of health professionals likely to be involved in FASD screening and diagnosis was included to ensure that the instruments developed will be practical and acceptable for use throughout Australia.
4. A consensus development workshop with members of the Australian FASD Collaboration to: consider the evidence from the systematic literature review, the Community Conversations and the Delphi survey; develop instruments for use in Australia for screening and diagnosis for FASD and recommend methods to validate the instruments and support their use.

KEY FINDINGS

The following key findings were identified from the literature review, Community Conversations, Delphi survey and consensus development workshop.

We found insufficient evidence on which to base the development of a valid screening instrument for FASD in Australia. Development of an instrument for targeted screening should be delayed until there is evidence to demonstrate its effectiveness, and adequate diagnostic and management services established to support the individuals undergoing screening and their families. Workshop participants concluded that the agreed criteria for referral could be used to guide health professionals on the need to refer individuals for specialist assessment.

There was consensus agreement that a multidisciplinary approach should be taken for the diagnosis of FASD in Australia. Diagnosis should be performed by specialist medical practitioners, ideally within an interdisciplinary team. Interdisciplinary diagnostic clinics are required in major cities, and outreach services could build local capacity and support diagnostic services in regional areas.

Consensus agreement was also reached on the need for a diagnostic instrument for FASD that is easy to understand and appropriate for use in Australia. A diagnostic instrument (Appendix A) was developed based on the University of Washington FASD Diagnostic Form, and includes a combination of elements from the University of Washington 4-Digit Diagnostic Code and the Canadian Guidelines for Diagnosis. The agreed diagnostic categories of FASD included in the instrument are Fetal Alcohol Syndrome, Partial Fetal Alcohol Syndrome and Neurodevelopmental Disorder – Alcohol Exposed. The agreed diagnostic categories of FASD and diagnostic criteria for each category included in the Australian diagnostic instrument are summarised in Table 1. Information on the use of the Australian FASD Diagnostic Assessment Form (Appendix A2) and the Australian FASD Diagnostic Assessment Summary Form (Appendix A3) is provided in the Guide to the Australian FASD Diagnostic Instrument (Appendix A1). The diagnostic instrument requires evaluation to ensure that it is appropriate for use in the Australian context, including among Indigenous Australians and those from culturally diverse backgrounds. This includes evaluation of the appropriateness of available population reference data for growth and facial features.

There was a recognised need for a comprehensive model of care for FASD to ensure that resources and support services are available for individuals undergoing diagnosis and their families, and to improve knowledge and awareness of FASD among health professionals.

Table 1 Australian FASD diagnostic categories and criteria

Diagnostic criteria[#]	Diagnostic category		
	Fetal Alcohol Syndrome (FAS)	**Partial Fetal Alcohol Syndrome (PFAS)**	**Neurodevelopmental 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: • 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: • 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 ≤ 10th percentile adjusted for gestational age, or postnatal height or weight ≤ 10th percentile	No deficit required[*]	No deficit required[*]
Central Nervous System (CNS) abnormality	At least 1 of the following: • clinically significant structural abnormality (e.g. head circumference ≤ 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) [‡]		

[†]University of Washington Lip-Philtrum Guides: http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm

[*]Not required for diagnosis but may be present

[#]Appropriate reference charts should be used, and other causes of growth deficit and CNS abnormality excluded.

[‡]Assessment of dysfunction based on evidence from standard validated assessments instruments interpreted by qualified professionals

CONCLUSIONS

There is an urgent need to increase national capacity for the prevention, diagnosis and management of FASD in Australia. This can be achieved through the development of standardised instruments for screening and diagnosis and guidelines for their use; training of health professionals; availability of interdisciplinary diagnostic services; and increased community awareness of the potential harms of alcohol use in pregnancy.

Targeted screening of children at risk of FASD is desirable but requires development and evaluation of an evidence based screening instrument. Screening should not be implemented until services are available to support individuals being screened and their families.

We have developed the basis for a nationally applicable, evidence-based, instrument for use in Australia for the diagnosis of FASD. Further work is required to develop detailed guidelines for diagnosis in Australia, to pilot test and evaluate the instrument, and to support its incorporation into cost effective strategies to improve the identification of FASD.

Inclusion in the instrument of standard diagnostic criteria for FASD will enable us to estimate the number of children with FASD in Australia and this in turn will inform programs for the management and prevention of FASD. Implementation of a standard diagnostic instrument would be an impetus for the improved reporting of FASD, improved training and awareness of FASD among health professionals, and appropriate planning for service development and prevention.

A comprehensive national implementation strategy is needed to ensure that the diagnostic instrument is embedded within evidence-based and locally relevant guidelines for the diagnosis and management of FASD. To optimise service provision and quality of life for individuals with FASD and their families, diagnosis should occur within a coordinated model of care that encompasses diagnosis, management and prevention of FASD and support for families. Within this model, interventions should also be available for women who use alcohol during pregnancy to help prevent alcohol-related harm in future pregnancies.

ABBREVIATIONS

Acronym/Abbreviation	Full Text
ADHD	Attention Deficit Hyperactivity Disorder
AERF	Alcohol Education and Rehabilitation Foundation
APSU	Australian Paediatric Surveillance Unit
ARBD	Alcohol-Related Birth Defects
ARND	Alcohol-Related Neurodevelopmental Disorders
CDC	Centers for Disease Control and Prevention (USA)
CNS	Central Nervous System
DoHA	Department of Health and Ageing (Commonwealth of Australia)
FAEE	Fatty Acid Ethyl Esters
FAS	Fetal Alcohol Syndrome
FASD	Fetal Alcohol Spectrum Disorders
HREC	Human Research Ethics Committee
IOM	Institute of Medicine (USA)
IQD	Inter-quartile Deviation
MCDS	Ministerial Council on Drug Strategy
ND-AE	Neurodevelopmental Disorder – Alcohol Exposed
NHMRC	National Health and Medical Research Council (Australia)
OFC	Occipito-frontal Circumference
PAE	Prenatal Alcohol Exposure
PEDS	Parents' Evaluation of Developmental Status
PFAS	Partial Fetal Alcohol Syndrome
PFL	Palpebral Fissure Length
SD	Standard Deviation
SE	Static Encephalopathy alcohol exposed
SNAPE	A 4-step guide to brief intervention: Smoking, Nutrition, Alcohol, Physical Activity and Emotional Health
T-ACE	Tolerance, Annoyed, Cut down, Eye opener A 4-item screening tool to identify of risky drinking during pregnancy
TWEAK	Tolerance, Worry, Eye opener, Amnesia, Cut down A 5-item screening tool developed to screen for risky drinking during pregnancy
UW	University of Washington
WAAHIEC	Western Australian Aboriginal Health Information and Ethics Committee
WHO ASSIST	The Alcohol, Smoking and Substance Involvement Screening Test (ASSIST) developed for the World Health Organization (WHO) to detect and manage substance use and related problems in primary and general medical care settings.

GLOSSARY OF TERMS

Term	Definition/Full Explanation
AUDIT-C	AUDIT is a simple 10-item questionnaire that is sensitive to early detection of risky and high risk drinking. AUDIT-C is a 3-item questionnaire that consists of the first three items of AUDIT.
Australian FASD Collaboration	A national group of highly qualified and experience health professionals, researchers and consumer and community members formed to guide the FASD Project. Also known as the 'Steering Group'.
Central nervous system abnormalities	Central nervous system (CNS) abnormalities are classified as structural, neurological, or functional Structural Abnormalities to brain structure including small head circumference (microcephaly) Neurological Includes seizures not attributable to a post natal insult (e.g. meningitis, trauma) Functional Cognitive impairment (decreased IQ; learning disabilities; developmental delays; problems with memory, attention or judgement etc
Community Conversation	Community Conversations are a way of engaging consumer and community members (using a world café process) to put forward their ideas about current research, gaps in research and priorities for future research.
Consumers	Patients and potential patients, carers, organisations representing consumers' interests, members of the public who are targets of health promotion programs and groups asking for research because they believe that they have been exposed to potentially harmful circumstances, products or services.
Delphi technique	The Delphi technique refers to a method that can be used to obtain the consensus opinion of a group of experts using a multi-stage survey.
Diagnosis	The act or process of identifying or determining the nature and cause of a disease, disorder or injury through evaluation of patient history, examination, and/or review of laboratory data.
Dichotomous questions	When a question has two possible responses, for example Yes/No, True/False or Agree/Disagree
Dysmorphology	Dysmorphology is a term to describe the study of human congenital anomalies (birth defects), particularly those affecting the morphology (the anatomy) of an individual. Dysmorphology literally means, "the study of abnormal form."
Epicanthic fold	The epicanthic fold is the skin fold of the upper eyelid covering the inner corner of the eye.
Executive functioning	The term executive function describes a set of cognitive abilities that control and regulate other abilities and behaviours – for example goal-directed behaviour, ability to initiate and stop actions, to monitor and change behaviour as needed, to plan future behaviour when faced with novel tasks and situations, anticipate outcomes and adapt to changing situations, form concepts and think abstractly.
FAS facial anomalies	The characteristic facial features of someone with Fetal Alcohol Syndrome include small palpebral fissures (eye openings), a thin upper lip, and an abnormal philtrum (absent or diminished ridges between the upper lip and nose).

FASD Project	Development of a diagnostic Instrument for FASD in Australia
Gestational age	The length of pregnancy from the first day of the last menstrual period
Health professionals	Includes medical practitioners, psychologists, dentists, pharmacists, physiotherapists, dieticians, nurses, occupational therapists, speech pathologists, social workers
Hypertelorism	Is an abnormally increased distance between the eyes
Interdisciplinary	An interdisciplinary team expands the multidisciplinary approach through collaborative communication (rather than shared communication) and interdependent practice. Members contribute their own professional specific expertise, but collaborate to interpret findings and develop a care plan. Team members negotiate priorities and agree by consensus.
Lifescripts	The Lifescripts initiative provides general practitioners with evidence-based tools and skills to help patients address the main lifestyle risk factors for chronic disease: smoking; poor nutrition; alcohol misuse; physical inactivity; and unhealthy weight. The initiative assists with the provision of tailored advice to patients on modifying their lifestyle. The AUDIT-C tool is included in the Lifescripts for pregnancy.
Likert scale	A multiple point response scale, for example (1= 'strongly agree', 2= 'agree', 3 = 'neither agree nor disagree', 4= 'disagree', 5= 'strongly disagree')
Lip Philtrum Guide	The Lip-Philtrum Guide as included in the UW Guidelines, is a 5-point pictorial rulers that is used to accurately assess and rank (from 1-5) the degree of philtrum smoothness (area between the upper lip and nose) and upper lip thinness.
Medical practitioner	Health professionals with a medical degree, such as physicians, paediatricians, general practitioners, psychiatrists
Meconium	Faeces present in the gut in utero and the earliest faeces produced by a baby
Microcephaly	Small head size for age and gestation or ≤ 3rd percentile
Multidisciplinary team	A FASD multidisciplinary team utilises the skills of health professionals from different disciplines (for example paediatrics, psychiatry, psychology, occupational therapy, speech pathology, social worker) with each discipline approaching the patient from their own perspective. Each team member conducts separate assessments. The team, directly or indirectly, then shares information and makes a joint plan regarding diagnostic category and future directions for patient care.
Nominal group technique	Established method for conducting structured group meetings
Palpebral fissure length	Horizontal length of the eye opening (distance from the endocanthion to the exocanthion)
Smooth philtrum	Diminished or absent ridges between the upper lip and nose
Polychotomous	When a question has more than two possible responses
Screening	The examination of apparently healthy individuals in order to identify those individuals who are at risk from a specific disorder to the extent that they would benefit from the use of a subsequent diagnostic test or procedure to confirm the presence or absence of the specific disorder. Screening can be applied across a whole population or used in a targeted way for select population groups.
Standard Drink Guide	The Standard Drink Guide is included in the NHMRC *Australian Guidelines to Reduce Health Risks from Drinking Alcohol.* (An Australian standard drink contains 10g of alcohol.)

Statistical terms used in this report	The **mean** (or average) of a set of data values is the sum of all of the data values divided by the number of data values. The **median** of a set of data values is the middle value of the data set when it has been arranged in ascending order, from the smallest value to the highest value. The **inter-quartile deviation** is a measure that indicates the extent to which the central 50% of values within the dataset are dispersed. It is based upon, and related to, the median. The **standard deviation** is a measure that summarises the amount by which every value within a dataset varies from the mean. **Percentiles** represent the value of a measurement or observation below which a specified percentage of observations fall. For example, if a 4-year-old boy's weight is in the 10th percentile that means that 10% of boys that age weigh less than he does and 90% of 4-year-old boys weigh more.
Steering Group	A national group of highly qualified and experience health professionals, researchers and consumer and community members formed to guide the FASD Project. Also known as the 'Australian FASD Collaboration'.
Teratogen	An agent (for example a drug, chemical or infection) that interrupts or alters the normal development of a fetus, including the development of the brain or other major organs. Examples of teratogens include alcohol, Rubella, mercury and thalidomide.
Thin vermillion border	Thin upper lip
World café process	A method which makes use of an informal café style format for participants to explore an issue by holding discussions in small groups. These are held in multiple rounds of 20-30 minutes and conclude with a summary of the discussions.

INTERNATIONAL FASD DIAGNOSTIC GUIDELINES

Throughout this report the following sources are referred to in the abbreviated form in parentheses.

Canadian Guidelines for Diagnosis (Canadian Guidelines)

Chudley, A. E., Conry, J., Cook, J. L., Loock, C., Rosales, T., & LeBlanc, N. (2005). Fetal Alcohol Spectrum Disorder: Canadian guidelines for diagnosis. *Canadian Medical Association Journal, 172*(5 Suppl), S1-S21.

Centers for Disease Control Guidelines for Referral and Diagnosis of FAS (CDC Guidelines)

Centers for Disease Control and Prevention. (2004). Fetal Alcohol Syndrome: Guidelines for referral and diagnosis. Atlanta: National Task Force on Fetal Alcohol Syndrome and Fetal Alcohol Effect.

Institute of Medicine Diagnostic Criteria (IOM Guidelines)

Stratton, K., Howe, C., & Battaglia, F. (Eds.). (1996). *Fetal Alcohol Syndrome: Diagnosis, epidemiology, prevent, and treatment*. Washington, D.C.: National Academy Press.

Hoyme Update of Institute of Medicine Diagnostic Criteria (Updated IOM)

Hoyme, H. E., May, P. A., Kalberg, W. O., Kodituwakku, P., Gossage, J. P., Trujillo, P. M., Buckley, D. G., Miller, J. H., Aragon, A. S., Khaole, N., Viljoen, D. L., Jones, K. L., & Robinson, L. K. (2005). A practical clinical approach to diagnosis of Fetal Alcohol Spectrum Disorders: Clarification of the 1996 institute of medicine criteria. *Pediatrics, 115*(1), 39-47.

University of Washington 4-Digit Diagnostic Code for FASD (UW Guidelines)

Astley, S. J. (2004). Diagnostic Guide for Fetal Alcohol Spectrum Disorders: The 4-Digit Diagnostic Code (Third ed.). Seattle: University of Washington.

LIST OF TABLES AND FIGURES

1. BACKGROUND

Key Points

- A third to a half of Australian women report drinking alcohol during pregnancy
- Alcohol may damage the fetal brain and the effects are life-long
- No nationally accepted diagnostic or screening instruments for FASD have been evaluated or validated for use in Australia
- Adoption of FASD screening and diagnostic instruments in Australia will enable earlier diagnosis and management thereby improving health outcomes and quality of life for individuals and families

In Australia, a large proportion of women of child-bearing age consume alcohol and often at high levels. Results from a 2007 study indicated that 59% of Australian women drank alcohol in at least one trimester of pregnancy (Colvin et al., 2007). This study also revealed that 47% of women did not plan their pregnancy. In a more recent study (Peadon et al., 2011) 34% of Western Australian women reported drinking in their last pregnancy.

Alcohol is a potent teratogen that may damage the developing fetal brain and other major organs *in utero*, often with devastating life-long consequences. Fetal Alcohol Spectrum Disorders (FASD) is an umbrella term covering a range of disorders caused by prenatal alcohol exposure (PAE), including Fetal Alcohol Syndrome (FAS), Partial Fetal Alcohol Syndrome (PFAS), Alcohol-Related Neurodevelopmental Disorders (ARND) and Alcohol-Related Birth Defects (ARBD) (Stratton, Howe & Battaglia 1996).

The effects of fetal alcohol exposure are life-long but may not be evident at birth. A minority of children exposed to alcohol during pregnancy will have FAS or PFAS, which can be identified by abnormal facial features, poor growth and abnormalities of the brain, with or without neurological problems. However, most children with FASD will look normal, despite having a range of learning, developmental and behavioural problems caused by damage to different parts of the brain. These can include a delay in reaching developmental milestones such as walking and talking; poor hand eye coordination and fine motor function; inability to complete complex tasks that involve planning, problem solving and judgement; and difficulties with motor function and social interactions. Children may also have poor academic performance, poor attention and lack the ability to control their emotions and a range of behavioural and mental health problems.

It is widely acknowledged that FAS is likely under-diagnosed and under-reported in both Indigenous and non-Indigenous children in Australia and that diagnosis is often delayed (Allen et al., 2007; Bower et al., 2000; E. Elliott et al., 2008; Harris & Bucens, 2003). FAS has been documented to occur more commonly in Indigenous children than in non-Indigenous children and there is concern that there may be some diagnostic bias in Indigenous children (Bower, et al., 2000).

Under-diagnosis may be related to health professionals' reluctance to ask about alcohol use and lack of knowledge about FASD. In a 2007 survey, just over half of Western Australian health professionals who cared for pregnant women routinely asked women about alcohol consumption in pregnancy and only 33% routinely provided information to pregnant women about the effects of alcohol use in pregnancy (Payne et al., 2011b). Only 18% of West Australian health professionals knew the four essential criteria for the diagnosis of FAS, 67% were concerned about stigmatising the child or the family with a diagnosis of FAS and only 6% felt very prepared to deal with FAS (Payne et al., 2011a). Similar findings applied to Australian paediatricians (E Elliott et al., 2006).

In a monograph presented to the Department of Health and Ageing (DoHA) in 2009 (Fetal Alcohol Spectrum Disorder in Australia: An Update), the Intergovernmental Committee on Drugs Working Group on Fetal Alcohol Spectrum Disorders recommended that criteria should be developed for the diagnosis of FASD in Australia and that a co-ordinated national screening and diagnostic service for children (and adults) with FASD be established. In March 2010, following a tender submitted by Clinical Professor Carol Bower and Professor Elizabeth Elliott, The Telethon Institute for Child Health Research and the University of Sydney were invited by DoHA to provide a quotation to supply a screening and diagnostic instrument for FASD.

An Australian FASD Collaboration, also known as the Steering Group, was established to guide the 'Development of a diagnostic instrument for FASD in Australia' (FASD Project). Led by Clinical Professor Carol Bower and Professor Elizabeth Elliott, the Collaboration is a group of highly qualified and experienced health professionals, epidemiologists, researchers, and consumer and community representatives committed to the prevention, diagnosis and care of individuals with FASD. They have established track records internationally and in Australia for the conduct and dissemination of research (including research in Aboriginal communities); health professional and community education; and advocacy about alcohol use in pregnancy and FASD. A list of the members of the Australian FASD Collaboration can be found in Appendix B (Vol 2 of this Report).

A Project Team comprising a Project Manager and Research Officer was appointed to take responsibility for the practical aspects of the project and the outputs required. This included conducting the literature review; coordinating the Community Conversations; undertaking the Delphi survey and holding a Steering Group Workshop to develop the screening and diagnostic instruments; analysing results; and producing all written reports. Following the resignation of the Research Officer in February 2011 a Project Analyst was appointed.

Since 1996, a number of screening and diagnostic instruments have been developed overseas (see International FASD Diagnostic Guidelines; page xiii). The Health Services Assessment Collaboration of New Zealand undertook a systematic review of the literature on the prevention, diagnosis and management of FASD, and included literature published from 1966 to July 2008. They concluded that there was insufficient high level evidence to identify which strategies may be suitable for implementation in New Zealand (L. Elliott et al., 2008).

In Australia, no nationally accepted screening or diagnostic instruments for FASD have been evaluated or validated; there are no national guidelines for the diagnosis or management of FASD; and there are few services for FASD. In 2011, pilot clinics in Sydney and Perth dedicated to the screening and diagnosis of FASD were funded by the Alcohol Education and Rehabilitation Foundation (AERF). The barriers to diagnosis of FASD are accentuated in rural, remote and Indigenous communities where access to relevant health services is limited or absent. Lack of diagnostic capacity for FASD means we are unable to quantify the problem and thus lack the evidence to advocate for much needed health professional training and diagnostic and therapeutic services. In addition, there is no means to advocate for clear referral pathways for affected children into diagnosis, therapy and life-long management.

OBJECTIVE

As specified in the Contract for Services between the Commonwealth of Australia (as represented by the DoHA); and the Telethon Institute for Child Health Research and the University of Sydney (on behalf of the Australian FASD Collaboration), the objective of the FASD Project was to develop an instrument that can be used to improve the identification and/or diagnosis of FASD in Australia. It is important to clarify the difference between screening and diagnosis, as the different needs of screening and diagnostic assessments require the development of separate instruments.

Diagnosis is the process of identifying or determining the nature and cause of a disease, disorder or injury through evaluation of patient history, examination, and/or review of laboratory data. Screening

can be defined as the examination of apparently healthy individuals in order to identify those individuals who are at risk from a specific disorder to the extent that they would benefit from the use of a subsequent diagnostic test or procedure to confirm the presence or absence of that specific disorder (Wald, 1991). Screening can be applied across a whole population or used within specific population groups.

Before a screening test is used, its performance must be known. Three indicators are required to describe the performance of a screening test:

i. the sensitivity of the screening test (the ability of the screening test to correctly identify with a positive result individuals who have the disorder);
ii. the specificity of the screening test (the ability of the screening test to correctly identify with a negative result individuals who do not have the disorder); and
iii. the positive predictive value of the screening test (the proportion of individuals who had a positive screening result who are diagnosed with the disorder) (Wald, 1991).

In addition tests used for screening must be cost effective and acceptable to health providers and the community. Adequate diagnostic services for FASD must also be available.

Outputs

In consultation with DoHA the following outputs were agreed:

1. Report on the outcomes of the first Steering Group teleconference
2. Report on a systematic literature review
3. Report on the Community Conversations
4. Report on the Delphi study methods and preliminary results
5. Final report

Potential outcomes

Adopting an instrument for the screening and diagnosis of FASD in Australia would:

1. Enable earlier diagnosis and management of FASD and improve health outcomes.
2. Improve functioning of persons diagnosed with FASD.
3. Improve the quality of life for individuals and families affected by FASD.
4. Enable more accurate estimates of the prevalence of FASD in Australia and planning for effective prevention strategies.

THE PROCESS

The FASD Project was conducted between August 2010 and August 2011. To ensure the requirements of the DoHA contract were achieved, a high level Project Management Plan, including a project risk analysis and timeline (Table 1.1), were developed. This was maintained by the Project Manager and used by the Steering Group to aid the process of communicating and integrating project work across interdisciplinary researchers, consumer and community representatives, health professionals and multiple organisations. It also helped us to clarify and agree to goals, identify resources needed, ensure accountability for results and performance, and focus on benefits to be achieved.

The Steering Group met via teleconference each month to provide high level expertise, advice and expert opinions on the following:

i. study design and methods;
ii. deliverables to DoHA; and
iii. documents for circulation to stakeholders.

The Steering Group held a two-day workshop in July 2011 to establish consensus on the content of the proposed Australian screening and diagnostic instruments for FASD. At this workshop the Steering Group considered the systematic literature review, the key issues arising from the Community Conversations and the results from the Delphi survey.

Figure 1.1 highlights the key components of the FASD Project.

Figure 1.1 FASD Project flow chart

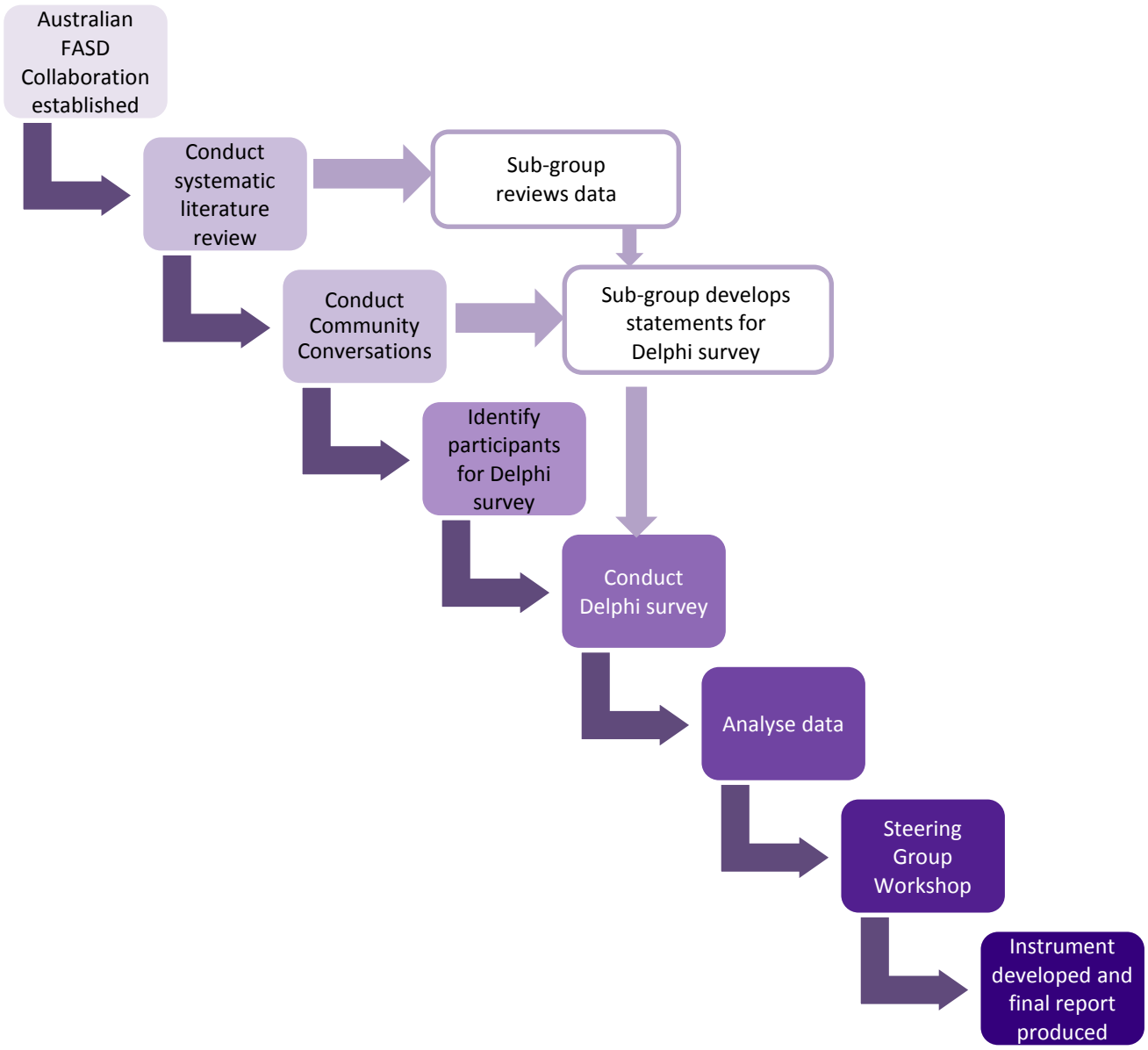

Table 1.1 FASD Project timeline

Date	Activity
March 2010	• DoHA requested a quotation from the Telethon Institute for Child Health Research and the University of Sydney • Australian FASD Collaboration (Steering Group) formed
April 2010	• The Telethon Institute for Child Health Research and University of Sydney submitted a quotation to DoHA on behalf of the Australian FASD Collaboration
May 2010	• Ethics approval from the University of Western Australia Human Research Ethics Committee (HREC)
June 2010	• Ethics approval from the Western Australian Aboriginal Health Information Ethics Committee (WAAHIEC) • Project plan developed, including dates for monthly Steering Group teleconferences • Contracts signed with amendments to project dates (August 2010 – August 2011)
July 2010	• Advertised for project staff
August 2010	• Project staff commenced work – based at the Telethon Institute for Child Health Research in Perth, Western Australia
September 2010	• **Deliverable 1:** Report on the outcomes from the first Steering Group teleconference
October 2010	• Systematic literature review conducted
November 2010	• **Deliverable 2:** Report on the systematic literature review
December 2010	• Development of Delphi statements and identification of participants for survey • Community Conversation – Perth, Western Australia
January 2011	• Development of Delphi statements and identification of participants for survey
February 2011	• On-line programming of Delphi survey • Community Conversation – Cairns, Queensland
March 2011	• Delphi survey pilot
April 2011	• Delphi survey round 1 (April 4 – 26) • **Deliverable 3:** Report on the Community Conversations
May 2011	• Delphi survey round 1 analysis (27 April – 25 May) • Delphi survey round 2 (26 May – 20 June)
June 2011	• Delphi survey round 2 analysis (23 June – 8 July) • **Deliverable 4:** Report on the Delphi study methods and preliminary results
July 2011	• Preparation of materials for Steering Group Workshop • Steering Group Workshop (25 – 26 July)
May 2012	• Clinician and consumer sub-group meetings and preparation of final report and review by the Steering Group • **Deliverable 5:** Final report

2. LITERATURE REVIEW: SUMMARY

Key Points

- The scope of this review was to identify in the published literature instruments for postnatal screening and diagnosis of FASD and clinical practice guidelines for their use
- The literature search did not identify any standardised screening or diagnostic instruments developed in Australia
- There is no internationally recognised gold standard for screening and/or diagnosis of disorders within the FASD spectrum
- It is important that individuals affected by alcohol exposure *in utero* are identified and assessed, as early as possible, preferably by a multidisciplinary team, so that tailored and specific treatment and support can be provided; the mother can be offered treatment; and the birth of affected siblings might be prevented

The Australian FASD Collaboration recognised the importance of prenatal screening. However the scope of this review was to identify literature describing instruments for postnatal screening and diagnosis and guidelines in order to inform the development of a postnatal screening and/or diagnostic instrument for use in Australia. A number of authors recommend that population-based screening should be implemented (Department of Health Western Australia, 2010; Gifford et al., 2010; Weiss et al., 2004). Others favour targeted screening (e.g., children in foster care or in the justice system) (Conry & Asante, 2010; Goh et al., 2008). Regardless of the type of screening, it should be introduced only if there are appropriate services for referral and follow-up (Canadian Association of Paediatric Health Centres, 2010; Gifford, et al., 2010; Weiss, et al., 2004).

In the literature search we did not locate any standardised screening or diagnostic instruments developed in Australia. A number of screening and diagnostic instruments that have been developed for use in other countries were identified. These include the Canadian Guidelines for Diagnosis (Canadian Guidelines), Centers for Disease Control Guidelines for Referral and Diagnosis of FAS (CDC Guidelines), Institute of Medicine Diagnostic Criteria (IOM Guidelines), Hoyme Update of Institute of Medicine Criteria (Updated IOM), and the University of Washington 4-Digit Diagnostic Code for FASD (UW Guidelines). The criteria for the diagnosis of FAS and PFAS are well defined and differ minimally between the guidelines identified; however the diagnostic instruments used to make the diagnosis differ. The criteria for diagnosis and the clinical description of other outcomes associated with fetal alcohol exposure vary between instruments, partly as a result of the wide range and complexity of outcomes in the FASD spectrum. The key criteria for a FAS diagnosis are relatively consistent: PAE,

growth abnormalities, characteristic facial features and neurological, functional and structural central nervous system (CNS) abnormalities, although the objective cut-off points used to determine abnormality vary between guidelines. The recommendation for a multidisciplinary team assessment is also common among the guidelines and criteria reviewed. Further guidelines were identified for use in the community and justice settings but they have not been rigorously evaluated in their application or success in diagnosing cases within the FASD spectrum.

A number of research papers identified in the systematic search provided further evidence of the applicability of technology such as digital photographs and computer software for the identification of facial characteristics. This was reported to be particularly useful for assessing cases from regions with limited access to clinicians trained in diagnosing FAS. However there is a need for norms for facial characteristics to be identified for Australia. Functional CNS domains were identified for evaluating and diagnosing disorders. Further to this, there is emerging evidence that brain imaging may be a useful, albeit expensive, method for measuring structural and neurological abnormalities.

Although there may be no gold standard for screening or diagnosis of disorders within the FASD spectrum, it is important that individuals affected by alcohol exposure are identified and assessed, preferably by a multidisciplinary team, in order to ascertain the specific limitations each individual has, so that tailored and specific treatment and support can be provided.

The full Literature Review can be found in Appendix D (Vol 2 of this Report).

3. COMMUNITY CONVERSATIONS: SUMMARY

Key Points

- The Community Conversations provided an important foundation for meaningful and inclusive consumer and community participation
- A standard set of questions (smoking, diet, exercise, alcohol) 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 for FASD
- Health professionals need education and training on FASD and how to speak to women about alcohol and 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 or breast feeding

Community Conversations were held in Perth (December 2010) and Cairns (February 2011) using a world café process (Brown & Isaacs, 2001) to ensure consumer and community members could be actively engaged in the FASD Project. The world café process is a method which makes use of an informal café style format for participants to explore an issue by holding discussions in small groups with a table facilitator. These are held in multiple rounds of 20-30 minutes and conclude with a summary of the discussions.

COMMUNITY CONVERSATION PURPOSE

The purpose of the Alcohol and Pregnancy Community Conversations was to provide input to the Steering Group when preparing the statements for the Delphi survey and for consideration when developing the screening and diagnostic instruments. The Community Conversations aimed to elicit information from consumer and the community members who are not as well informed about alcohol and pregnancy and FASD as the Steering Group consumer and community representatives, and in a manner that was open, friendly and inclusive. The Community Conversations were a foundation for meaningful and inclusive consumer and community participation in the FASD Project.

PROCESS

A total of 32 women participated in the Community Conversations. Each Community Conversation included two presentations. An overview of current alcohol and pregnancy research and information on FASD was presented by a researcher from the FASD Project. The second presentation was given by a parent/foster parent and was based on personal insight and lived experience, and provided compelling stories of living with children with FASD. The agenda was designed to allocate the majority of time to small group and whole group discussion, keeping presentations to a minimum.

QUESTIONS USED IN THE WORLD CAFÉ PROCESS

The questions for the Community Conversations were developed in consultation with the facilitator and members of the Steering Group and Project Team. The questions were designed to:

- elicit responses on what information women expected health professionals to provide with respect to alcohol use in pregnancy;
- facilitate discussion on the manner in which health professionals interact with women; and
- elicit responses on the level of information women would provide to health professionals about their alcohol use in pregnancy.

Evaluation forms from the Perth Community Conversation included participant comments that some questions appeared to be repetitive. The questions were revised for the Cairns Community Conversation.

STATEMENTS FROM COMMUNITY CONVERSATION PARTICIPANTS

Using the world café process participants in each small group were asked to write their individual statements on sticky notes and place them onto a large sheet of paper. All statements were transposed (no editing and in no particular order) into a spreadsheet and grouped into themes. The themes were counselling/support; family/community; feelings; health professionals; health professional training; how to ask questions of women; information for the public; information for women; language; resources; schools; timing; and general.

EVALUATION

In the evaluation process participants commented that the world café process gave them the opportunity to put forward their ideas. The Community Conversations were viewed positively by

participants and seen as a good opportunity to meet and interact with different people; learn about alcohol and pregnancy and FASD; and contribute to research. The Community Conversations were considered to be very informative and the voices and points of view of all participants were heard. The majority of participants stated that they would be interested in attending future Community Conversations.

> *"Good forum to share information"*
> Community Conversation participant

> *"The best thing was the opportunity to talk about something that is important to the community"*
> Community Conversation participant

> *"Everyone had their say"*
> Community Conversation participant

KEY ISSUES AND DISCUSSION

Issues arising from the Community Conversations specific to the development of the screening and diagnostic instruments:

- A standard set of questions (diet, smoking, drugs, lifestyle etc) that are asked by health professionals (general practitioner, obstetrician, midwife, maternal/child health nurse) of all pregnant women, no individuals or groups singled out
- Questions should be asked in private – not in front of partners or family
- Language should be culturally sensitive and questions should be easy to understand by all women and not use medical terminology
- Parents/guardians/kinship carers should be asked to provide informed consent before proceeding with screening and diagnosis for FASD
- The instrument needs to provide a guide and referral pathways
- The instrument should be appropriate for all Australian children (urban, rural and remote) and for different ages

Some statements were relevant to the topic of alcohol and pregnancy and FASD generally, but were not specific to the development of the screening and diagnostic instruments.

Issues arising from the Community Conversations that are related to the introduction of screening and diagnostic instruments and which require action on completion of the FASD Project:

- Health professionals need education and training on FASD and how to speak to women about alcohol and pregnancy
- Education and training should not be restricted to paediatricians – children in rural and remote areas will not have ready access to a paediatrician. Education and training is important for general practitioners, maternal and child health nurses, health workers
- 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 or breast feeding
- Resources for health professionals to be used in discussion with women should be visual (pictorial/DVD/models) and explain how alcohol reaches the baby and how it affects the baby

Women participating in the Community Conversations were concerned that health professionals were not providing them with information about alcohol and pregnancy and in some cases inconsistent messages were given to pregnant women. Women were of the view that without an education and training program for health professionals, information will not be provided and screening and diagnosis will not occur. As the screening of children will be undertaken by a range of health professionals, professional development in the area of FASD should be offered to health workers, maternal and child health nurses, general practitioners, paediatricians, psychologists and psychiatrists.

Important issues arising from the Community Conversations that are related to alcohol and pregnancy and FASD but outside the scope of the FASD Project:

- Prevention is the key to reducing the incidence of FASD – a national awareness campaign with information on alcohol use in pregnancy should be implemented across a range of media and venues. Examples included television, radio, social media (YouTube, Twitter), buses, clubs, bars, restaurants, nightclubs, Centre Link, Medicare, doctors and clinic waiting rooms.
- Education on alcohol use in pregnancy should be incorporated in the health curriculum in schools (12 – 16 year olds)

The full Community Conversations Report can be found in Appendix E (Vol 2 of this Report).

4. DELPHI STUDY METHODS

Key Points

- A Delphi survey was used to identify consensus on methods for screening and diagnosis of FASD
- Two survey rounds were conducted with a panel of 139 Australian and international health professionals and researchers
- The questionnaire included open-ended and closed-ended questions that sought to elicit opinions on different approaches to screening and diagnosis and identify alternative methods or viewpoints
- Findings from the Delphi survey were subsequently used to inform instrument development

OBJECTIVE

To establish consensus among health professionals on methods for screening and diagnosis of FASD in Australia.

BACKGROUND

The lack of clear empirical evidence to inform clinical decision-making in many areas of health care has motivated researchers and policy makers to use expert judgement and consensus as a basis for the development of clinical tools and practice guidelines (Biondo et al., 2008; Black et al., 1999; Jones & Hunter, 1995; McKenna, 1994). Formal consensus methods are frequently used to develop clinical guidelines as they provide a systematic and structured mechanism to evaluate and integrate multiple sources of evidence (Rycroft-Malone, 2001) and deal with uncertainty (Black, et al., 1999). In the absence of sufficient empirical evidence to support the design of a screening and diagnostic instrument for FASD in Australia, consensus methods enable a wide range of knowledge and experience to be considered in the instrument development process.

The Delphi consensus development method was designed to counter the disadvantages of unstructured group judgement processes, where there is direct interaction of participants, through the use of three main techniques: anonymity, multiple response rounds and feedback of the group response (Woudenberg, 1991). Classical Delphi studies use an open first round to determine the content of subsequent questionnaire rounds (Keeney et al., 2011). The modified Delphi design generally diverges from the classical Delphi method in the use of alternative means to derive the content of the initial

quantitative questionnaire round (Keeney, et al., 2011) while still allowing the collection of rich data based on multiple questionnaire iterations and revision due to feedback (Okoli & Pawlowski, 2004).

Formal consensus methods (Black, et al., 1999; Jones & Hunter, 1995) were used to develop screening and diagnostic instruments for FASD in Australia. For instrument development a two stage process was used to achieve agreement on the content and form of the proposed instruments:

i. a modified Delphi process was used to harness the insights of a large number of experts on the development of screening and diagnostic instruments for FASD in Australia without the need for face-to-face meetings; and
ii. findings from the modified Delphi process were subsequently used to inform instrument development using a consensus development workshop with members of the Steering Group.

Use of the modified Delphi process in the first stage of this study enabled the perspectives of experienced clinicians who understand the context, requirements and limitations of screening and diagnosis to guide the development of screening and diagnostic instruments.

> *"Great to see this level of consultation when deciding clinical policy. I am impressed by the process."*
> Delphi Study participant

QUESTIONNAIRE DEVELOPMENT

In the systematic review of the literature on FASD screening and diagnosis, which is described in detail in Appendix D (Vol 2 of this Report), a considerable existing body of knowledge relevant to the design of both screening and diagnostic instruments for FASD in Australia was identified. Relevant international literature includes detailed specifications for five different guidelines for the diagnosis of FASD (see International Diagnostic Guidelines: page xiii). Thus we used a modified or reactive Delphi process, which requires participants to react to previously prepared information rather than generate the initial questionnaire information (McKenna, 1994), to enable existing knowledge to be used as a foundation for the development of screening and diagnostic instruments in Australia. A modified Delphi process generates a more highly structured initial questionnaire.

The Delphi questionnaire was developed in collaboration with a sub-group of the Steering Group. A total of 174 Likert statements were designed to elicit opinion on the key components of screening and diagnostic instruments for FASD, and explore opinions on screening and diagnostic processes in

Australia, including referral guidelines and diagnostic criteria. The Likert statements were developed to represent approaches to screening and diagnosis in the literature, including the methods used in existing instruments, and key issues identified in the Community Conversations. Agreement with the statements was evaluated on a 5-point Likert scale (1='strongly agree', 2='agree', 3='neither agree nor disagree', 4='disagree', 5='strongly disagree'). Due to the broad scope of the questionnaire content and the varied professional background of participants, an additional response option 'no comment' was provided to enable participants to indicate if individual statements were outside their area of expertise.

To ensure that participant responses were not constrained by the use of a modified Delphi process, each area of questioning was accompanied by open-ended questions that sought to elicit both the identification of alternative methods or viewpoints and general comments on each area of enquiry. Participants were specifically encouraged to provide comments on their responses and identify where the statements did not adequately reflect their beliefs about screening and diagnosis. The collection of supporting qualitative data is essential to understand panel perspectives on screening and diagnosis, and ensure that subsequent questionnaire rounds adequately represent the diversity of opinion present. A total of 59 open-ended questions and 67 dichotomous or polychotomous questions were included in the questionnaire to enable participants to specify alternative approaches and elicit comment or further information on the concepts represented. Table 4.1 provides a summary of the number and type of questions included in each section of the round 1 questionnaire.

"This is a very concise piece of work which I am very pleased to have been able to participate in. I know this will lead to further vital input in developing a much needed national approach to this problem for both early intervention and prevention when the full extent of the problem has been realised. Best wishes to you all"
Delphi Study participant

"An excellent survey. It highlights the difficulty of diagnosis and the problems with diagnostic categories"
Delphi Study participant

"The introduction estimated completion time of 45mins but I found that it took 2 hours by the time I wrote comments."
Delphi Study participant

Table 4.1 Round 1 questionnaire composition by section and question type

Questionnaire Section	Likert statements (n)	Other closed questions[†] (n)	Open-ended questions (n)	Total (n)
1. Demographic information	0	16	0	16
2. Screening programs	19	2	5	26
3. Targeted screening	18	2	4	24
4. Screening providers	17	0	1	18
5. Screening methods (part 1)	8	6	3	17
6. Screening methods (part 2)	11	3	6	20
7. Screening methods (part 3)	17	1	2	20
8. Definition of abnormal screening findings	22	6	13	41
9. Criteria for conducting a full diagnostic evaluation	23	1	3	27
10. Diagnostic systems and guidelines	0	21	8	29
11. Diagnostic processes	10	1	3	14
12. Diagnostic criteria for Fetal Alcohol Syndrome	22	5	6	33
13. Diagnostic criteria for other FASD	7	3	4	14
14. Acknowledgement and feedback	0	2[‡]	1	3
Total	**174**	**69**	**59**	**302**

[†]Other closed questions included dichotomous and polychotomous questions

[‡]These 2 questions sought participant consent regarding acknowledgement, and did not collect data related to the objective of the questionnaire

ONLINE DEVELOPMENT

The Delphi survey was administered as an online password-protected questionnaire. The online questionnaire content was translated into HTML (hypertext mark-up language), and the website was created using PHP (PHP: Hypertext Pre-processor, The PHP Group, 2001-2011) which is a widely-used scripting language that can be embedded into HTML. The script files were created using Komodo Edit version 6.0.2. The questionnaire content was delivered to participants using HTML forms which enabled responses to be saved to and retrieved from a secure MySQL database. Due to the length of the questionnaire, the online program structure was designed so that participants could complete the questionnaire over a number of sessions. Following the secure login screen, a contents page was developed to provide access to each of the 14 questionnaire sections, summarise the completion status of each section, and display an overall progress indicator. The round 1 questionnaire (Appendix F: Vol 2 of this report) was hosted on a secure web server (http://fasd.ichr.uwa.edu.au) located at the Telethon Institute for Child Health Research.

QUESTIONNAIRE PRETESTING

Prior to pretesting, the proposed questionnaire was reviewed by three members of the Steering Group and researchers experienced in using the Delphi technique, to ensure that the questionnaire was clear, comprehensible and had a consistent format (Keeney et al., 2001; Kingston et al., 2009). The questionnaire was subsequently pretested with a group of 16 clinicians and researchers, including members of the Steering Group who were not participating in the Delphi survey. Pretesting was conducted to ensure that the content was clear and comprehensible; that the online format functioned on a range of web browsers and computer operating systems; and that the questionnaire had face validity as determined by the reviewers. Pretesting indicated that the online format functioned well and that the questionnaire was clear, comprehensible and collected the information required to address the study aims.

Following pretesting of the round 1 questionnaire, six new statements were included and one redundant statement was deleted. Several minor revisions to statement wording were also made to improve clarity or distinctions between statements. A mechanism was introduced to allow participants to indicate that entire diagnostic criteria sections were outside their expertise in order to make completion of the questionnaire easier for those who did not wish to complete these sections. Based on pilot testing, the round 1 questionnaire was estimated to take 45 minutes to complete.

PANEL COMPOSITION

The Delphi study method requires recruitment of a panel with specific expertise that is relevant to the study objective (Keeney, et al., 2001; Kingston, et al., 2009), because a representative population sample is unlikely to be sufficiently knowledgeable to provide accurate responses to the questions (Okoli & Pawlowski, 2004). Panel members for the Delphi survey were recruited based on their expertise or experience in the screening or diagnosis of FASD.

Delphi studies are typically conducted with panels comprising 20 or fewer individuals (Ludwig, 1997; Okoli & Pawlowski, 2004), although there is no agreement regarding the minimum or maximum number of individuals that should form a panel (Keeney et al., 2006; Williams & Webb, 1994). Small panels may not provide a representative pool of judgement, and large panels can result in a low response rate (Hsu & Sandford, 2007). Recommendations for Delphi panel sizes range from 15 to 60 individuals, with greater numbers recommended if more than one reference group is involved (Hasson et al., 2000; Hsu & Sandford, 2007). The development of screening and diagnostic instruments for FASD

must involve the range of health professionals who provide services for individuals who may have FASD, including specialist and generalist medical practitioners, nurses and allied health professionals. Thus, a large panel was recruited to allow representation of perspectives from the range of health professionals who may be involved in screening and diagnosis.

RECRUITMENT

Three recruitment strategies were used to identify panel members and issue invitations to participate. All medical practitioners who had reported a diagnosis of FAS to the Australian Paediatric Surveillance Unit (APSU) in the surveillance study conducted in the period 2001-2004 were invited to participate (EJ. Elliott, et al., 2008). In addition, members of the Steering Group identified individuals known to have expertise or experience in the screening or diagnosis of FASD, and these individuals were contacted via email and invited to participate. Other clinicians and researchers who have expertise or experience in the screening or diagnosis of FASD were also identified through canvassing members of relevant professional associations, including national, state and territory medical and nursing organisations. The Lead Investigators reviewed and approved the final list of panel members. Recruitment aimed to obtain a predominantly Australian expert panel, although a small number of international experts were identified and included.

A formal invitation was sent via email to 57 medical practitioners who had reported a diagnosis of FAS to the APSU, 128 health professionals who were identified by the Steering Group to have experience or expertise in FASD screening and/or diagnosis, and 35 health professionals who responded to calls for volunteers and who had relevant experience or expertise in FASD screening or diagnosis. Of these 220 invited panel members, 81 either did not respond to the email invitation to participate in this study, or declined to participate and were excluded from the study. In the case of panel members recruited through the APSU, all were included in the study apart from those who requested to withdraw from the study. All other panel members were required to actively indicate their agreement to participate in the study. The first round of the Delphi survey commenced with 139 panel members.

> *"I think the FASD questionnaire was very well researched and put together"*
> Delphi Study participant

This study was approved by the Human Research Ethics Committee (HREC) at the University of Western Australia and the Western Australian Aboriginal Health Information and Ethics Committee (WAAHIEC).

QUESTIONNAIRE ADMINISTRATION AND FOLLOW-UP

An email containing a personal username and password and a link to the Delphi survey website was sent to all panel members who were asked to complete the first round within two weeks. This is consistent with Delbecq and co-workers (1975) who suggest allowing two weeks for Delphi subjects to respond to each round. Panel members received a reminder about questionnaire completion seven days following the initial email and again approximately one day prior to the round 1 deadline.

Follow-up emails distinguished between panel members who had completed the entire questionnaire, completed part of the questionnaire, or had not yet logged onto the questionnaire website. This enabled targeted follow-up, so that those who had completed the questionnaire could be thanked for their contribution, those who had started the questionnaire could be encouraged to complete it, and those who had not yet logged onto the website could be encouraged to respond and reminded of the importance of their participation.

Due to a slower than anticipated response to the round 1 questionnaire, the deadline for completion was extended to include three additional working days as well as the adjacent five-day Easter public holiday period. On the original round 1 closing date the response rate was approximately 57% (79/139 completed, with an additional 13 started but not completed). A follow-up email was distributed to all panel members who had not yet completed the questionnaire to advise them of the extension to the round 1 deadline and encourage them to participate.

Persistent follow-up of panel members is critical to achieving a high response rate (Keeney, et al., 2006). In an attempt to improve the round 1 response rate, panel members who had neither logged on to the study web site nor contacted the Project Manager to advise their intent to complete the questionnaire before the extended round 1 closure date were targeted for additional follow-up. Steering Group members performed personal follow-up when possible for the 15 panel members whom they had personally recruited. Although telephone contact details were not originally collected, internet and project correspondence searches identified telephone contact details for an additional 28 non-respondents who were recruited via the APSU or the call for volunteers. One contact attempt was made to follow-up these 28 panel members at least five days before the closure of round 1. Following closure of round 1, panel members who had completed or part-completed the questionnaire were thanked for their contribution and advised of the anticipated date of the round 2 questionnaire administration. Panel members who did not complete the round 1 questionnaire were thanked for their interest and advised of their exclusion from future study rounds.

DATA ANALYSIS

Data entered from the online questionnaires were automatically saved in a MySQL database located on a secure webserver. Following the closure of each questionnaire round the data were exported from the MySQL database in text format and stored in a Filemaker Pro database. PASW Statistics version 18.0.1 was used to analyse the quantitative response data. Results for each statement were summarised using the per cent agreement, median and interquartile deviation (IQD) of responses to provide indicators of central tendency and dispersion (Rycroft-Malone, 2001). Item-level analyses aimed to determine which statements had achieved consensus and which required revision. Item non-response was also assessed. The Chi-square Test of Independence was used to test the association between two categorical variables.

Qualitative data were independently coded and analysed by the Project Analyst and Project Manager using qualitative inductive content analysis methods (Elo & Kyngas, 2008; Streubert-Speziale & Carpenter, 2003). Data from each open-ended question were reviewed alongside the quantitative data and coded inductively, based on the underlying meaning of the data. Responses were read line by line prior to coding for each question, and significant words and phrases were identified. The main intent of each response was then conceptualised and coded. For questions where a sufficient number and depth of responses existed, first level codes were also reviewed and categorised based on their characteristics or properties (Hsieh & Shannon, 2005; Streubert-Speziale & Carpenter, 2003). Both analysts' independent coding schemes were documented and then reviewed for consistency to ensure credibility and trustworthiness of the analysis process (Elo & Kyngas, 2008). For all 59 open-ended questions included in round 1, the review of first-level coding identified a high level of consistency of coding. For several questions some differences were identified in the categorisation of first level codes, and in these cases the data were jointly reviewed and agreement was reached on the final coding scheme to be applied.

CONSENSUS

The appropriate criteria to define the level of consensus in Delphi studies vary based on considerations unique to each study, with definitions based on percentage of agreement in published studies ranging between 51% and 100% (Keeney, et al., 2006; Kingston, et al., 2009; Williams & Webb, 1994). In addition to indicators of central tendency, definitions of consensus can also incorporate measures of spread. The response IQD provides an additional measure that can be used to ensure that bimodal distributions are excluded from the definition of consensus.

Prior to the commencement of data collection, the Steering Group agreed on the criteria for consensus. Statements were considered to reach consensus when 70% or more of valid responses lie within two categories on the 5-point Likert scale. Consensus agreement required 70% of responses to be 'strongly agree' or 'agree', and consensus disagreement required 70% of responses to be 'strongly disagree' or 'disagree'. In addition, statements were considered to reach a high level of consensus when, as well as meeting the 70% threshold for consensus, the statement IQD was less than or equal to one, meaning that the middle 50% of responses were also located on two adjacent rating scale points. Similarly, consensus agreement or disagreement for the five items that assessed agreement using a 3-point scale ('yes', 'unsure', 'no') required either 70% agreement ('yes') or 70% disagreement ('no').

QUESTIONNAIRE REVISION PROCESS

In order to provide Delphi participants with feedback and an opportunity to revise their responses, Delphi studies should be conducted over at least two rounds (Keeney, et al., 2006). Due to the broad scope of the questionnaire and the high response burden this placed on participants, revisions were made to the questionnaire following round 1 to reduce the length of the questionnaire.

The following criteria were used to determine whether items in the round 1 questionnaire had reached agreement, were re-assessed or were rejected:

i. if at least 70% of participants agreed or strongly agreed with an item, it was considered endorsed and removed from further rounds;
ii. if at least 60% but less than 70% of participants agreed or strongly agreed with an item, it was re-administered in the second questionnaire round either in its original form, or, if indicated based on the qualitative data, in a modified form;
iii. if less than 60% of participants agreed or strongly agreed with an item and qualitative comments also indicated a low level of support for the item, or for screening or diagnostic methods items, if a corresponding alternative item from within the question area reached a significantly higher level of consensus, the item was rejected and removed from the round 2 questionnaire.

In addition, due to the large number of inter-related criteria for referral for diagnosis that we examined in round 1, items that reached at least 70% agreement were re-administered in round 2, and agreement with these criteria was reassessed. Finally, if qualitative data indicated that the questionnaire did not enable representation of a relevant alternative perspective, a new item was included in the round 2 questionnaire.

As outlined above (criterion iii), the inclusion of statements that represented alternative methods for screening and diagnosis in the round 1 questionnaire required consensus to be evaluated for each area of questioning, rather than for individual statements alone, as all statements were not expected to reach consensus. As such, decisions about the achievement of consensus, feedback of results, and exclusion of statements from future rounds were performed separately for each question area. Due to the length of the questionnaire, statements that achieved consensus in round 1 were generally excluded from further quantitative evaluation in round 2 as indicated above (criterion i). However, in several questionnaire sections where clear consensus was not reached, re-inclusion of statements that had achieved consensus allowed participants to reconsider their responses.

The provision of feedback to participants is a fundamental feature of the Delphi process. Key results from round 1 were summarised in the round 2 questionnaire to enable broad feedback through the inclusion of group-level findings. Feedback was interspersed with related follow-up questions that explored remaining areas of uncertainty or areas in which consensus was not reached. The round 2 questionnaire included for each area of questioning a summary of group-level results for statements that had reached consensus, a summary of individual and group-level results for statements that had not reached consensus, a small number of questions that required rating or re-rating, and an opportunity to provide further comment on the perspectives represented.

> *"I have found this survey to be above my expectations, and in particular I like the feedback from my previous contribution being available to me as I progress through the second survey."*
> Delphi Study participant

> *"High level model of what can be achieved with Delphi qualitative research process. I await the final product with interest."*
> Delphi Study participant

> *"A little more time between sending the initial questionnaire and the less than subtle reminders would be nice. An 11 day turnaround time is not long for busy clinicians with on-call responsibilities."*
> Delphi Study participant

The revised questionnaire structure allowed a reduction in length of the questionnaire, as well as a mechanism for participants to provide feedback on areas of consensus and ensure that responses to the round 1 questionnaire had been accurately interpreted. Table 4.2 provides a summary of the number and type of questions included in each section of the round 2 questionnaire.

Table 4.2 Round 2 questionnaire composition by section and question type

Questionnaire Section	Likert statements (n (repeat*))	Other closed questions[†] (n)	Open-ended questions (n)	Total (n)
1. Screening programs	13 (4)	0	3	16
2. Screening methods	4 (1)	0	1	5
3. Definition of abnormal screening findings	8 (1)	0	1	9
4. Criteria for conducting a full diagnostic evaluation	3 (1)	0	2	5
5. Diagnostic criteria for FAS and PFAS	3 (3)	0	1	4
6. Diagnostic criteria for other FASD	5 (4)	0	1	6
7. Existing diagnostic guidelines and diagnostic processes	4 (2)	0	2	6
8. Acknowledgement and feedback	0	2[‡]	1	3
Total	**40 (16)**	**2**	**12**	**54**

[†]Other closed questions included dichotomous and polychotomous questions

[‡]These 2 questions sought participant consent regarding acknowledgement, and did not collect data related to the objective of the questionnaire

*Number of round 1 statements re-administered in round 2

ROUND 2 QUESTIONNAIRE PRETESTING

The round 2 Delphi questionnaire was reviewed by three members of the Steering Group and researchers experienced in using the Delphi technique to establish face validity and ensure that statements were clear and comprehensible. The round 2 questionnaire was then pretested with a group of 10 clinicians and researchers, including members of the Steering Group who were not participating in the Delphi process. Following pretesting, one new statement was included in the questionnaire. Several minor revisions to the wording of instructions were also made to improve clarity. Based on pilot testing, the round 2 questionnaire (Appendix F: Vol 2 of this Report) was estimated to take 20-25 minutes to complete.

> *"Excellent survey. It highlights the difficulty of diagnostic categories. There is a need for more definitive terminology that captures the diversity and continuum of disabilities associated with alcohol exposure prenatally."*
>
> Delphi Study participant

ROUND 2 QUESTIONNAIRE ADMINISTRATION AND FOLLOW-UP

An email containing a personal username and password and a link to the Delphi survey website was sent to all 103 participants who completed the round 1 questionnaire. In response to feedback indicating that the response period for round 1 should have been longer; participants were asked to complete the second questionnaire within 4 weeks.

Follow-up of non-respondents in round two utilised email, personal communication and telephone to maximise the response rate. On the original round 2 closing date, the response was approximately 77% (79/103 completed, with an additional 7 started but not completed). The deadline for completion of round 2 was extended by one week to improve the response rate. Data collection ceased following round 2 due to the in-principle achievement of consensus for all main areas of inquiry, and the likelihood of low response rates if further rounds were conducted (Keeney, et al., 2001).

> *"Great study providing much information to further look up and understand towards improving knowledge in the field of FASD – Thank you"*
> Delphi Study participant

5. DELPHI STUDY RESULTS

Key Points

- The response rates for rounds 1 and 2 were 74.1% and 85.4% respectively
- There was consensus agreement for targeted screening at birth (75.3%) and in childhood (84.5%), as well as the need for a screening checklist at birth (83.7%) and in childhood (88.1%)
- There was consensus agreement on the diagnostic criteria for FAS (82.1%) and PFAS (75.8%)
- Most participants (89.9%) agreed that a multidisciplinary diagnostic clinic should be available in major cities

PARTICIPATION

Response rates by recruitment strategy and professional group are summarised in Table 5.1 based on information collected at the time of recruitment. Among round 1 respondents, 93 completed the whole questionnaire, and 11 completed between 1 and 10 of the 14 questionnaire sections. One round 1 respondent who only completed the demographic questions was excluded from both the analysis and participation in subsequent rounds. Participants were defined as respondents who completed at least 2 of the 14 questionnaire sections.

Overall, individuals who were passively recruited via the APSU were the least likely to respond to both round 1 and 2 questionnaires. Individuals who were actively recruited by Steering Group members were the most likely to respond to the round 1 questionnaire (79.4%), followed by those recruited through professional bodies (71.0%). Non-response did not vary significantly by professional group, with an overall response of 73.2% among medical practitioners and 76.2% among other health professionals in round 1 (χ^2=0.14 p=0.71), and 85.9% among medical practitioners and 84.4% among other health professionals in round 2 (p=1.00, Fisher's exact test).

Table 5.1 Study participation by recruitment strategy and professional group[†]

Recruitment strategy	Round 1		Round 2	
	Invited n	Responded n (%)	Invited n	Responded n (%)
Steering group				
medical practitioner	34	27 (79.4)	27	25 (92.6)
other	34	27 (79.4)	27	22 (81.5)
Professional bodies				
medical practitioner	23	17 (73.9)	17	16 (94.1)
other	8	5 (62.5)	5	5 (100.0)
Passively enrolled APSU				
medical practitioner	40	27 (67.5)	27	20 (74.1)
Total	139	103 (74.1)	103	88 (85.4)

[†]Based on data collected on professional group at the time of recruitment

PARTICIPANT CHARACTERISTICS

The demographic characteristics of participants are described in Table 5.2. Participants were predominantly from Western Australia, New South Wales and Queensland, and almost three quarters were female. Paediatricians were the most frequently represented professional group, and most participants practised in metropolitan areas.

Table 5.2 Socio-demographic characteristics of round 1 participants

Characteristic	Round 1 (%) (n=103)	Round 2 (%) (n=88)
Country of residence		
Australia	92.2	92.0
other	7.8	8.0
State or Territory of residence		
WA	27.0	30.3
NSW	25.8	26.3
QLD	25.8	23.7
SA	6.7	5.3
NT	5.6	3.9
VIC	4.5	5.3
TAS	3.4	3.9
ACT	1.1	1.3
Sex		
female	74.8	75.0
male	25.2	25.0

Table 5.2 Socio-demographic characteristics of round 1 participants (con't)

Characteristic	Round 1 (%) (n=103)	Round 2 (%) (n=88)
Occupation		
paediatrician	43.7	40.9
allied health professional[†]	14.6	14.8
midwife/nurse	10.7	11.4
psychiatrist	8.7	8.0
general practitioner	6.8	8.0
clinical geneticist	4.9	5.7
other medical practitioner/specialist[‡]	3.9	4.5
health researcher	3.9	4.5
Aboriginal health/community worker	2.9	2.3
*Location of practice**		
metropolitan	62.1	62.5
regional	31.1	31.8
rural	29.1	26.1
remote	25.2	26.1

[†]Includes psychologist, occupational therapist, physiotherapist, social worker, speech pathologist

[‡]Includes obstetrician, paediatric senior registrar, addiction medicine specialist, forensic clinician

*Some participants practised in more than one location

Over three quarters of participants (77.2% in round 1 and 76.1% in round 2) reported experience in either the screening or diagnosis of FASD. Participants reported up to 30 years experience in screening, and up to 40 years experience in diagnosis (Table 5.3). Only a quarter of participants had completed specific training on the diagnosis of FASD.

Table 5.3 Participant experience in FASD screening and diagnosis

Characteristic	Round 1 (%) (n=103)	Round 2 (%) (n=88)
Involved in screening	29.7	29.5
Years experience in screening (n=29)		
≤ 5	41.4	48.0
6-10	27.6	24.0
≥ 11	31.0	28.0
Involved in diagnosis	43.6	43.2
Years experience in diagnosis (n=43)		
≤ 5	30.2	32.4
6-10	23.3	24.3
≥ 11	46.5	43.2

Table 5.3 Participant experience in FASD screening and diagnosis (con't)

Characteristic	Round 1 (%) (n=103)	Round 2 (%) (n=88)
Number of cases diagnosed (n=43)		
≤ 5	32.6	32.4
6-10	14.0	16.2
11-15	14.0	13.5
16-20	20.9	18.9
≥ 21	18.5	18.9
Involved in contributing to diagnosis	69.3	69.8
Years experience contributing to diagnosis (n=69)		
≤ 5	34.8	37.3
6-10	26.1	28.8
≥ 11	39.1	33.9
Number of cases contributed to diagnosis (n=67)		
≤ 5	38.8	38.6
6-10	17.9	17.5
11-15	17.9	17.5
16-20	10.4	8.8
≥ 21	15.0	17.5
Completed specific training on FASD screening	15.8	18.6
Completed specific training on FASD diagnosis	25.7	27.9

Survey results for questionnaire rounds 1 and 2 are presented below by area of questioning, with statements identified in the tables by questionnaire section (S) and question number (Q). Likert statement scores ranged from 1 ('strongly agree') to 5 ('strongly disagree'). Statement agreement frequencies (responses of 'strongly agree' or 'agree' on the 5-point Likert scale) are presented in bold when agreement frequency equals or exceeds the 70% consensus level, and IQD are presented as a summary indicator of the spread of responses.

SCREENING PROGRAMS

Targeted screening in childhood received the highest level of support in round 1 (Table 5.4). There was neither clear endorsement nor rejection of the different screening strategies at birth, and all four questions on screening coverage were re-administered in round 2. Only targeted screening reached consensus agreement in round 2 (Table 5.5). Participant comments indicated support for universal screening as an ideal and ethical approach to decrease the risk of missing cases and enable early diagnosis and early intervention. However, targeted screening of high risk groups was described as a more feasible and effective approach. Concerns were raised about the sensitivity, specificity and cost-effectiveness of any screening program. Participants commonly identified the need for provider training and the availability of adequate intervention services to accompany any screening program. The

universal collection of maternal history of alcohol consumption during the prenatal period and at birth was deemed necessary to enable targeted follow-up of exposed children and preventive interventions at the individual and community level.

Table 5.4 Round 1 statement ratings: screening program timing

Statement	N	Agree (%)	Median	IQD
S2Q1. Screening for FASD at birth should be universal	95	57.9	2	3
S2Q2. Screening for FASD at birth should be targeted	95	69.5	2	2
S2Q3. Screening for FASD in childhood should be universal	93	48.4	3	3
S2Q4. Screening for FASD in childhood should be targeted	93	**78.5**	2	1

Table 5.5 Round 2 statement ratings: screening program timing

Statement	N	Agree (%)	Median	IQD
S1Q6. Screening for FASD at birth should be universal	85	57.6	2	3
S1Q7. Screening for FASD at birth should be targeted	85	**75.3**	2	2
S1Q12. Screening for FASD in childhood should be universal	84	40.5	3	2
S1Q13. Screening for FASD in childhood should be targeted	84	**84.5**	2	1

Consensus was reached for five of the six statements on the content of screening programs at birth (Table 5.6) and all nine statements on the components of screening in childhood (Table 5.7). Over half the participants (56.3%) indicated that the question about measuring fatty acid esters in meconium was outside their area of expertise. The limited support for testing fatty acid esters in meconium was consistent with participant comments which indicated: the inability of this test to reflect alcohol consumption in the early prenatal period; its lack of sensitivity for lower levels of alcohol consumption which may still pose a risk to the fetus; the lack of an agreed scoring method; and that it is inappropriate for low risk settings.

General comments indicated that screening for maternal alcohol use, although potentially unreliable, was the most important factor in FASD screening. Comments also highlighted a range of difficulties in the provision of screening, including: the evolving clinical presentation of FASD; that facial anomalies were specific to FAS; the difficult nature of screening for PAE; the difficulty in the detecting CNS dysfunction in very young children; the need for a clearer distinction between screening and diagnosis; and the lack of services for referral and intervention for children who screened positive. Participants also highlighted the need for training of screening providers, and that screening programs must be cost effective, efficient, and acceptable to the population being screened.

Table 5.6 Round 1 statement ratings: components of screening at birth

Statement: screening at or around birth should assess and record:	N	Agree (%)	Median	IQD
S2Q5. prenatal alcohol exposure	100	**98.0**	1	0
S2Q6. birth weight, length and head circumference	98	**100.0**	1	1
S2Q7. fatty acid esters (FAEE) in meconium collected within 72 hours of birth	42	45.2	3	2
S2Q8. characteristic FAS facial anomalies	97	**97.9**	1	1
S2Q9. birth defects	97	**96.9**	1	1
S2Q10. evidence of withdrawal from alcohol or other drugs	98	**98.0**	1	1

Table 5.7 Round 1 statement ratings: components of screening in childhood

Statement: screening for FASD in childhood should assess and record:	N	Agree (%)	Median	IQD
S2Q12. prenatal alcohol exposure	98	**96.9**	1	1
S2Q13. growth (height and weight)	97	**97.9**	1	1
S2Q14. head circumference	94	**98.9**	1	1
S2Q15. developmental delay	97	**99.0**	1	1
S2Q16. neurological signs	95	**92.6**	1	1
S2Q17. functional central nervous system abnormalities (e.g. cognition, behaviour disorders)	95	**97.9**	1	1
S2Q18. hearing and vision	93	**91.4**	1	1
S2Q19. characteristic FAS facial anomalies	97	**97.9**	1	1
S2Q20. birth defects	97	**95.9**	1	1

Agreement with additional suggested components of screening and participant comments that most of the screening assessments described are a part of routine care was assessed in round 2, and findings are summarised in Table 5.8.

Table 5.8 Round 2 statement ratings: components of screening

Statement	N	Agree (%)	Median	IQD
Screening for FASD at or around birth should also assess and record:				
S1Q1. family history of FASD or developmental delay	86	**91.9**	2	1
S1Q2. evidence of CNS dysfunction including irritability, feeding difficulties or other neurological signs	84	**89.3**	2	1
S1Q3. Most of the information required for FASD screening at birth is routinely collected at birth	79	54.4	2	2
S1Q4. Screening for FASD at birth primarily requires health professionals to assess prenatal alcohol exposure and consider it as a potential cause of other relevant abnormalities identified	86	**87.2**	2	1
S1Q5. A checklist is needed to support the implementation of screening for FASD at birth that identifies the components to be assessed and criteria for conducting a full diagnostic evaluation	86	**83.7**	2	1

Table 5.8 Round 2 statement ratings: components of screening (con't)

Statement	N	Agree (%)	Median	IQD
Screening for FASD at or around birth should also assess and record:				
S1Q8. family history of FASD, developmental delay, abuse or neglect	84	**97.6**	2	1
S1Q9. Most of the information required for FASD screening in childhood is routinely assessed as part of a general clinical assessment of children with neurodevelopmental or other related presentations	77	59.7	2	2
S1Q10. Screening for FASD in childhood primarily requires health professionals to assess prenatal alcohol exposure and consider it as a potential cause of other relevant abnormalities identified (e.g. abnormalities of development, learning, behaviour)	84	**89.3**	2	1
S1Q11. A checklist is needed to support the implementation of screening for FASD in childhood that identifies the components to be assessed and criteria for conducting a full diagnostic evaluation	84	**88.1**	2	1

TARGETED SCREENING

Consensus was reached for all indications for targeted screening (Tables 5.9 and 5.10). Participants suggested that behavioural problems or diagnoses (including Attention Deficit Hyperactivity Disorder (ADHD), autism, Asperger syndrome, social communication deficits, poor impulse control, and learning and behavioural problems) should also prompt targeted screening.

Table 5.9 Round 1 statement ratings: targeted screening indications

Statement: targeted screening for FASD should be conducted for all children who present with:	N	Agree (%)	Median	IQD
S3Q1. an alcohol-related event, illness or dependency in the birth mother	99	**96.0**	1	1
S3Q2. a parent or foster parent who is concerned that their child might have a FASD	99	**99.0**	1	1
S3Q3. prenatal alcohol exposure	98	**91.8**	1	1

Table 5.10 Round 1 statement ratings: targeted screening indications – presentations

Statement: in the absence of other known causes, targeted screening for FASD should be conducted for all children who present with:	N	Agree (%)	Median	IQD
S3Q4. developmental delay	96	**91.7**	1	1
S3Q5. growth retardation or failure to thrive	95	**91.6**	2	1
S3Q6. structural central nervous system abnormalities	90	**86.7**	2	1
S3Q7. neurological signs	92	**82.6**	2	1
S3Q8. functional central nervous system abnormalities	92	**88.0**	2	1
S3Q9. characteristic FAS facial anomalies	97	**96.9**	1	1
S3Q10. birth defects	93	**92.5**	2	1
S3Q11. reported or observed problems with behaviour	96	**86.5**	2	1

Consensus was reached for all indications for targeted screening in high risk groups (Tables 5.11 and 5.12). Participants suggested that other high risk groups should also undergo targeted screening, including individuals attending health and mental health services (child and adolescent health services, alcohol and drug service, colic and feeding services), Indigenous communities that request screening to be implemented, learning support services and recurrent emergency department admissions.

Table 5.11 Round 1 statement ratings: targeted screening indications – high risk groups

Statement: the following children should be screened for FASD:	N	Agree (%)	Median	IQD
S3Q13. children of mothers attending alcohol treatment services	99	**93.9**	1	1
S3Q14. siblings of identified cases of FASD	98	**95.9**	1	1
S3Q15. children who are diagnosed with ADHD	90	**74.4**	2	2

Table 5.12 Round 1 statement ratings: targeted screening indications – high risk groups

Statement: All children should be screened for FASD when they enter:	N	Agree (%)	Median	IQD
S3Q16. a child development service	96	**87.5**	2	1
S3Q17. child protection	92	**85.9**	2	1
S3Q18. foster care or adoptive placements (including kinship care)	93	**87.1**	2	1
S3Q19. a juvenile justice setting	91	**83.5**	2	1

General comments about targeted screening for FASD in high risk groups indicated that it is 'impossible to screen all children where there are child protection concerns', and the need to ensure that the criteria are not over-inclusive. Criteria for targeted screening were considered to require refinement so that screening is only relevant if there are other indications of FASD and if no other obvious cause for the child's problems is identified. Other suggested groups for screening included Department of Education early intervention programs, and an inter-agency approach required to promote sharing of information. No additional statements on targeted screening were included in the round 2 questionnaire.

SCREENING PROVIDERS

Of the 17 statements on screening providers, 15 achieved consensus (Table 5.13). Participants endorsed the involvement of a wide range of health professionals in the provision of screening, although there was a lower level of support for the involvement of physiotherapists and social workers. Participant comments about which professionals should be involved in screening reflected three main issues that are described in detail below.

Comments that supported the involvement of all health professionals in screening included that 'all of the above are in the business of screening for difficulties in infants and/or children', and that once trained well, any professional could perform screening in the course of their routine professional activities. Different professionals were recognised to have different roles and 'one cannot assume that health professionals know or have an understanding of FASD simply because of their professions.' Participants believed that training was required to improve general awareness about what features to look for and who to refer to, with 'what is more required is knowledge of the condition rather than repetitive screening of the long suffering client'.

Responses that did not support the involvement of a broad range of health professionals in screening indicated that it requires specialised skills, expertise and experience with other conditions that can cause overlapping phenotypes. For example, 'obstetricians and midwives should flag high risk neonates but do not have the expertise to do the screening themselves' and allied health professionals are the 'most likely not to have the in-depth training needed'. Other respondents identified frontline workers who were considered to be the gatekeepers of referral pathways, and particularly those for high risk groups (for example Aboriginal health workers, general practitioners, child health nurses, remote area nurses and midwives), were recommended for targeted education and training. Participants identified the need for health professionals to work collaboratively and to use a central referral system to avoid duplication, and stated that all professionals would not be required to screen if early screening was established in the obstetric setting and through community or school health nurses. Rather than involving a range of professionals in screening it was considered 'more important to get across the message to all health professionals that they have a role in considering the possible diagnosis, and referring to those with specialty knowledge'.

Other responses described uncertainty about what screening would involve and the need to define screening, and indicated that the appropriate person to perform screening 'depends on how and when screening would be done'. Concerns were also raised about the predictive value of screening, the need for different screening tools for different age and ethnic groups, the need for services to which individuals with FASD can be referred, and the need for screening to be affordable, accurate, acceptable and easy to administer. No additional questions on screening providers were included in round 2.

Table 5.13 Round 1 statement ratings: screening providers

Statement: the following health professionals should screen for FASD:	N	Agree (%)	Median	IQD
S4Q1. paediatricians	95	**97.9**	1	1
S4Q2. neonatologists	93	**97.8**	1	1
S4Q3. obstetricians	86	**74.4**	2	2
S4Q4. psychiatrists	89	**83.1**	2	1
S4Q5. general practitioners	92	**91.3**	2	1
S4Q6. nurse practitioners	92	**80.4**	2	1
S4Q7. midwives	89	**82.0**	2	1
S4Q8. child health nurses	93	**92.5**	1	1
S4Q9. school health nurses	91	**83.5**	2	1
S4Q10. community health nurses	91	**85.7**	2	1
S4Q11. occupational therapists	86	**72.1**	2	2
S4Q12. speech pathologists	86	**70.9**	2	2
S4Q13. social workers	88	62.5	2	2
S4Q14. psychologists	89	**78.7**	2	1
S4Q15. physiotherapists	84	58.3	2	2
S4Q16. Aboriginal health workers	90	**88.9**	1	1
S4Q17. All health professionals who screen for FASD require appropriate FASD-specific training	95	**94.7**	1	1

SCREENING METHODS: PRENATAL ALCOHOL EXPOSURE

Of the four statements that assessed the necessary content of PAE assessment during screening, all achieved consensus (Table 5.14). Participants also indicated that other factors should be assessed and recorded, including what alcohol was consumed and when, evidence of dependency, history of drinking in previous pregnancies, other drug use, psychosocial assessment and social situation including drug and alcohol use in the partner. Participants also identified that screening may not obtain accurate responses.

Table 5.14 Round 1 statement ratings: assessment of prenatal alcohol exposure

Statement: assessment of prenatal alcohol exposure should identify and record:	N	Agree (%)	Median	IQD
S5Q1. number of standard alcoholic drinks consumed during a typical drinking occasion	93	**97.8**	1	1
S5Q2. frequency of drinking occasions	94	**97.9**	1	1
S5Q3. frequency of excessive (binge) drinking (5 or more standard drinks per occasion)	94	**95.7**	1	1
S5Q4. timing of alcohol intake during pregnancy	94	**96.8**	1	1

Only two of the four statements that assessed how PAE should be assessed during screening achieved consensus (Table 5.15). There was limited support for the use of informal methods to assess PAE, and the assessment of PAE at every antenatal visit.

Table 5.15 Round 1 statement ratings: assessment of prenatal alcohol exposure

Statement		N	Agree (%)	Median	IQD
S5Q6.	Alcohol exposure should be assessed at every antenatal visit	90	65.6	2	2
S5Q7.	Alcohol exposure should be assessed alongside other lifestyle factors (e.g. diet)	93	**92.5**	1	1
S5Q8.	Prenatal alcohol exposure can be effectively assessed using an informal approach (e.g. inquiring during a consultation)	90	52.2	2	2
S5Q9.	Prenatal alcohol exposure should be assessed using a formal tool	77	**71.4**	2	2

Most participants were unfamiliar with specific screening tools to identify alcohol use in pregnancy and potentially harmful PAE (Table 5.16). Among participants who were familiar with the AUDIT-C screening tool (42), 54.8% recommended its use, compared with 48.6% for the Lifescripts tool (which incorporates the AUDIT-C), 45% for the T-ACE and 42.1% for the TWEAK.

Table 5.16 Round 1: use of formal tools to assess prenatal alcohol exposure

Statement: prenatal alcohol exposure should be assessed using one of the following tools:	N	Yes (%)	Unsure (%)	No (%)	Not Familiar with (%)
S5Q10. AUDIT-C (3-item Alcohol Use Disorders Identification Test)	94	24.5	16.0	4.3	55.3
S5Q11. Lifescripts tool (includes 3-item AUDIT-C)	94	19.1	18.1	2.1	60.6
S5Q12. TWEAK (5-item Tolerance, Worry, Eye-opener, Amnesia, Cut down)	93	17.2	12.9	10.8	59.1
S5Q13. T-ACE (4-item: Tolerance, Annoyed, Cut down, Eye-opener)	94	19.1	12.8	10.6	57.4

General comments about the assessment of PAE indicated the need to use a tool that is brief, sensitive, reliable, valid and culturally appropriate for remote Aboriginal communities and cross-cultural contexts. Specific comments included: that the Smoking, Nutrition, Alcohol, Physical Activity and Emotional Health (SNAPE) Tool is used by remote area Aboriginal health workers, nurses and midwives; that the formal tools are indicators of dependence and lack sensitivity for young people and hidden or binge use; and the potential need for more specific assessment for high risk groups. Some participants stated a preference for an informal assessment approach during clinical interview, with questioning considered to be less confronting when alcohol history is integrated with the general history and able to retrieve a more detailed frequency, quantity and timing of alcohol use than the AUDIT-C. Establishing a trusting relationship and using a sensitive approach to assess the many risk factors for the fetus was believed to be essential, with repeated questioning likely to be counterproductive and risk causing

shame and blame. The importance of a team approach was emphasised as was the need to seek reliable collateral information on PAE in some circumstances, including information on hospital admissions and treatment for alcohol-related events.

In round 2, a brief description of the AUDIT-C was included with feedback on the round 1 results, and further information was sought on the role of informal assessment and endorsement of the AUDIT-C as a possible assessment tool (Table 5.17). Round 2 findings indicated consensus agreement on the combination of formal assessment tools for PAE with use of the AUDIT-C to assess PAE and a clinical interview to obtain more detailed information on consumption.

Table 5.17 Round 2 statement ratings: assessment of prenatal alcohol exposure

Statement	N	Agree (%)	Median	IQD
S2Q1. The use of formal tools for the assessment of prenatal alcohol exposure should be combined with a clinical interview to obtain more detailed information about alcohol consumption patterns, potential indicators of addiction and other relevant contextual information	87	**88.5**	2	1
S2Q2. Prenatal alcohol exposure can be effectively assessed using an informal approach (e.g. inquiring during a consultation)	85	44.7	3	2
S2Q3. Information on alcohol use from family members, other health professionals or community members (if appropriate) should be sought if indicated	85	**78.8**	2	0
S2Q4. The AUDIT-C would be a useful tool for the formal assessment of prenatal alcohol exposure	80	**87.5**	2	0

SCREENING METHODS: GROWTH DEFICIT

Participants most strongly agreed with the assessment of growth by comparing height and weight with population standards (Table 5.18). Participant comments included: that growth is age-specific and must be monitored over time; that nutritional and other factors must be considered in interpretation; that the absence of growth deficit should not exclude FASD; and that the reference standards used should include the World Health Organization (WHO) growth charts until two years of age, and the WHO or Centers for Disease Control (CDC) growth charts for children aged two years or more. General comments also included: uncertainty about the worth of growth as part of a screening tool; that it is time consuming and expensive; that growth criteria apply to a host of other problems in children who do not have FAS; that assessment of growth is a usual component of child health assessments; and uncertainty about how screening assessments differ from diagnostic assessments. Consensus was reached for two of the four statements on the definition of growth deficit (Table 5.19), with greatest support for the evaluation of growth deficit based on comparison of height and weight with population

standards at the 10th percentile. No further questions on screening for growth deficit were included in round 2.

Table 5.18 Round 1 statement ratings: assessment of growth deficit

Statement	N	Agree (%)	Median	IQD
S6Q1. Growth should be assessed by comparing height and weight with population standards	75	**92.0**	2	1
S6Q2. Growth should be assessed by comparing weight to height ratio with population standards	71	64.8	2	2
S6Q3. Growth should be assessed by comparing weights over time (to identify decelerating weight over time)	73	**86.3**	2	1
S6Q4. Assessment of growth deficit should consider other factors that may affect growth (e.g. gestational age parental size, gestational diabetes, nutritional status, illness)	85	**98.8**	1	1

Table 5.19 Round 1 statement ratings: definition of growth deficit

Statement: growth deficit should be defined as:	N	Agree (%)	Median	IQD
S8Q8. prenatal or postnatal growth deficit in height or weight at or below the 10th percentile based on comparison with population standards for age/gestational age (and sex and race where available)	70	**85.7**	2	1
S8Q9. prenatal or postnatal height or weight low for age/gestational age	63	65.1	2	1
S8Q10. disproportionately low weight-to-height ratio at or below the 10th percentile	58	**70.7**	2	1
S8Q11. disproportionately low weight-to-height ratio	55	56.4	2	2

SCREENING METHODS: FACIAL ANOMALIES

Both general statements about the assessment of characteristic facial anomalies reached consensus agreement (Table 5.20). Participants also indicated that the presence of other facial anomalies that may 'travel with FASD' should be recorded (e.g. low ears, railroad track ears, flattened midface, epicanthal folds, cleft lip, hypertelorism, depressed nasal bridge, micrognathia), due to the need to consider other neurodevelopmental anomalies as alternative diagnoses or comorbidities.

Table 5.20 Round 1 statement ratings: assessment of characteristic facial anomalies

Statement	N	Agree (%)	Median	IQD
S6Q6. The presence of the following characteristic FAS facial anomalies should be assessed: smooth philtrum, thin upper lip, and small palpebral fissures	89	**98.9**	1	1
S6Q7. Assessment of characteristic FAS facial anomalies should use appropriate anthropometric population standards for race and age where available	85	**94.1**	1	1

Among methods for the assessment of characteristic facial anomalies, The University of Washington (UW) Lip-Philtrum Guide received the highest level of support (Tables 5.21 and 5.22). General comments about the assessment of facial anomalies included: that screening needs to be efficient, with overly detailed methods and measurement adding to screening time and making it less likely to be done; that clinical observation is operator dependent and measurements should be used to confirm the clinical impression; disagreement with a focus on facial features during screening because the face is only abnormal in FAS and the absence of characteristic facial features may encourage professionals to dismiss the possibility of FASD; the need for training to use facial dysmorphology assessment tools; that measures are often inaccurate; the need to consider normal variation, family characteristics and ethnicity; the need for a guide for Indigenous Australians; the need for better technologies such as three-dimensional assessment; and that this identification of facial features is diagnostic and not part of screening.

Table 5.21 Round 1 statement ratings: assessment of characteristic facial anomalies

Statement: at the screening stage, characteristic FAS facial anomalies can be effectively assessed using:	N	Agree (%)	Median	IQD
S6Q9. clinical observation	77	68.8	2	1
S6Q10. physical measurement of palpebral fissures	58	**75.9**	2	1
S6Q11. the University of Washington Lip-Philtrum Guide	57	**86.0**	2	1
S6Q12. the facial photographic screening tool	52	**73.1**	2	2

Table 5.22 Round 1 statement ratings: definition of thin upper lip and smooth philtrum

Statement: thin upper lip and smooth philtrum should be defined as:	N	Agree (%)	Median	IQD
S8Q5. positive finding based on the University of Washington Lip-Philtrum Guide (rank 4 or 5)	52	**96.2**	1	1
S8Q6. positive finding based on visual assessment/clinical impression	58	58.6	2	2

Consensus was not reached for all three statements on the definition of short palpebral fissures (Table 5.23). Comments about the definition of short palpebral fissures indicated the need for appropriate norms; that experience is required to achieve reliable measurement; that clinical impression from an experienced clinician is acceptable; that clinical photographs would assist clinical impression; and that the assessment is diagnostic and not part of screening.

Table 5.23 Round 1 statement ratings: definition of short palpebral fissures

Statement: short palpebral fissures should be defined as:	N	Agree (%)	Median	IQD
S8Q1. fissure length at or below the 10th percentile based on comparison with population standards, with physical or photographic measurement of length	47	53.2	2	2
S8Q2. fissure length below the 3rd percentile based on comparison with population standards, with physical or photographic measurement of length	50	68.0	2	2
S8Q3. positive finding based on visual assessment/clinical impression	56	55.4	2	2

Further questions were included in round 2 to clarify participant perceptions of the distinction between screening and diagnostic assessment of facial anomalies, the definition of short palpebral fissures, and the use of clinical observation at the screening stage. Round 2 results indicated that measurement of facial anomalies was not considered essential at the screening stage (Table 5.24).

Table 5.24 Round 2 statement ratings: assessment of characteristic facial anomalies

Statement	N	Agree (%)	Median	IQD
S3Q6. Facial anomalies can be assessed using clinical observation for evidence of the characteristic FAS facial anomalies, with formal physical measurement of these features not essential at the screening stage	71	**77.5**	2	0
S3Q2. At the screening stage it is not necessary to formally assess and measure suspected facial anomalies	74	63.5	2	2
S3Q7. Palpebral fissure length must be assessed using formal physical measurement and comparison with population references at the screening stage	67	37.3	4	2
S3Q8. Thin upper lip and smooth philtrum must be assessed using formal tools such as the University of Washington Lip-Philtrum Guide at the screening stage	67	46.3	3	2

SCREENING METHODS: BIRTH DEFECTS

Consensus was reached for the single statement about the assessment of birth defects, that FASD screening should assess and record the presence of birth defects as part of the clinical examination (Table 5.25). Comments by participants about the assessment of birth defects included: that any paediatric examination should assess birth defects; that some birth defects are age dependent; and that birth defects are not sufficiently sensitive or specific to FASD to warrant documenting them during screening.

Table 5.25 Round 1 statement ratings: assessment of birth defects

Statement	N	Agree (%)	Median	IQD
S6Q14. FASD screening should assess and record the presence of birth defects as part of the clinical examination	89	**97.8**	1	1

SCREENING METHODS: CENTRAL NERVOUS SYSTEM ABNORMALITIES

Consensus was reached for 13 of the 14 statements about the assessment of CNS abnormalities during screening (Table 5.26). Participant comments included: the need to screen for mental health issues or behavioural problems and symptoms such as anxiety, oppositional behaviour, obsessive symptoms, inattention, distractibility and short term memory problems; that diffusion tensor imaging would provide a better picture of brain functioning but referrals must have a fair chance of being diagnosed with FASD; and that standardised screening tests need to be used to assess CNS abnormalities.

Table 5.26 Round 1 statement ratings: assessment of CNS abnormalities during screening

Statement: Assessment of CNS abnormalities in FASD screening may include:	N	Agree (%)	Median	IQD
S7Q1. developmental milestones	88	**96.6**	1	1
S7Q2. motor and sensory function	85	**88.2**	2	1
S7Q3. cognition (IQ)	87	**92.0**	1	1
S7Q4. memory	86	**89.5**	2	1
S7Q5. academic achievement	87	**90.8**	2	1
S7Q6. executive functioning and abstract reasoning	87	**89.7**	2	1
S7Q7. adaptive behaviour	83	**92.8**	1	1
S7Q8. attention and hyperactivity	88	**95.5**	1	1
S7Q9. communication (receptive and expressive language)	86	**94.2**	2	1
S7Q10. social skills and social communication	88	**93.2**	1.5	1
S7Q11. hard and soft neurologic signs (including sensory-motor signs)	82	**86.6**	2	1
S7Q12. seizures that are not due to a postnatal insult or other postnatal process	82	**79.3**	2	1
S7Q13. head circumference	86	**95.3**	1	1
S7Q14. brain imaging	72	56.9	2	1

Participants endorsed the use of valid and reliable neurobehavioural assessment methods that are appropriate for the age and cultural background of each patient (Table 5.27). General comments about the assessment of CNS abnormalities in FASD screening included: the need for distinction between screening and diagnostic assessments; the need for a pragmatic approach considering cost and resources, including the possibility of qualitative assessment of core areas at the screening stage; that testing must be age and culturally appropriate; that testing requires experienced clinicians; that an

evidence-based standardised approach is required; that multidisciplinary input is required; and that causes may be multifactorial.

Table 5.27 Round 1 statement ratings: assessment of CNS abnormalities during screening

Statement: the choice of tests for neuro-behavioural assessments should be guided by:	N	Agree (%)	Median	IQD
S7Q16. the availability of valid and reliable instruments	84	**88.1**	2	1
S7Q17. clinician preference and experience	82	59.8	2	2
S7Q18. test appropriateness for patient age and cultural background	86	**97.7**	1	1

Agreed definitions of CNS abnormalities that would be considered significant if found on screening included head circumference below the 3rd percentile, brain abnormalities observable on imaging, clinical findings of hard or soft neurological signs, and evidence of functional impairment at two or more standard deviations below the mean based on standard psychometric testing (Table 5.28). General comments about the definition of structural and neurological CNS abnormalities included: the need for distinction between screening and diagnostic assessments; that imaging is diagnostic; that these findings are not specific to FASD; that seizure disorders often have no cause found and should not be labelled as FASD; that functional problems are not necessarily accompanied by structural changes; and that soft signs should not be used as diagnostic criteria as they are difficult to assess and may be a variation of normal.

General comments about the definition of functional CNS abnormalities included: that standardised testing must be used as far as possible; that these assessments are diagnostic and not screening tests; that there is a lack of Australian normative data for standard psychometric assessments suitable for use with multi-cultural and Indigenous populations; that CNS abnormalities are not specific or diagnostic; that cognitive impairment may not be less than two standard deviations below the mean; that the use of a threshold as severe as two standard deviations below the mean is not necessary for screening; and that interpretations should be made about constructs using test scores as a tool, with a purely psychometric definition of impairment considered out-dated and problematic.

Table 5.28 Round 1 statement ratings: definition of CNS abnormalities during screening

Statement: CNS abnormalities should be defined as:	N	Agree (%)	Median	IQD
S8Q13. head circumference at or below the 10th percentile	60	63.3	2	1
S8Q14. head circumference below the 3rd percentile	62	**88.7**	1.5	1
S8Q15. clinically significant brain abnormalities observable through imaging techniques (e.g. hydrocephaly, size or shape of the corpus callosum, cerebellum, or basal ganglia) determined by an appropriately trained professional	62	**87.1**	2	1
S8Q17. presence of hard or soft neurological signs (including sensory-motor signs) based on clinical assessment	68	**89.7**	2	1
S8Q18. presence of a seizure disorder that is not due to a postnatal insult	63	69.8	2	2
S8Q20. evidence of functional impairment on standard psychometric testing, with performance 2 or more standard deviations below the mean, assessed by qualified professionals	66	**89.4**	2	1
S8Q21. evidence of functional impairment on standard psychometric testing, with performance 1 or more standard deviations below the mean, assessed by qualified professionals	65	47.7	3	2
S8Q22. clinical judgement of functional impairment or deficit in domains where standardised measurements are not available	72	63.9	2	1
S8Q23. clinical judgement of functional impairment or deficit based on clinical observation and assessment	69	62.3	2	2

Participants strongly agreed with the use of standardised assessment tools and comparisons with population data in assessments of abnormalities during screening (Table 5.29). General comments about the definition of abnormal screening findings included: that screening should be simple and quick; that multidisciplinary evaluation should be used; that appropriate reference data are not available for non-Caucasians; that screening tools should not be limited to those that have Australian population reference data; and that false negatives and false positives are common and concerning.

Table 5.29 Round 1 statement ratings: definition of abnormal screening findings

Statement	N	Agree (%)	Median	IQD
S8Q25. Evidence of abnormality or dysfunction should be based on normative data	75	**92.0**	2	1
S8Q26. The population standards required for comparison during FASD screening are available in Australia	53	45.3	3	2
S8Q27. Evidence of abnormality or dysfunction should be based on valid and reliable standard assessment tools where available	83	**95.2**	2	1
S8Q28. A full diagnostic evaluation for FASD should occur outside standard criteria when health professionals have concerns or doubts about FASD screening results	75	**82.7**	2	1

Due to conflicting participant comments about screening methods for CNS abnormalities, additional questions were included in round 2 to identify agreement with the need for formal testing of CNS domains at the screening stage. Round 2 results indicated that formal assessment of CNS abnormalities was not required at the screening stage (Table 5.30).

Table 5.30 Round 2 statement ratings: assessment of CNS abnormalities during screening

Statement:	N	Agree (%)	Median	IQD
S3Q1. At the screening stage it is not necessary to formally assess and measure suspected CNS anomalies	74	**71.6**	2	1
At the screening stage the following are acceptable indicators of possible CNS abnormalities (neurological, functional or structural):				
S3Q3. clinical identification	72	**95.8**	2	0
S3Q4. parent or other credible third party report	72	**88.9**	2	0
S3Q5. results of previous relevant formal assessments (e.g. psychological report)	71	**100**	2	1

CRITERIA FOR REFERRAL

CRITERIA FOR PRENATAL ALCOHOL EXPOSURE

There was consensus agreement for referral for a diagnostic evaluation based on evidence of moderate or heavy levels of PAE alone (Table 5.31). Participant comments about the criteria for referral for diagnosis included: that PAE should always lead to screening and monitoring as any PAE may be significant and information about exposure may not be accurate; that suspected heavy exposure (e.g. a history of alcohol-related illness or dependency) should prompt diagnostic evaluation and, if appropriate, assessment and support for the mother; that heavy PAE is not alone sufficient to warrant diagnostic evaluation as there are not sufficient resources to deal with this, and identification and management of the child's difficulties is the key; that it is impossible to get this level of information in all settings; that the absence of information on PAE should not preclude the need to consider FASD; that referral for high levels of exposure would send an inappropriate public health message that lower levels of exposure are safe; and that it is difficult to define the relevant level of exposure.

Table 5.31 Round 1 statement ratings: referral criteria for PAE alone

Statement: what level of alcohol exposure, at any time during pregnancy, would alone be sufficient to indicate the need for a full diagnostic evaluation for FASD:	N	Agree (%)	Median	IQD
S9Q1. less than 7 standard drinks per week, and no more than 2 standard drinks on any one day	80	37.5	3	2
S9Q2. less than 7 standard drinks per week, and between 3 and 4 standard drinks on any one day	78	61.5	2	1
S9Q3. 7 or more standard drinks per week, and no more than 2 standard drinks on any one day	79	59.5	2	2
S9Q4. 7 or more standard drinks per week, and between 3 and 4 standard drinks on any one day	81	**81.5**	2	1
S9Q5. binge drinking (5 or more standard drinks per occasion) less than once per week	84	**78.6**	2	1
S9Q6. binge drinking (5 or more standard drinks per occasion) once or twice per week	83	**84.3**	1	1
S9Q7. no level of prenatal alcohol exposure is alone sufficient to indicate the need for a full diagnostic evaluation for FASD	72	45.8	3	3

CRITERIA FOR OTHER COMBINATIONS OF SCREENING FINDINGS

Consensus agreement was reached for 13 of the 16 criteria for other combinations of screening findings that require a diagnostic evaluation (Table 5.32). General comments about the criteria for referral included: support for sensitive criteria for referral to reduce the risk of undetected cases; that diagnostic evaluations for FASD do not stand alone and are part of an integrated assessment process for neurological or developmental concerns; that the criteria for parental concern is too non-specific and should instead indicate the need for screening; that the distinction between screening and diagnosis is unclear; and that prior assessment is required to rule out other known causes of these criteria, with FASD possibly a differential diagnosis.

Table 5.32 Round 1 statement ratings: referral criteria for other combinations of screening findings

Statement: in the absence of other known causes, a full diagnostic evaluation for FASD is required when there is evidence of:	N	Agree (%)	Median	IQD
S9Q8. concern by a parent or foster parent that their child might have a FASD	88	**88.6**	2	1
S9Q9. all 3 of the characteristic FAS facial anomalies (smooth philtrum, thin vermillion border, and small palpebral fissures)	83	**95.2**	1	1
S9Q10. 2 of the characteristic FAS facial anomalies	78	**76.9**	2	1
S9Q11. the characteristic pattern of FAS facial anomalies (number unspecified)	79	**72.2**	2	2
S9Q12. 2 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality (structural, neurological or functional)	80	**93.8**	2	1
S9Q13. 2 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	82	**92.7**	1	1

Table 5.32 Round 1 statement ratings: referral criteria for other combinations of screening findings (con't)

Statement: in the absence of other known causes, a full diagnostic evaluation for FASD is required when there is evidence of:	N	Agree (%)	Median	IQD
S9Q14. 1 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality	81	67.9	2	1
S9Q15. 1 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	81	**85.2**	2	1
S9Q16. known or probable prenatal alcohol exposure, **and** 1 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality	83	**92.8**	2	1
S9Q17. known or probable prenatal alcohol exposure, **and** 1 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	82	**96.3**	1	1
S9Q18. growth deficit **and** any CNS abnormality	79	55.7	2	1
S9Q19. known or probable prenatal alcohol exposure, **and** growth deficit **and** any CNS abnormality	83	**94.0**	2	1
S9Q20. known or probable prenatal alcohol exposure, **and** any CNS abnormality	82	**87.8**	2	1
S9Q21. 2 or more CNS abnormalities	73	43.8	3	1
S9Q22. known or probable prenatal alcohol exposure, **and** 2 or more CNS abnormalities	82	**95.1**	2	5
S9Q23. known or probable prenatal alcohol exposure, **and** 1 or more birth defects	81	**87.7**	2	1

Round 2 results confirmed agreement with the criteria for referral endorsed in round 1 (Figure 2) and with referral based on moderate or heavy PAE alone (Table 5.33).

Figure 5.2 Round 1 consensus criteria for referral apart from PAE alone

i. concern by a parent or foster parent that their child might have a FASD
ii. 2 or more of the characteristic FAS facial anomalies or the characteristic pattern of FAS facial anomalies
iii. 1 of the characteristic FAS facial anomalies, and a growth deficit and any CNS abnormality
iv. known or probable prenatal alcohol exposure, and 1 of the characteristic FAS facial anomalies, and a growth deficit or any CNS abnormality
v. known or probable prenatal alcohol exposure, and any CNS abnormality
vi. known or probable prenatal alcohol exposure, and 1 or more birth defects

Table 5.33 Round 2 statement results: referral criteria

Statement	N	Agree (%)	Median	IQD
S4Q1. If any of the combinations of criteria listed in [Figure 2] above are met, a full diagnostic evaluation is required	79	**91.1**	2	1
S4Q2. Evidence of significant prenatal alcohol exposure on its own is sufficient to require a full diagnostic evaluation	81	**71.6**	2	2
S4Q3. The above criterion of significant prenatal alcohol exposure (Q2) can be generally understood to indicate exposure to 7+ standard drinks per week and 3+ drinks on any one occasion, or regular exposure to 5+ standard drinks on any one occasion, or strong clinical suspicion of heavy prenatal alcohol exposure	80	**86.3**	2	0

DIAGNOSIS

EXISTING DIAGNOSTIC GUIDELINES

Approximately half of the participants were familiar with the UW Guidelines, and just over a third were familiar with the Canadian Guidelines (Table 5.34). Almost one third of participants who were familiar with the UW and Canadian Guidelines respectively recommended that these be adopted in Australia (Table 5.35).

Table 5.34 Round 1: familiarity with existing diagnostic guidelines

Statement: are you familiar with the following diagnostic guidelines for FASD:	N	Yes % (n)
S10Q1. Institute of Medicine Guidelines (1996)	88	30.7 (27)
S10Q2. University of Washington 4-Digit Diagnostic Code (2004)	89	50.6 (45)
S10Q3. Centers for Disease Control and Prevention (CDC) Guidelines (2004)	90	31.1 (28)
S10Q4. Canadian Guidelines (2005)	91	35.2 (32)
S10Q5. Hoyme (an update based on Institute of Medicine) Guidelines (2005)	88	13.6 (12)

Table 5.35 Round 1: adoption of existing diagnostic guidelines in Australia

Statement: should this diagnostic guideline be adopted in Australia:	N	Yes % (n)
S10Q1.2 Institute of Medicine Guidelines (1996)	25	4.0 (1)
S10Q2.2 University of Washington 4-Digit Diagnostic Code (2004)	40	32.5 (13)
S10Q3.2 Centers for Disease Control and Prevention (CDC) Guidelines (2004)	23	8.7 (2)
S10Q4.2 Canadian Guidelines (2005)	26	30.8 (8)
S10Q5.2 Hoyme (an update based on Institute of Medicine) Guidelines (2005)	11	9.1 (1)

Participant comments about the adoption of existing FASD diagnostic systems or guidelines in Australia included: that education and training of health professionals who are going to use the diagnostic tool is the key; that the development of an Australian guideline appropriate for use in rural and remote areas based on the adoption and adaptation of existing guidelines is required; that Australian normative data are required, particularly Indigenous Australian facial norms; that there is a need to ensure good access to diagnostic, intervention and support services within an interdisciplinary and inter-agency model; that an Australian tool would need to be valid, specific and sensitive; that a standard national approach is required to support accurate data collection; and that an Australian system would need to be easy to understand, for example the UW Guidelines diagnostic categories would need to be simplified, and practical to use, with possibly a simpler system required by generalists such as general practitioners.

DIAGNOSTIC PROCESSES

There was consensus agreement that a paediatrician or clinical geneticist is required to confirm the diagnosis of FASD and exclude alternative diagnoses, and that diagnosis should involve multidisciplinary assessment (Table 5.36). Participants also indicated that psychiatrists (child and neurodevelopmental), addiction medicine specialists (for older patients with delayed diagnosis) and interdisciplinary teams should also be able to confirm the diagnosis of FASD and exclude alternative diagnoses. General comments about who should confirm the diagnosis of FASD indicated that a multidisciplinary team with a specialist paediatrician was the gold standard for diagnosis, and that resource constraints limit the provision of diagnostic services, particularly outside metropolitan areas where multidisciplinary teams are not available or practical. Participants suggested that, in rural areas, general practitioners may have to diagnose FASD, and that appropriate interest, education, training and experience can increase the capacity of non-specialists, such as general practitioners, to confirm and exclude FASD in the absence of an ideal level of resources.

Table 5.36 Round 1 statement ratings: diagnostic processes

Statement	N	Agree (%)	Median	IQD
S11Q1. Exclusion of differential diagnoses is essential for the accurate diagnosis of Fetal Alcohol Spectrum Disorders	85	**91.8**	2	1
S11Q2. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to confirm the diagnosis of a FASD	77	**74.0**	2	2
S11Q3. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to exclude alternative diagnoses	81	**82.7**	2	1
S11Q4. With appropriate FASD-specific training, general practitioners can confirm the diagnosis of a FASD	79	46.8	3	2
S11Q5. With appropriate FASD-specific training, general practitioners can exclude alternative diagnoses	77	29.9	4	2
S11Q6. Diagnosis of FASD should involve multidisciplinary assessment by FASD accredited paediatricians and other health professionals (e.g. social worker, psychologist, speech pathologist, occupational therapist, physiotherapist, nurse practitioner)	86	**83.7**	1.5	1

Participants strongly agreed with the need for multidisciplinary assessment clinics in major cities and scheduled visits by teams to support workforce development in regional centres (Table 5.37). General comments about the provision of FASD screening and diagnosis capacity outside metropolitan areas indicated: that it is vital to build local capacity in diagnosis, and support existing services through providing on-going training, screening and diagnostic tools, and assistance in complex cases; and that although the involvement of multidisciplinary teams was recognised as best practice, screening was not considered to require the involvement of specialist assessment teams. Participants highlighted the

need for diagnostic capacity to be linked to intervention services to enable local access, and an outreach service model was perceived to provide little on-going patient support.

Table 5.37 Round 1 statement ratings: screening and diagnostic service delivery

Statement	N	Agree (%)	Median	IQD
S11Q8. A multidisciplinary FASD assessment clinic should be available in major cities	89	**89.9**	1	1
S11Q9. Scheduled visits by FASD assessment teams to regional centres should be used to perform FASD screening and diagnosis	86	**76.7**	2	1
S11Q10. Scheduled visits by FASD assessment teams to regional centres should be used to support workforce training and development for FASD screening and diagnosis	87	**92.0**	1	1
S11Q11. Telehealth should be used by FASD assessment teams to support FASD screening and diagnosis	84	**79.8**	2	1

Because participant comments indicated the need to build local capacity for screening and diagnosis outside metropolitan areas, four questions were included in round 2 to identify support for the involvement of general practitioners in diagnosis in rural and remote areas. Round 2 results did not indicate consensus support for the involvement of general practitioners from rural or remote settings in the confirmation of a diagnosis of FASD or the exclusion of alternative diagnoses (Table 5.38).

Table 5.38 Round 2 statement results: diagnostic service delivery

Statement	N	Agree (%)	Median	IQD
S7Q1. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to confirm the diagnosis of a FASD	74	**79.7**	2	0
S7Q2. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to exclude alternative diagnoses	76	**89.5**	2	1
S7Q3. With appropriate FASD-specific training, general practitioners in rural and remote settings can confirm the diagnosis of a FASD	76	60.5	2	2
S7Q4. With appropriate FASD-specific training, general practitioners in rural and remote settings can exclude alternative diagnoses	75	44.0	3	2

DIAGNOSTIC CRITERIA FOR FETAL ALCOHOL SYNDROME

The proportion of participants who indicated that evaluating the diagnostic criteria for FAS was outside their area of expertise ranged from 22.3% to 43.7% for the Likert statements, and up to 55.3% for other closed survey questions.

Consensus was not reached on the general definition of the diagnostic criteria for FAS until round 2 (Tables 5.39 and 5.40). Participant comments on the definition of FAS included: that all four features

should not be a prerequisite for diagnosis, including when cognition and behaviours strongly predict FAS, or when features may not be evident at birth or may normalise in adolescence; that facial features should be supportive but not essential criteria; difficulties confirming the absence of PAE; that FAS is syndromic and clinically obvious without the need to measure cognition or growth; and that strict case definitions are needed for facial anomalies, growth deficit, CNS abnormality and PAE.

Table 5.39 Round 1 statement ratings: general diagnostic criteria for FAS

Statement	N	Agree (%)	Median	IQD
S12Q1. A diagnosis of FAS should only be made in the presence of all 4 of the following: characteristic FAS facial anomalies, growth deficit, CNS abnormalities and confirmed or unknown prenatal alcohol exposure	63	61.9	2	3
S12Q2. A confirmed absence of prenatal alcohol exposure (in the presence of all other required FAS findings) should rule out a diagnosis of FAS and be recorded under a different diagnostic category	69	**71.0**	2	1

Table 5.40 Round 2 statement results: general diagnostic criteria for FAS

Statement	N	Agree (%)	Median	IQD
S5Q1. A diagnosis of FAS should only be made in the presence of all 4 of the following: confirmed or unknown prenatal alcohol exposure, all three characteristic FAS facial anomalies, growth deficit and CNS abnormality	67	**82.1**	2	1

Participants agreed that all three characteristic FAS facial anomalies are required for a diagnosis of FAS (Table 5.41). Participant comments also included that characteristic FAS facial anomalies could include a flattened midface, and that the anomalies may be lost in other strong features, grown out, or not yet developed.

Table 5.41 Round 1 statement ratings: diagnostic criteria for characteristic FAS facial anomalies

Statement: FAS diagnostic criteria for facial anomalies should include:	N	Agree (%)	Median	IQD
S12Q4 all 3 of the following characteristic FAS facial anomalies; (short palpebral fissures, thin upper lip, smooth philtrum)	54	**74.1**	2	2
S12Q5. 2 or more of the following characteristic FAS facial anomalies; (short palpebral fissures, thin upper lip, smooth philtrum)	52	55.8	2	2
S12Q6. 2 or more of the following characteristic FAS facial anomalies; (short palpebral fissures, thin upper lip, smooth philtrum, flat midface)	53	47.2	3	2
S12Q7. evidence of a characteristic pattern of FAS facial anomalies that includes features such as short palpebral fissures and abnormalities in the premaxillary zone (e.g. flat upper lip, flattened philtrum)	54	55.6	2	2

Participants agreed that prenatal growth deficit (birth weight or length below the 10th percentile when corrected for gestation) or postnatal growth deficit in height or weight at or below the 10th percentile is required for a diagnosis of FAS (Table 5.42). Participant comments included: that the 3rd percentile could be used to define growth deficit in some population groups such as Asian and Indigenous Australians to account for trends towards lower weight and height; and that growth is not always restricted in FASD.

Table 5.42 Round 1 statement ratings: diagnostic criteria for growth deficit

Statement: FAS diagnostic criteria for growth deficit should include:	N	Agree (%)	Median	IQD
S12Q9. prenatal or postnatal growth deficit in height or weight at or below the 10th percentile	50	**80.0**	2	1
S12Q10. disproportionately low weight to height ratio at or below the 10th percentile	45	55.6	2	1
S12Q11. disproportional low weight to height	46	50.0	2.5	2
S12Q12. low birth weight for gestational age	50	56.0	2	1
S12Q13. decelerating weight over time not due to nutrition	49	53.1	2	1

Consensus agreement was reached only for the UW Guidelines general definition of FAS diagnostic criteria for CNS abnormalities (Table 5.43). Participants suggested the need to clarify that the abnormalities are of presumed prenatal origin, and that soft neurological signs should not be a diagnostic feature as they are difficult to assess using standardised methods and do not provide definitive evidence of neurological dysfunction.

Table 5.43 Round 1 statement ratings: general diagnostic criteria for CNS abnormalities

Statement: FAS diagnostic criteria for CNS abnormalities should include:	N	Agree (%)	Median	IQD
S12Q15. at least 1 structural CNS abnormality (including decreased cranial size),	45	55.6	2	1
S12Q16. at least 1 of the following CNS abnormalities: structural (abnormal brain structure, including decreased cranial size), **or** neurological (hard or soft neurological signs)	49	44.9	3	1
S12Q17. at least 1 of the following CNS abnormalities: structural (abnormal brain structure, including decreased cranial size), **or** neurological (hard or soft neurological signs), **or** functional (global cognitive or intellectual deficits representing multiple domains of deficit (including significant developmental delay in young children), **or** deficits in three or more specific functional domains (e.g. developmental milestones, cognition, memory, executive functioning, attention, hyperactivity, social, communication and language, motor and sensory)	49	**75.5**	2	1
S12Q18. 3 or more of the following CNS abnormalities: structural (abnormal brain structure, including decreased cranial size), neurological (hard or soft neurological signs), cognition, communication, academic achievement, memory, executive functioning and abstract reasoning, attention deficit or hyperactivity, adaptive behaviour, social skills, social communication	50	60.0	2	2

There was consensus agreement that the FAS diagnostic criteria for CNS abnormalities should include decreased cranial size at or below the 3rd percentile and functional performance below the 3rd percentile or two standard deviations below the mean (Table 5.44). General comments about the diagnostic criteria for CNS abnormalities included: the need for multidisciplinary assessment; the importance of clinical judgement in evaluation given the child's genetic background and experience; the need for microarray analysis of chromosomes to exclude other causes; and that the diagnostic criteria may not be highly specific.

Table 5.44 Round 1 statement ratings: specific diagnostic criteria for CNS abnormalities

Statement: CNS abnormalities should be defined as:	N	Agree (%)	Median	IQD
S12Q20. decreased cranial size at or below the 3rd percentile	50	**88.0**	2	1
S12Q21. decreased cranial size at or below the 10th percentile	47	31.9	3	2
S12Q22. global functional performance (cognitive or intellectual) below the 3rd percentile	52	**78.8**	2	1
S12Q23. performance for specific functional domains below the 3rd percentile	50	**82.0**	2	0
S12Q24. performance for specific functional domains below the 16th percentile	46	28.3	3	2
S12Q25.clinical judgement of functional impairment or deficit in domains where standardised measurements are not available	53	58.5	2	1
S12Q26. clinical judgement of functional impairment or deficit based on clinical assessment	56	44.6	3	2

DIAGNOSTIC CRITERIA FOR OTHER FETAL ALCOHOL SPECTRUM DISORDERS

The proportion of participants who indicated that evaluating the diagnostic criteria for other FASD was outside their area of expertise ranged from 39.8% to 46.6% for the Likert statements, and up to 57.3% for other closed survey questions.

In round 1 consensus agreement was reached for both general definitions of the diagnostic criteria for PFAS which were based on the Canadian and IOM Guidelines (Table 5.45). Both questions were repeated in round 2 with similar results (Table 5.46).

Table 5.45 Round 1 statement ratings: general diagnostic criteria for PFAS

Statement: A diagnosis of PFAS in the absence of FAS requires:	N	Agree (%)	Median	IQD
S13Q1. confirmed prenatal alcohol exposure, **and** evidence of some components of the pattern of characteristic FAS facial anomalies, **and either**: growth deficit, **or** structural or neurological CNS abnormality, **or** evidence of multiple behavioural or cognitive abnormalities that are inconsistent with developmental level (e.g. learning, academic achievement, poor impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement)	50	**70.0**	2	1
S13Q2. confirmed prenatal alcohol exposure, **and** 2 of the 3 characteristic FAS facial anomalies (short palpebral fissure, thin upper lip, smooth philtrum), **and** CNS abnormality in 3 of the following areas (hard and soft neurologic signs, brain structure, cognition, communication, academic achievement, memory, executive functioning and abstract reasoning, attention deficit or hyperactivity, adaptive behaviour, social skills, social communication)	47	**74.5**	2	2

Table 5.46 Round 2 statement ratings: general diagnostic criteria for PFAS

Statement: A diagnosis of PFAS in the absence of FAS requires:	N	Agree (%)	Median	IQD
S5Q2. confirmed prenatal alcohol exposure, **and** evidence of some components of the pattern of characteristic FAS facial anomalies, **and either:** growth deficit, **or** structural or neurological CNS abnormality, **or** evidence of multiple behavioural or cognitive abnormalities that are inconsistent with developmental level (e.g. learning, academic achievement, poor impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement)	64	**70.3**	2	1
S5Q3. confirmed prenatal alcohol exposure, **and** 2 of the 3 FAS facial anomalies (short palpebral fissures, thin upper lip, smooth philtrum), **and** CNS abnormality in 3 of the following areas (hard and soft neurologic signs, brain structure, cognition, communication, academic achievement, memory, executive functioning and abstract reasoning, attention deficit or hyperactivity, adaptive behaviour, social skills, social communication)	62	**75.8**	2	0

Consensus agreement was reached on the diagnostic criteria for ARND as defined in the IOM Guidelines in round 1 (Table 5.47), and neither of the definitions evaluated in round 2 (Table 5.48). Participant comments included: that the Canadian Guidelines definition prevents early diagnosis due to the need for cognitive testing; that alcohol exposure can be unknown and should not be a determining factor for diagnosis; that all three statements meet the diagnostic criteria for Static Encephalopathy alcohol exposed (SE); that Neurobehavioural Disorder should be included as a diagnostic classification that has been validated; that the terminology is an inadequate reflection of the multifactorial and often unknown aetiology of neurological impairments and this labelling of complex problems can be counterproductive, with a need for more research before these FASD diagnoses are used; that it is

difficult to know what is significant prenatal alcohol exposure; that differential diagnosis should exclude other causes; and that a universal health record would assist diagnosis.

Table 5.47 Round 1 statement ratings: general diagnostic criteria for ARND or SE

Statement: A diagnosis of ARND or SE in the absence of FAS requires:	N	Agree (%)	Median	IQD
S13Q4. confirmed prenatal alcohol exposure, **and** evidence of CNS abnormality (decreased cranial size, abnormal brain structure or neurological hard or soft signs, including fine motor skills, neurosensory hearing loss and co-ordination), **or** evidence of multiple behavioural or cognitive abnormalities that are inconsistent with developmental level (e.g. learning, academic achievement, poor impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement)	43	**72.1**	2	2
S13Q5. confirmed prenatal alcohol exposure, **and** evidence of decreased cranial size or abnormal brain structure	42	40.5	3	2
S13Q6. confirmed prenatal alcohol exposure, **and** CNS abnormality in 3 of the following areas (hard and soft neurologic signs, brain structure, cognition, communication, academic achievement, memory, executive functioning and abstract reasoning, attention deficit or hyperactivity, adaptive behaviour, social skills, social communication)	42	66.7	2	1

Table 5.48 Round 2 statement ratings: general diagnostic criteria for ARND or SE

Statement: A diagnosis of ARND or SE in the absence of FAS requires:	N	Agree (%)	Median	IQD
S6Q4. confirmed prenatal alcohol exposure, **and** evidence of CNS abnormality (decreased cranial size, abnormal brain structure or neurological hard or soft signs, including fine motor skills, neurosensory hearing loss and co-ordination), **or** evidence of multiple behavioural or cognitive abnormalities that are inconsistent with developmental level (e.g. learning, academic achievement, poor impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement)	53	62.3	2	2
S6Q5. confirmed prenatal alcohol exposure, **and** CNS abnormality in 3 of the following areas (hard and soft neurologic signs, brain structure, cognition, communication, academic achievement, memory, executive functioning and abstract reasoning, attention deficit or hyperactivity, adaptive behaviour, social skills, social communication)	50	58.0	2	2

In round 1, consensus agreement was not reached on the general definition of the diagnostic criteria for ARBD (Table 5.49) or the usefulness of this diagnostic category (Table 5.50). Participant comments included: the need to be more specific about musculoskeletal deformities; that the case definition must be refined to include birth defects with a high likelihood of being related to alcohol exposure; that the differential diagnosis must exclude other causes; and lack of agreement with the terminology 'alcohol-related'.

Table 5.49 Round 1 statement ratings: general diagnostic criteria for ARBD

Statement: A diagnosis of ARBD in the absence of FAS requires:	N	Agree (%)	Median	IQD
S13Q8. confirmed prenatal alcohol exposure, **and** identification of alcohol-related birth defects on clinical examination (including cardiac, skeletal, renal, ocular, auditory or other malformations, including facial anomalies)	46	69.6	2	1

Table 5.50 Round 1 statement ratings: usefulness of ARBD as a diagnostic category

Statement	N	Agree (%)	Median	IQD
S13Q10. Alcohol-related birth defects is not sufficiently well defined to be a useful diagnostic category	43	55.8	2	2

In round 2, consensus agreement was reached on the lack of evidence to support the diagnostic category of ARBD (Table 5.51). General comments included: that ARBD is required as a diagnostic category and may become more concrete over time; that significant alcohol exposure should be defined and the level of alcohol exposure taken into account when making a diagnosis of ARBD; and that the diagnostic term needs to be clinically useful, is rarely used, and cannot be medically confirmed.

Table 5.51 Round 2 statement ratings: general diagnostic criteria for ARBD

Statement: A diagnosis of ARBD in the absence of FAS requires:	N	Agree (%)	Median	IQD
S6Q2. confirmed prenatal alcohol exposure, **and** identification of alcohol-related birth defects on clinical examination (including cardiac, skeletal, renal, ocular, auditory or other malformations, including facial anomalies)	54	33.3	3	2
S6Q3. confirmed significant prenatal alcohol exposure, **and** identification of alcohol-related birth defects on clinical examination (including cardiac, skeletal, renal, ocular, auditory or other malformations, including facial anomalies)	56	62.5	2	1
S6Q1. Better evidence of the association between alcohol and particular birth defects is required for alcohol-related birth defects (ARBD) to be a clinically useful diagnostic category	65	**90.8**	2	1

6. WORKSHOP METHODS

Key Points

- Steering Group members met to consider the evidence from the literature review, Community Conversations and Delphi survey
- A range of methods were used to develop consensus on the content of screening and diagnostic instruments for FASD

PURPOSE

The purpose of the workshop was to establish consensus on:

1. The content of a diagnostic instrument for FASD in Australia.
2. The content of a screening instrument for FASD in Australia.
3. Recommendations to support the implementation of a standardised approach to FASD screening and diagnosis in Australia.

This consensus development workshop brought Steering Group members together to consider the evidence on FASD screening and diagnosis identified in a systematic literature review, the Community Conversations and the Delphi survey of health professionals, in the context of their own expertise and experience. The nominal group technique was used, in combination with other methods, as a basis for the design of the workshop to develop consensus on the content of instruments for FASD screening and diagnosis in Australia. The nominal group technique is an established method for conducting structured group meetings that facilitates efficient problem exploration and consensus development (Delbecq, et al., 1975; Gallagher et al., 1993) and has been commonly applied in the health context (Bond & Watson, 2003; Carney et al., 1996; Trickey et al., 1998).

PARTICIPANTS

Thirteen of the seventeen members of the Steering Group attended the 2-day workshop in July 2011. The workshop was jointly facilitated by a member of the Steering Group, with facilitation experience, and the Project Analyst. The Project Manager co-ordinated the workshop activities, the preparation and distribution of workshop resources, and performed the role of scribe. The twelve remaining Steering Group members attended the workshop as participants. All twelve workshop participants had experience in FASD research, nine participants were qualified health professionals, including three practising paediatricians, and two participants were consumer and community representatives who had

extensive experience in consumer and community advocacy, support and training about FASD. The diverse expertise of Steering Group members provided a basis for rich discussion and exploration of this complex issue. Three Steering Group members who were unable to attend the workshop (a paediatrician, a paediatric senior registrar and a drug and alcohol researcher with a nursing background) participated in a post-workshop review of workshop outcomes as outlined below.

PRE-WORKSHOP PROCEDURE

The Steering Group, including workshop participants, met monthly via teleconference during the project (August 2010 - August 2011) to design, review and oversee the collection and evaluation of evidence to be considered at the workshop. Information from these preliminary evaluation processes was documented, summarised and circulated to workshop participants prior to the workshop. Evidence reviewed prior to the workshop included the literature review (Appendix D: Vol 2 of this Report) and Community Conversations (Appendix E: Vol 2 of this Report), as well as findings from the Delphi survey which were summarised in three short reports. These three short reports aimed to place the Delphi survey findings in the context of the current literature on the screening and diagnosis of FASD.

WORKSHOP PROCEDURE

A flexible approach was used to conduct each workshop session in recognition of the challenges involved in reaching consensus on this complex topic with a diverse group of experts who met face-to-face as a group for the first time at this workshop. A Lead Investigator and facilitator opened the workshop and group work commenced with an introductory, participant-driven, global conceptual mapping of the proposed screening and diagnostic process for FASD in Australia. Presentations of results from the Delphi survey were then delivered to facilitate the review of findings on the diagnosis of FASD in Australia. Two workshop participants who were paediatricians experienced in making FASD diagnoses each delivered a presentation from a clinical perspective on the strengths and limitations of the two existing diagnostic guidelines that were most widely supported in the Delphi survey (UW and Canadian Guidelines). Following these presentations an open group session allowed participants to discuss and debate alternative approaches to diagnosis. During this discussion participants agreed that specific justification would be required if workshop recommendations were contrary to the findings from the Delphi survey of health professionals.

The nominal group process (Delbecq, et al., 1975) commenced with participants engaging in individual reflection for five minutes to identify their ideas in response to the following task statement: ‘You are familiar with the literature review and Delphi study findings, and have heard clinician perspectives. We

now need you to use your expertise and experience to identify what YOU think should be in the diagnostic instrument.' The facilitator then asked participants to report ideas one by one to the group, and a scribe listed all ideas on a flipchart. Facilitated group discussion was used to clarify the ideas, and identify their logic and importance. The list of ideas was then sorted into coherent groups (Bartunek & Murnighan, 1984), and through the use of voting, participants evaluated the inclusion of each idea in the diagnostic instrument. Group consensus was not established on the diagnostic criteria for FASD, and additional extended group discussion and a formal voting process were used to identify which diagnostic criteria should be included in the diagnostic instrument.

It was agreed during the workshop that a diagnostic subgroup of the Steering Group should review the workshop outcomes related to diagnosis and provide final confirmation of the recommended diagnostic criteria and final form of the diagnostic instrument. This was due to the inability of all Steering Group members with relevant diagnostic experience to attend the workshop, and the scope and complexity of information relevant to the diagnostic criteria and diagnostic assessment process reviewed during the workshop.

Subsequent workshop sessions were conducted using similar discussion and voting processes to develop consensus on additional components of the screening and diagnostic instruments, including detailed criteria for diagnosis and referral for a diagnostic evaluation. These sessions were also guided by the review of relevant findings from the Delphi survey and information on existing diagnostic guidelines, and this information was used as a basis for discussion and criteria generation. During the session on development of a screening instrument, a participant also read out the key issues identified during the Community Conversations to help direct the discussion. Potential instrument components and criteria were evaluated using facilitated group discussion, small group discussion, and small group activities, including the construction of screening and diagnostic instruments. In the final session of the workshop facilitated discussion was used to identify conclusions and recommendations to support the implementation of screening and diagnostic instruments for FASD in Australia.

POST-WORKSHOP PROCEDURE

Following the workshop, the diagnostic subgroup of the Steering Group, which included two medical practitioners and four paediatricians, reviewed the workshop outcomes related to diagnosis, confirmed the recommended diagnostic criteria and constructed the final diagnostic instrument. Diagnostic subgroup members met by teleconference, and 5 subgroup members and two facilitators attended a final face-to-face review meeting in April 2012. A three-member consumer subgroup of the Steering

Group was also formed to review the workshop outcomes related to consumer resources and construct the final consumer resources for inclusion in the diagnostic instrument.

Recorded outcomes of formal and informal voting processes, and transcribed workshop discussions were used to draft this report of workshop findings. Qualitative descriptive analysis (Sandelowski, 2000) of participant contributions in open discussion sessions, based on identifying and categorising the underlying meaning of participant contributions and statements (Streubert-Speziale & Carpenter, 2003), was used to describe the main discussion content. All participants checked the description of workshop findings for consistency.

Due to the broad scope of the information generated in the workshop, and the additional contributions received from the diagnostic and consumer subgroups, all Steering Group members were asked to review the workshop report to ensure that group processes and findings were accurately reported. The final workshop report was approved by all Steering Group members.

7. WORKSHOP RESULTS

Key Points

- A diagnostic instrument for FASD for use in Australia was developed with agreed diagnostic criteria based on both the UW and Canadian Guidelines
- Diagnostic categories include FAS, PFAS and ND-AE
- Criteria for targeted screening for FASD were proposed; however, the criteria require evaluation to establish their validity and cost effectiveness in at risk populations
- Improved diagnostic capacity for FASD in Australia and training of health professionals were identified as high priorities

DEVELOPMENT OF A DIAGNOSTIC INSTRUMENT

Participant contributions on the content of a diagnostic instrument proposed for use in Australia were sorted into two coherent categories, reflecting:

1. Principles of instrument design
2. Instrument content

PRINCIPLES OF INSTRUMENT DESIGN

Workshop participants identified that the following general principles should be incorporated in the design of an instrument for the diagnosis of FASD in Australia. The diagnostic instrument needs to include standardised assessment protocols, use terminology that is understood by all health professionals, be easy to implement, be applicable in diverse settings, and be suitable for use across the lifespan. To support specific high quality evaluation, the diagnostic instrument should: provide a pro forma for all required examinations, investigations and assessments; facilitate the use of valid and reliable assessment methods; and include a standardised method to assess alcohol consumption.

The instrument must be linked to comprehensive guidelines for the diagnosis of FASD and training for health professionals that includes how to perform a diagnostic assessment among different age and cultural groups and in diverse settings. Participants also required that the instrument be compatible with an interdisciplinary approach to assessment, and facilitate standardised national reporting of FASD diagnoses. Guided by the diagnostic instrument, the diagnostic process should direct and inform management, and ensure that support is available for the individual and family, parent or carer.

It was also considered important to establish standard processes to set the agenda for the diagnostic assessment. Parents, carers and individuals should be informed about the diagnostic and management process and goals prior to the diagnostic assessment, and provided with access to resources that offer on-going support. Informed consent should also be obtained and recorded prior to the communication of diagnostic findings to other individuals or organisations, excluding mandatory reporting requirements.

DIAGNOSTIC INSTRUMENT CONTENT

All participants agreed on the essential components of the diagnostic instrument as listed in Table 7.1. Three different diagnostic criteria were proposed for inclusion in the instrument: the UW criteria, Canadian criteria, or a combination of the UW and Canadian criteria. Formal voting was used to identify group consensus on the diagnostic criteria to be included in the proposed instrument. Voting indicated that the combination of UW and Canadian criteria had the highest level of support (71.7%) followed by the UW criteria (15.0%) and the Canadian criteria (13.3%).

Both the UW and the Canadian Guidelines recommend that the UW 4-Digit Diagnostic Code should be used in the diagnostic assessment to document the presence and severity of facial anomalies, CNS abnormalities, growth deficit, and PAE. A review of the UW New Patient Information Form and the UW FASD Diagnostic Form (Astley, 2004) found that all required areas of instrument content identified by participants were included (Table 7.1). Thus modified versions of these diagnostic forms might be appropriate for use in Australia. Although areas of strength are not explicitly identified on the UW FASD Diagnostic Form, the UW Guidelines indicate that a comprehensive assessment will identify areas of strength as well as areas of impairment, and that documentation of the outcomes of all areas of assessment should be incorporated in the assessment findings to inform treatment planning (Astley, 2004/pp.40).

The AUDIT-C (Bush et al., 1998) was supported by workshop participants as a useful standard tool to assess alcohol consumption during pregnancy. It is currently recommended by the DoHA for assessment of alcohol intake during pregnancy as part of Pregnancy Lifescripts (Australian Government Department of Health and Ageing, 2006). The assessment is quick and easy to complete and the score derived indicates the level of risk to the woman and fetus from alcohol use. Participants recommended that the AUDIT-C be used in combination with a clinical interview to obtain more detailed information about, or alternative indicators of, consumption patterns and timing. Workshop participants recognised that detailed and accurate information on prenatal alcohol consumption will not always be available.

The UW FASD Diagnostic Form is used to collect similar information to the AUDIT-C, including the frequency and usual amount of alcohol consumed, and the maximum number of drinks per occasion. In addition, the UW FASD Diagnostic Form includes fields to record information on alcohol consumption prior to and during pregnancy, the timing of alcohol consumption during pregnancy, reported alcohol dependence, and other information that might help describe the mother's level of alcohol use during pregnancy.

Table 7.1 Required content of a standard diagnostic instrument for FASD

Instrument required content	**Content included on the UW FASD Diagnostic Form or New Patient Information Form**
History	-
Family/social	Yes
Prenatal medical	Yes
Obstetric	Yes
Neonatal	Yes
Academic	Yes
Developmental	Yes
Current problems	Yes
Pre + post natal alcohol + other prenatal exposures	Yes
Paternal drinking	Yes
Drug and alcohol use in the child or individual	Yes
Early life trauma	Yes
Examination	-
Growth	Yes
Head circumference	Yes
Dysmorphology	Yes
CNS	Yes
Birth defects	Yes
Medical investigations	Yes
Diagnostic criteria (combined UW and Canadian[†])	Yes (UW criteria)
Exclusion of other diagnoses	Yes
Reporting final diagnosis by category	Yes
Results summary: strengths and areas of need	Yes
Follow-up/management plan	Yes

[†]Diagnostic criteria originally proposed for inclusion in the instrument by workshop participants were the UW criteria, the Canadian criteria, and a combination of the UW and Canadian criteria. A combination of the UW and Canadian criteria was selected using a formal vote.

DIAGNOSTIC CATEGORIES

There was consensus among workshop participants that the UW diagnostic terminology should be modified for use in the clinical environment to ensure that the diagnostic instrument is easy to implement, and meaningful to clinicians, individuals with FASD and their families. Workshop participants agreed that the term SE should not be adopted in Australia as it is not commonly used and fails to describe the nature of the observed impairment. The term ARND was also rejected due to the potentially unknown and multifactorial causes of neurodevelopmental disorders.

Diagnostic categories for FASD included in the instrument for use in Australia are: FAS, PFAS and Neurodevelopmental Disorder-Alcohol Exposed (ND-AE). The diagnostic categories for use in Australia reflect the categories used in the Canadian Guidelines, although the terminology differs from that used in the UW and Canadian Guidelines. The diagnostic term ND-AE is used in place of the terms SE (UW Guidelines) and ARND (Canadian Guidelines), and uses the UW Guidelines convention of designating a diagnostic category as alcohol exposed, rather than alcohol-related.

Consistent with both the UW and Canadian Guidelines, ARBD was not considered a clinically useful diagnostic category and was excluded from the proposed instrument. However, it was emphasised that all birth defects identified during the diagnostic assessment should be recorded on the diagnostic instrument. In accordance with four existing diagnostic guidelines (Canadian, CDC, Hoyme, IOM), workshop participants agreed to omit the UW diagnostic category of Neurobehavioural Disorder-Alcohol Exposed from the proposed diagnostic criteria, and identified the need for further evidence on the validity and usefulness of this diagnostic category in Australia.

DIAGNOSTIC CRITERIA

Participants recognised that there are only minor differences between diagnostic criteria recommended by different groups, but that there is a growing evidence base for the UW diagnostic criteria. They agreed that the diagnostic instrument should include a combination of elements from both the UW and Canadian diagnostic criteria as outlined in Tables 7.2-7.4. With regard to the diagnostic criteria for PFAS, we have removed the requirement for the presence of a growth deficit.

There was considerable debate among workshop participants about the recommended criteria for the diagnosis of CNS abnormality, identifying the criteria to be of critical importance to the diagnosis of FASD and indicating a perceived lack of evidence to describe the relative performance of alternative criteria. Discussion identified a lack of agreement about the adequacy of microcephaly alone as an

indicator of CNS damage. There was only limited support for the inclusion of CNS dysfunction in two domains instead of three, to fulfil the criteria for the diagnostic category ND-AE. Consistent with the Delphi survey findings, the UW criteria for CNS abnormality, based on a significant structural abnormality or significant dysfunction in three or more domains, were provisionally endorsed by workshop participants, pending a review of this decision by the diagnostic subgroup following the workshop.

After considerable debate, the diagnostic subgroup subsequently endorsed the requirement for significant dysfunction in at least three CNS domains for the diagnostic category ND-AE. The high level of dysfunction required to fulfil this diagnostic criterion was acknowledged, and future evaluation of the validity of diagnosis based on two rather than three domains of dysfunction was recommended. Although the diagnostic subgroup also debated revision of the requirement for confirmed prenatal alcohol exposure for the diagnosis of ND-AE to confirmed high-risk prenatal alcohol exposure, the requirement for confirmed prenatal alcohol exposure was maintained.

Appropriate population growth standards for use in Australia also need to be identified, particularly among Indigenous Australians and those from culturally diverse backgrounds. An evaluation of the performance of WHO Child Growth Standards (World Health Organization Multicentre Growth Reference Study Group, 2006) and the CDC Clinical Growth Charts (Centers for Disease Control and Prevention, 2000) for FASD screening in a remote Indigenous population in Western Australia is currently underway as a part of The Lililwan Project: FASD prevalence in the Fitzroy Valley (Latimer et al., 2010).

Having taken into consideration the literature and the results of the Delphi survey, workshop participants agreed on the diagnostic criteria for FAS as shown in Table 7.2, PFAS as shown in Table 7.3, and ND-AE as shown in Table 7.4.

Table 7.2 Diagnostic criteria for Fetal Alcohol Syndrome (FAS)

	FAS: diagnostic criteria
General	Requires all 4 of the features listed below
Prenatal alcohol exposure	Confirmed or unknown prenatal alcohol exposure
Facial anomalies[†]	All 3 characteristic FAS facial anomalies at any age: short palpebral fissure length (2 or more SD below the mean); smooth philtrum (Rank 4 or 5 on the UW Lip-Philtrum Guide); and thin upper lip (Rank 4 or 5 on the UW Lip-Philtrum Guide)
Growth[†]	Prenatal or postnatal growth deficit indicated by birth length or weight ≤ 10th percentile adjusted for gestational age, or postnatal height or weight ≤ 10th percentile
CNS[†]	At least 1 of the following: i. Structural abnormality (e.g. OFC ≤ 3rd percentile, abnormal brain structure), or neurological abnormality (seizure disorder or hard neurological signs), **or** ii. Severe dysfunction (impairment in 3 or more domains of function, 2 or more SDs below the mean) [‡]

CNS-central nervous system; SD-standard deviation; OFC-occipital-frontal circumference

[†]Please refer to the Guide to the Australian FASD Diagnostic Instrument (Appendix A1) for information on the diagnostic assessment process

[‡]The indication of impairment will be guided by the parameters of the tests used and clinician judgement

Table 7.3 Diagnostic criteria for Partial Fetal Alcohol Syndrome (PFAS)

	PFAS: diagnostic criteria
General	Requires confirmed prenatal alcohol exposure, the presence of 2 of the 3 characteristic FAS facial anomalies, and CNS criteria to be met
Prenatal alcohol exposure[†]	Confirmed prenatal alcohol exposure
Facial anomalies[†]	Presence of any 2 of the following facial anomalies at any age: short palpebral fissure length (2 or more SD below the mean); smooth philtrum (Rank 4 or 5 on the UW Lip-Philtrum Guide); and thin upper lip (Rank 4 or 5 on the UW Lip-Philtrum Guide)
Growth	No deficit required
CNS[†]	At least 1 of the following: i. Structural abnormality (e.g. OFC ≤ 3rd percentile, abnormal brain structure), or neurological abnormality (seizure disorder or hard neurological signs), **or** ii. Severe dysfunction (impairment in 3 or more domains of function, 2 or more SDs below the mean) [‡]

CNS-central nervous system; SD-standard deviation; OFC-occipital-frontal circumference; PAE-prenatal alcohol exposure

[†]Please refer to the Guide to the Australian FASD Diagnostic Instrument (Appendix A1) for information on the diagnostic assessment process

[‡]The indication of impairment will be guided by the parameters of the tests used and clinician judgement

Table 7.4 Diagnostic criteria for Neurodevelopmental Disorder-Alcohol Exposed (ND-AE)

	ND-AE: diagnostic criteria
General	Requires confirmed prenatal alcohol exposure and CNS criteria to be met
Prenatal alcohol exposure[†]	Confirmed prenatal alcohol exposure
Facial anomalies	No anomalies required
Growth	No deficit required
CNS[†]	At least 1 of the following: i. Structural abnormality (e.g. OFC ≤ 3rd percentile, abnormal brain structure), or neurological abnormality (seizure disorder or hard neurological signs), **or** ii. Severe dysfunction (impairment in 3 or more domains of function, 2 or more SDs below the mean) [‡]

CNS-central nervous system; SD-standard deviation; OFC-occipital-frontal circumference; PAE-prenatal alcohol exposure

[†]Please refer to the Guide to the Australian FASD Diagnostic Instrument (Appendix A1) for information on the diagnostic assessment process

[‡]The indication of impairment will be guided by the parameters of the tests used and clinician judgement

The Australian FASD Diagnostic Instrument was developed based on the workshop findings and is included in Appendix A. This instrument is based on a modified version of the UW FASD Diagnostic Form that incorporates the diagnostic categories and criteria agreed during the workshop and indicators of PAE derived from the AUDIT-C. This instrument will require evaluation prior to its use in the clinical environment.

DEVELOPMENT OF A SCREENING INSTRUMENT

Consistent with the Delphi survey findings, workshop participants did not support the establishment of dedicated universal screening programs for FASD at this stage. Opportunities to conduct screening for FASD within current programs were identified, should a feasible and effective screening instrument be developed. Workshop participants agreed that an instrument for targeted screening for FASD is required in Australia. Moderated group discussion revealed group consensus around four main issues associated with the development of a targeted screening instrument for FASD in Australia.

> *"Primary care providers would not see the point in screening if their experience is that it is impossible to get help or that there are no effective management strategies."*
> Delphi Study participant

> *"Screening can only occur once there is diagnostic and intervention capacity."*
> Delphi Study participant

1. Improved identification

   Targeted screening for FASD was considered necessary to facilitate the early identification of individuals and families in need of services, and enable the provision of information, referral and support. Systems are also required to enable individuals and families to understand the screening and diagnostic process, the implications of diagnosis, and to enable access to support while waiting for a diagnostic evaluation. Targeted screening was also recognised as an important mechanism to improve current estimates of the prevalence of FASD and provide valuable information for service delivery and the design of prevention programs.

2. Diagnostic capacity

   Although workshop participants thought that screening for FASD was required to document the need for diagnostic and intervention services for FASD, they agreed that targeted screening for FASD should not be implemented without ensuring access to such services. In view of the current limited capacity for FASD diagnosis in Australia, including the lack of access to interdisciplinary assessment clinics, workshop participants agreed that the national implementation of standard guidelines for FASD diagnosis and the establishment of interdisciplinary diagnostic clinics to improve diagnostic capacity is an immediate priority.

3. Screening program design

   Participants agreed that screening should be a quick and efficient process, suitable for use in a range of settings, and for administration by a range of health professionals with training in the use of the screening instrument. Relevant information from birth records such as growth, head circumference and alcohol exposure should be integrated into the screening process. Currently there is a proposal that data about alcohol exposure during pregnancy is incorporated in the minimum data set for the national perinatal data collection.

   Screening for FAS and PFAS was recognised as more straightforward, better established, and likely to have better performance characteristics than screening for ND-AE. However, screening for the full range of FASD was preferred, as there was concern that ND-AE may be overlooked if screening was limited to FAS and PFAS. An inclusive screening model for FASD in Australia would improve the identification and management of all individuals with FASD.

4. Screening program evaluation

   Participants identified the novel nature of screening for FASD in Australia and the need to evaluate screening performance according to classical criteria prior to implementation. These include

whether screening delivered by trained health professionals can yield sufficient sensitivity, specificity, predictive value and cost-effectiveness. Reported difficulties with the clinical assessment of facial anomalies and the application of the UW Lip-Philtrum Guides and the UW FAS Facial Photographic Analysis Software in culturally diverse populations, including Indigenous populations, also require investigation. Appropriate facial anomaly assessment tools and reference data must be established for the Australian population.

> *"[We]…should consider identifying the positive areas of all guidelines and develop Australian guidelines for diagnosis. Within all guidelines there are techniques that are the same or similar. I agree that we should not reinvent the wheel but I feel that here in Australia we have a unique situation especially relating the facial characteristics of Indigenous people."*
> Delphi Study participant

SCREENING INSTRUMENT CONTENT

Workshop participants identified four assessment components required for FASD screening.

1. Prenatal alcohol exposure

   Consistent with the Delphi survey results, workshop participants agreed that assessment of PAE should include the standard AUDIT-C questions alongside non-judgemental enquiry about alcohol consumption and other lifestyle factors and indicators of alcohol dependency. The assessment should identify exposures immediately prior to pregnancy and after the pregnancy is confirmed. Screening at birth should also include the assessment of alcohol withdrawal in the mother and neonate.

2. Characteristic FAS facial anomalies

   Assessment for the thin vermillion border and smooth philtrum characteristics of FAS should be conducted using the UW Lip-Philtrum Guide. Ranks 4 and 5 are considered abnormal. Physical measurements of palpebral fissure length by inexperienced assessors may be unreliable. Ideally, measurement of palpebral fissure length should be performed by trained and experienced assessors. The UW Facial Photographic Analysis Software, which enables measures to be made using a digital photograph, may be appropriate for use in specific screening contexts.

3. Growth

   Growth should be assessed using standard objective methods and compared with population references, with correction for gestational age if required.

4. Central nervous system abnormalities

   Assessment of CNS abnormalities at the screening stage should include measurement of head circumference. Other indicators of CNS abnormality may be identified in: the physical examination; reports by parents, carers or other credible sources; formal assessments of CNS function, including the results of investigations or scans; or the results of valid and reliable behavioural and developmental screening tools e.g. Parents' Evaluation of Developmental Status (PEDS) (Centre for Community Child Health The Royal Children's Hospital), Ages and Stages Questionnaire (ASQ) (Paul H. Brookes Publishing Co. Inc., 2011), Strengths and Difficulties Questionnaire (SDQ) (Goodman, 1997). Participants acknowledged that different screening tests for CNS abnormalities are required for different age groups and in different settings. A range of valid and reliable developmental and behavioural screening tools that are routinely used could indicate the need for referral.

DEVELOPMENT OF CRITERIA FOR REFERRAL

Although workshop participants concluded that further evidence on the performance of screening criteria is required to enable the development of an instrument for screening for FASD in Australia, agreed criteria for referral were developed to provide guidance for health professionals on the need to refer individuals for specialist assessment. These criteria for referral for diagnosis were based on a review of the Delphi survey findings and existing criteria for referral included in the Canadian and CDC Guidelines. Workshop participants endorsed five independent criteria for referral as outlined in Table 7.5. Referral was considered appropriate if individuals satisfied all the requirements of any of the five criteria for referral.

The inclusion of a criterion for referral based only on confirmed PAE was the most contentious element of the agreed criteria for referral, with strong debate among workshop participants on the ability to identify and assess an appropriate level of PAE sufficient to warrant referral. Anecdotal evidence discussed at the workshop on the adverse effects of PAE was consistent with advice that there is no known minimum safe level of exposure during pregnancy. However, this was contrasted with current empirical evidence that suggests adverse outcomes are significantly linked to moderate or high-level PAE. In addition, the development of criteria for screening must consider other factors that influence the ability to report PAE, and that the factors mediating prenatal alcohol-related harm are not fully understood. Workshop participants decided that referral based on PAE alone should be limited to reported or suspected moderate or high levels of consumption, similar to the Canadian and CDC Guidelines.

In contrast with existing referral criteria included in the Canadian and CDC Guidelines, the presence of two characteristic FAS facial anomalies were considered by workshop participants to be sufficient to refer for a diagnostic evaluation due to the potential unreliability of palpebral fissure measurement by inexperienced assessors, and the importance of these findings. Agreement with referral based on the existing CDC criterion of the presence of a facial anomaly, growth deficit and CNS anomaly provides a second means for referral that does not require the presence of confirmed PAE.

The final two agreed criteria for referral listed in Table 7.5 include any confirmed PAE as a common element in conjunction with the specified CNS abnormalities, and provide a mechanism for the referral of individuals with possible ND-AE, for which confirmed PAE is required for diagnosis. Participants agreed that these criteria for referral require evaluation to ensure that they effectively and efficiently identify individuals at high risk of FASD. Guidelines and training resources for health professionals to support the use of the criteria also need to be developed.

Table 7.5 Criteria for referral and their relation to the Canadian and CDC criteria.

Proposed criteria for referral	**Equivalent existing Canadian criteria**	**Equivalent existing CDC criteria**
1. Confirmed moderate or high level[†] prenatal alcohol exposure	Yes	Yes
2. 2 or more characteristic FAS facial anomalies	No	No
3. 1 facial anomaly and growth deficit and 1 or more CNS abnormalities	No (Yes)[‡]	Yes
4. Microcephaly and any confirmed prenatal alcohol exposure	No (Yes)[‡]	No
5. 2 or more CNS abnormalities and any confirmed prenatal alcohol exposure	No (Yes)[‡]	No

CDC-Centers for Disease Control; [†]based on the Delphi survey findings, moderate or high level exposure was defined as exposure to 7+ standard drinks per week, or regular exposure to 5+ standard drinks on any one occasion, or strong clinical suspicion of heavy prenatal alcohol exposure; [‡]equivalent criteria not included in primary description of the Canadian Guidelines (Chudley et al., 2005), but related criteria are included in a published description of the Canadian Guidelines (Loock et al., 2005).

WORKSHOP CONCLUSIONS AND RECOMMENDATIONS

DIAGNOSIS

Workshop participants developed a diagnostic instrument for FASD for use in Australia (Appendix A). The instrument is based on a revision of the UW FASD Diagnostic Form which has been modified to include information relevant to the diagnostic categories and criteria required for Australia. The Australian diagnostic instrument needs to be accompanied by comprehensive guidelines for its use and training resources for health professionals, which should be developed based on the existing UW

Guidelines and training resources. The Australian FASD Diagnostic Instrument also requires pilot evaluation to demonstrate its clinical applicability and appropriateness in the Australian context, and facilitate implementation and adoption by health service providers. These diagnostic methods require the availability of accurate and locally appropriate population reference data and assessment tools, and the validity of using existing population reference data among Indigenous Australians and those from culturally diverse backgrounds must also be established.

An implementation plan is required to embed the diagnostic instrument in a model of care that facilitates adoption of standard diagnostic practices. The implementation plan should identify strategies for: improving health professionals' awareness of national diagnostic guidelines for FASD; facilitating adoption of standard diagnostic practices; and providing training and support for health professionals. Support services and resources should also be established for individuals undergoing diagnosis and their families, parents and carers. Implementation planning should also include, and be based on, the findings of a health economic assessment.

In summary, to support the implementation of the standard diagnostic instrument, workshop participants endorsed the need to:

1. Evaluate and demonstrate the clinical applicability of the proposed diagnostic instrument in at least two interdisciplinary clinics to ensure its suitability for the Australian context.
2. Establish the validity of using available standard reference data for growth, head circumference and facial features in the Australian population, including among Indigenous Australians.
3. Develop a national implementation plan to embed the diagnostic resources in a sustainable model of care, and identify strategies for instrument dissemination, adoption, training and support.

SCREENING

There is little information to guide the development of targeted screening for FASD in Australia. Workshop participants concluded that the proposed screening criteria require performance evaluation prior to implementation to establish their sensitivity, specificity, predictive value and cost effectiveness in at-risk populations. Evaluation of the screening criteria will enable the development of a final checklist for screening and referral, and guidelines for its use. Forthcoming data from a high risk Indigenous population (The Lililwan Project: FASD prevalence in the Fitzroy Valley (Latimer, et al., 2010)), a high risk foster care population in Western Australia and state referral clinics in Western Australia and New South Wales could be used to develop and evaluate a targeted screening instrument.

In summary, to support the development and implementation of an instrument for targeted screening for FASD in Australia, workshop participants endorsed the need to:

4. Develop a screening and referral instrument and evaluate its performance in high risk populations, including sensitivity, specificity, predictive value and cost effectiveness.
5. Develop a national implementation plan and detailed guidelines for use which identify strategies for screening instrument dissemination, adoption, training and support.

REFERRAL

Workshop participants concluded that the agreed criteria for referral, which were developed for use in conjunction with the screening instrument, could be used in a stand-alone capacity to guide health professionals on the need to refer individuals for specialist assessment. The effectiveness of the agreed criteria for referral would be evaluated with the screening instrument. Brief guidelines should be developed to support the use and implementation of the proposed criteria for referral. Training for health professionals and materials for individuals, families, parents and carers are also required.
In summary, to support the implementation of the agreed criteria for referral for a diagnostic evaluation for FASD in Australia, workshop participants endorsed the need to:

6. Develop a referral checklist and guidelines for its use and implementation, including strategies for dissemination, training and support.

CLOSING REMARKS

In the closing session of the workshop participants expressed optimism about the progress made toward improving the capacity for screening, diagnosis and management of FASD in Australia, and a strong commitment to the implementation of these findings to ultimately benefit all Australians who are affected by FASD. Participants also spoke of a sense of collective purpose, where outcomes were created through the combined efforts of a diverse range of experts who each contributed with equal voice. We thank all individuals involved in this initiative, and particularly the health professionals and community members who took time to share their knowledge and insights in earlier stages of this project, providing the foundation on which these workshop outcomes are based.

> *"Privilege to be part of the group and value the input of the community representatives"*
> Workshop participant

8. CONCLUSIONS

Consensus was reached on the development of a diagnostic instrument for FASD for use in Australia, on the diagnostic criteria for FASD and on the referral criteria for FASD. However, there was insufficient evidence to support the development of a screening instrument. There are few formal screening instruments for FASD, and limited empirical information on which to base the design of an Australian screening instrument. Specific conclusions are presented below.

DIAGNOSIS

National diagnostic criteria for FASD are required to provide a foundation for improved diagnostic capacity in Australia. National diagnostic criteria would promote consistent diagnostic practices and help identify the size of the FASD population. This in turn would improve the evidence base for prevention, diagnosis, and management. We have developed a diagnostic instrument for FASD for use in Australia. It includes nationally applicable criteria for diagnosis that are based on both the UW and Canadian Guidelines. Use of the UW diagnostic instrument as the foundation for the proposed diagnostic instrument was based on its use of specific, quantifiable diagnostic criteria and the accumulated evidence base resulting from its use. This outcome also recognises that the UW Guidelines were most commonly recommended for adoption in the Delphi survey.

The UW and Canadian diagnostic criteria have been incorporated into a diagnostic instrument for FASD that is simple to use and appropriate for use in the Australian clinical context. Some revisions to the UW diagnostic criteria were considered necessary. In common with the Canadian Guidelines, only the three most well established diagnostic categories are included. Growth deficit was excluded from the diagnostic criteria for PFAS, which is consistent with the Canadian Guidelines, and the UW diagnostic category 'Static Encephalopathy alcohol exposed' was renamed 'Neurodevelopmental Disorders – Alcohol Exposed'. Our recommendation is that FASD be reported in Australia using the three diagnostic categories Fetal Alcohol Syndrome, Partial Fetal Alcohol Syndrome and Neurodevelopmental Disorders-Alcohol Exposed, but this does not preclude the use of the UW diagnostic terminology for research or other purposes.

In light of the modifications made to the UW diagnostic criteria it will be important to develop comprehensive guidelines and training resources specifically for use in the Australian context. A brief guideline (Appendix A1) was produced for clinicians to facilitate use of the diagnostic instrument in the clinical setting, and information for individuals, families and clinicians about the diagnostic process was

also developed (Appendix A5-A6). The diagnostic instrument and training resources should undergo pilot evaluation prior to national implementation to establish their clinical applicability and validity in the Australian context, and facilitate their later adoption by health service providers.

The appropriateness of available population reference data for palpebral fissure length, growth and tests for functional CNS abnormalities must be established in the Australian population, including among Indigenous Australians and those from culturally diverse backgrounds. This also applies to the UW Lip-Philtrum Guides which are normalised to Caucasians and African Americans. Research should also identify challenges encountered in the use of the diagnostic instrument and the delivery of diagnostic services, and facilitate the improvement of the diagnostic methods used in Australia.

A multidisciplinary approach to diagnosis is recommended and the ideal is an interdisciplinary model where the team performs joint assessment. However, given the current lack of diagnostic clinics in Australia, this model is unlikely to be feasible in the short term, and particularly in regional, rural and remote areas. Diagnostic services must be accessible and provided within a sustainable model. Thus diagnosis by a specialist medical practitioner with input from other relevant health professionals is acceptable in the absence of access to interdisciplinary diagnostic services, or where the specialist medical practitioner considers that an interdisciplinary assessment is not required. Interdisciplinary diagnostic clinics are needed in major cities and could provide outreach services to regional centres to conduct diagnostic clinics, support workforce training and build local capacity.

An implementation plan, informed by a health economic assessment, is required to embed the diagnostic instrument and resources in a comprehensive model of care and facilitate their national adoption. The implementation plan should also establish resources and support services for individuals undergoing diagnosis and their families, parents and carers.

Our findings support the need to:

1. Develop comprehensive Australian guidelines for the diagnosis of FASD.
2. Evaluate the validity and clinical applicability of the proposed diagnostic instrument in at least two interdisciplinary clinics to ensure appropriateness in the Australian context.
3. Develop a national implementation plan for dissemination and adoption of the diagnostic instrument. The instrument should be embedded in a sustainable model of care, and strategies for health professional training and support identified.

4. Establish interdisciplinary teams for diagnosis of FASD in major capital cities, and a plan for outreach to regional centres to conduct diagnostic clinics and build capacity and support services.
5. Establish clinical research programs to build a locally relevant evidence base for diagnosis, with initial priorities being to:
    i. establish the validity of available population reference data (and assessment instruments) for growth, head circumference and facial features in the Australian population, including among Indigenous Australians and those from culturally diverse backgrounds;
    ii. evaluate the diagnostic criteria for CNS abnormality, including the adequacy of a significant structural abnormality (such as microcephaly) in the absence of functional impairment, as an indicator of FASD diagnoses;
    iii. identify methods for assessing functional CNS abnormalities and establish their cultural appropriateness in the Australian population, including among Indigenous Australians and those from culturally diverse backgrounds; and
    iv. evaluate the evidence for inclusion of a diagnostic category for neurobehavioural disorders that requires significant CNS dysfunction in only two domains instead of three.

SCREENING

We found that health professionals desired a standardised, accurate and cost-effective screening instrument to identify individuals who have a high likelihood of FASD and who require further assessment to confirm their diagnosis. However, a screening instrument for FASD was not developed because of insufficient evidence for a simple, valid test and the absence of established diagnostic and management services to support the individuals undergoing screening and their families. Information collected during this project will inform future research to develop and evaluate targeted screening methods. Additional strategies to facilitate effective identification and diagnosis of individuals with FASD, including improving awareness and knowledge of FASD among health professionals, may be cost-effective and should be considered.

Our findings support the need to:

6. Conduct research to develop and evaluate the performance of a targeted screening instrument in high risk populations, including feasibility, sensitivity, specificity, predictive value and cost effectiveness.

7. Ensure that targeted screening for FASD is only implemented when:
    i. there is a sufficient evidence base to demonstrate the effectiveness of screening; and
    ii. adequate diagnostic and management services are established.
8. Evaluate additional strategies to improve the identification of individuals with FASD, including interventions to improve awareness among health professionals.

REFERRAL

We developed criteria to assist health professionals to make decisions about the need to refer individuals for a diagnostic evaluation following routine clinical evaluation. To support implementation of these agreed criteria for referral, brief guidelines and training resources for health professionals should be developed and evaluated. Performance evaluation of the standard assessment methods used to identify criteria for referral, and the effectiveness of the criteria for referral is also required.

Our findings support the need to:

9. Develop and evaluate guidelines and resources for training and support of health professionals, and identify strategies for implementation of the criteria for referral.

GENERAL CONCLUSIONS

Additional mechanisms are required to ensure that improvements in the identification and diagnosis of FASD in Australia benefit population health. National mandatory reporting of FASD diagnoses, using the standard diagnostic criteria we have developed should be introduced to improve the evidence base for prevention, diagnosis, and management of FASD in Australia. The outcomes of this project demonstrate the value of a collaborative approach to research involving health professionals, consumer and community members and researchers. Continued operation of the Australian FASD Collaboration is recommended to: develop comprehensive guidelines for the diagnosis of FASD in Australia; evaluate the Australian FASD Diagnostic Instrument; develop and evaluate strategies to improve the identification of individuals with FASD and inform policy and practice on the prevention, screening and diagnosis of FASD in Australia.

Our findings support the need to:

10. Establish national mandatory reporting of FASD diagnoses based on our diagnostic criteria for FAS, PFAS and ND-AE in Australia, and mandatory recording of alcohol use in pregnancy in the minimum data set for the national perinatal data collection.

11. Fund a national collaboration to develop comprehensive Australian guidelines for the diagnosis of FASD, evaluate the Australian FASD Diagnostic Instrument, develop and evaluate strategies to improve the identification of individuals with FASD, and inform policy and practice on the prevention, screening and diagnosis of FASD in Australia.

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10. APPENDIX A: AUSTRALIAN FASD DIAGNOSTIC INSTRUMENT

APPENDIX A1: GUIDE TO THE AUSTRALIAN FASD DIAGNOSTIC INSTRUMENT

APPENDIX A2: AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT FORM

APPENDIX A3: AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT SUMMARY FORM

APPENDIX A4: AUSTRALIAN FASD MANAGEMENT PLAN FORM

APPENDIX A5: INFORMATION FOR CLINICIANS ON ISSUES THAT PARENTS OR CARERS MAY EXPERIENCE DURING THE FASD DIAGNOSTIC ASSESSMENT

APPENDIX A6: INFORMATION ON FASD DIAGNOSTIC ASSESSMENT FOR PARENTS AND CARERS

APPENDIX A7: AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT CONSENT FORM

APPENDIX A8: AUSTRALIAN FASD CRITERIA FOR REFERRAL FOR DIAGNOSTIC ASSESSMENT

GUIDE TO THE AUSTRALIAN FASD DIAGNOSTIC INSTRUMENT

Purpose

The ***Australian Fetal Alcohol Spectrum Disorders (FASD) Diagnostic Instrument*** was developed to facilitate and standardise the identification of FASD in Australia. This guide provides clinicians with the background information needed to apply standard national diagnostic criteria for FASD in the Australian context. The recommended clinical diagnostic assessment methods are based on the University of Washington (UW) 4-Digit Diagnostic Code[1] method of interdisciplinary team assessment, and the Australian diagnostic categories and criteria combine elements of the UW and Canadian Guidelines for the diagnosis of FASD.[2] Although the actual 4-Digit Diagnostic Code does not need to be routinely derived during the diagnostic assessment, it can be derived from information recorded on the ***Australian FASD Diagnostic Assessment Form*** (Appendix A2).

The diagnosis of FASD is complex, and ideally requires an interdisciplinary team of clinicians to evaluate individuals for a range of potential outcomes that are associated with, but not unique to, prenatal alcohol exposure. In addition, alternative diagnoses must be excluded and the potential influence of other adverse exposures assessed. Diagnostic criteria used internationally are based on criteria developed in North America. Research is required to ensure that diagnostic criteria and methods recommended for use in Australia are validated in the Australian context, and can be updated to include new diagnostic technologies or diagnostic classifications when appropriate.

Historical context

In 1996 the United States Institute of Medicine (IOM) published diagnostic criteria for four disorders that were known to be associated with prenatal exposure to alcohol: Fetal Alcohol Syndrome (FAS), Partial FAS (PFAS), Alcohol-related Neurodevelopmental Disorders (ARND) and Alcohol-related Birth Defects (ARBD).[3] These four disorders are now collectively referred to using the non-diagnostic descriptive term Fetal Alcohol Spectrum Disorders (FASD). However, there is controversy about the reliability with which disorders within the FASD spectrum can be diagnosed and the terminology used to describe these disorders. For example, the United States Centers for Disease Control and Prevention (CDC) only provide criteria for the diagnosis of FAS,[4] while the Canadian guideline recommends diagnosis of FAS, PFAS and ARND.[2]

Australian diagnostic categories and criteria

Following a consultation involving Australian clinicians, consensus was reached on the use of the following FASD diagnostic categories in Australia:

i) Fetal Alcohol Syndrome (FAS)
ii) Partial Fetal Alcohol Syndrome (PFAS)
iii) Neurodevelopmental Disorder–Alcohol Exposed (ND-AE)

The diagnostic criteria for each of these categories are summarised in Table 1. The aetiological role of alcohol is most clearly established in the presence of all three characteristic FAS facial anomalies, where a diagnosis of FAS can be made when prenatal alcohol exposure is unknown,[5] provided there is also growth deficit and central nervous system abnormality. The aetiological role of alcohol is least well established for ND-AE. Because the current diagnostic assessment process is unable to causally link alcohol exposure to observed impairments, irrespective of the extent of the exposure, the Neurodevelopmental Disorder diagnostic category is designated 'Alcohol Exposed'. Nevertheless, research supports the validity of this diagnostic category within the spectrum of FASD.[5]

Table 1 Australian FASD diagnostic categories and criteria

Diagnostic criteria[#]	Diagnostic category		
	Fetal Alcohol Syndrome (FAS)	**Partial Fetal Alcohol Syndrome (PFAS)**	**Neurodevelopmental 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: • 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: • 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 ≤ 10th percentile adjusted for gestational age, or postnatal height or weight ≤ 10th percentile	No deficit required[*]	No deficit required[*]
Central Nervous System (CNS) abnormality	At least 1 of the following: • clinically significant structural abnormality (e.g. head circumference ≤ 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) [‡]		

[†]University of Washington Lip-Philtrum Guides: http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm

[*]Not required for diagnosis but may be present

[#]Appropriate reference charts should be used, and other causes of growth deficit and CNS abnormality excluded.

[‡]Assessment of dysfunction based on evidence from standard validated assessment instruments interpreted by qualified professionals

Diagnostic assessment

There is insufficient evidence to support the use of formal screening tests for FASD in the Australian population. However, the ***Australian FASD Criteria for Referral for Diagnostic Assessment*** (Appendix A8) is a resource that provides guidance for primary health care providers on the appropriateness of referral for a diagnostic assessment. Ideally, the FASD diagnostic process is conducted by a specialist interdisciplinary assessment team to enable accurate assessment of the range of outcomes that may be associated with prenatal alcohol exposure; however there are at present limited resources to support implementation of this diagnostic model in Australia.

The ***Australian FASD Diagnostic Assessment Form*** (Appendix A2) identifies the information required to diagnose FASD according to the Australian diagnostic criteria (Table 1). Use of this form is recommended following assessment of patient history and identification of a suspicion of FASD. A summary version of the FASD diagnostic assessment form which identifies the essential information required for diagnosis is also available (***Australian FASD Diagnostic Assessment Summary Form:*** Appendix A3). Although this instrument was developed for use among individuals with suspected FASD, it is essential that other causes and conditions that could explain the clinical presentation are considered and excluded during the diagnostic process. Information on the diagnostic assessment process should be provided (***Information on FASD Diagnostic Assessment for Parents and Carers*** Appendix A6) prior to the diagnostic assessment, and completion of the ***Australian FASD Diagnostic Assessment Consent Form*** (Appendix A5) is recommended.

In an individual with prenatal alcohol exposure and or suspicion of FASD the following essential areas should be assessed: growth, maternal alcohol use, facial and physical features, central nervous system (CNS) structure and function, and other exposures.

Growth

The purpose of the growth assessment is to identify whether there has been growth deficiency at any time that is characteristic of a teratogenic insult, and is not due to genetic or other environmental factors, including nutritional deprivation and acute or chronic illness. Growth deficiency due to prenatal alcohol exposure generally presents as a relatively consistent impairment in growth over time.[1]

Growth deficit is required for the diagnosis of FAS, and is defined as a height and or weight measurement at ≤10th percentile for age and sex at any time that is not attributable to any other known cause. Growth should be assessed and the data plotted on appropriate sex-specific growth reference charts by gestational age (at birth) or age to identify percentile ranks for birth length, birth weight, height and weight. Correction for prematurity should be used until 2 years of age.[6] Evaluation of growth deficiency in height or weight should be based on locally appropriate reference charts (e.g. CDC 2000[7]).

Due to the strong genetic contribution to attained height, adjust height for mid-parental height, when available, using measured rather than reported parental height where possible.[8 9] Target adult height in centimetres can be estimated using the method developed by Hermanussen and Cole[10] and recommended for use in Australia[11] based on the CDC growth charts as follows:

Target height (cm) for females = $163.3 + 2.33\left(\frac{Father's\ height - 176.9}{7.1} + \frac{Mother's\ height - 163.3}{6.5}\right)$

10^{th} percentile of target height for females (cm) = target height – 8.3

Target height (cm) for males = $176.9 + 2.57\left(\frac{Father's\ height - 176.9}{7.1} + \frac{Mother's\ height - 163.3}{6.5}\right)$

10^{th} percentile of target height for males (cm) = target height – 9.1

The above target height estimate can be used to evaluate whether projected (or actual) adult height is equal to or less than the 10^{th} percentile of the predicted target height:

1. Estimate target adult height based on mid-parental height, and calculate the 10^{th} percentile of target height
2. Evaluate current height using the appropriate age and gender-specific CDC growth chart. Identify the projected adult height by following the current height percentile line to 20 years[6]
3. Compare the 10^{th} percentile of estimated target height with projected adult height.

The above method for estimating target height provides a significant improvement over the frequently used Tanner method, which is based on adding 6.5cm to the mean of the mid-parental heights for males, or subtracting 6.5cm from this mean for females.[11]

Maternal alcohol use

The timing, frequency and quantity of prenatal alcohol exposure are linked to the pattern and severity of fetal outcomes, but may not be available or known with any accuracy.[2 5] In addition, both maternal and fetal characteristics are associated with variability in alcohol-related outcomes. Adverse cognitive, behavioural and developmental outcomes may result from exposure at any time during pregnancy and may occur in the absence of facial anomalies or structural central nervous system abnormalities.[12] Although there is no strong evidence associating low levels of prenatal alcohol exposure with risks to fetal development,[13] there is no agreed 'safe' level of alcohol exposure in pregnancy.[14]

Assessment of prenatal alcohol exposure requires careful evaluation of a range of information that may provide confirmation of maternal alcohol use and quantification of intake. Evidence of confirmed prenatal alcohol exposure may include: information reported by the birth mother about consumption during the index pregnancy; reports by others, including a relative, partner, household or community member about direct observation of drinking during the index pregnancy; or documentation in medical or other records of alcohol exposure, alcohol-related disorders, or other problems directly related to drinking during the index pregnancy, including alcohol-related injury and intoxication. A history of alcohol dependence without evidence of consumption during the index pregnancy is not sufficient to indicate confirmed exposure, but should raise suspicion.[1 2]

The required evidence for prenatal alcohol exposure for each diagnostic category is summarised in Table 1. When detailed information on maternal alcohol use is available, consumption during pregnancy should be assessed using the AUDIT-C questions,[15 16] as included on the ***Australian FASD Diagnostic Assessment Form*** (Appendix A2). The AUDIT-C questions provide a standardised method for the assessment of maternal alcohol use and are based on a validated sex-specific version of the instrument.[17 18] The use of a sex-specific threshold of 5 or more drinks on one occasion for question 3 of the AUDIT-C reflects known levels of maternal alcohol consumption associated with increased risk of FASD and other harms.[14 19 20] Derivation of the AUDIT-C score, although not essential for diagnosis, allows the clinician to categorise the level of risk associated with maternal drinking. This can indicate the potential risk to the

fetus. The AUDIT-C can be used as a screening test for the identification of maternal heavy drinking, alcohol abuse or dependence.[15-18] Information on the definition of a standard drink for different types of alcoholic drinks (e.g. http://www.alcohol.gov.au/internet/alcohol/publishing.nsf/Content/drinksguide-cnt) should be provided prior to using the AUDIT-C.

Evidence of exposure can be evaluated to estimate the overall level of risk using the following broad risk categories:

i. none (confirmed absence),
ii. unknown (alcohol use is unknown),
iii. confirmed (confirmed use, but exposure less than high risk level for FASD; or confirmed use, but not known if exposed at a high risk level for FASD), and
iv. confirmed-high risk (confirmed use, exposure at high risk level for FASD).

Confirmed high risk exposures for FASD can be considered to include, at any time during pregnancy:

i. reported consumption of 7 or more standard drinks per week (derived from AUDIT-C questions 1 and 2), or
ii. reported consumption of 5 or more standard drinks on one occasion (AUDIT-C question 3), or
iii. an AUDIT-C score of 5 or more.

The identification of high risk alcohol exposure for FASD should be guided by Australian and international research,[13 20-24] and updated as required.

Other reliable information on prenatal alcohol exposure includes medical or legal documentation, such as recorded blood alcohol levels or medical diagnosis, forensic records, or observation by a public officer of alcohol consumption during pregnancy. If recalled information from different informants is in direct conflict (confirmed absence and confirmed presence) and reliable information on exposure is not available, alcohol exposure should be recorded as unknown. A diagnosis of FASD is not appropriate where there is confirmed absence of prenatal alcohol exposure, but may be appropriate where there is unknown prenatal alcohol exposure (Table 1). Information such as when the birth mother realised she was pregnant, may influence the evaluation of reported information on prenatal alcohol exposure.

Facial features and physical examination

The facial anomalies required for a diagnosis of FAS or PFAS are shown in Table 1. Either two or three anomalies must be present at the same point in time. All three characteristic FAS facial features should be assessed using either direct measurement, or computerised analysis of a digital facial photograph. Computerised analysis of a facial photograph taken using a standard digital camera, as described by Astley and Clarren[25] is useful.[1]

Direct measurement of palpebral fissure length and assessment of upper lip thinness and philtrum smoothness should be performed using the techniques and tools described in the UW 4-Digit Diagnostic Code,[1] as summarised below. For palpebral fissure lengths, a clear plastic ruler should be used to measure the distance from the endocanthion to the exocanthion when the eye is fully open. Care should be taken to ensure that the ruler is held as close as possible to the patient's eye without touching the eye or eyelashes. Measurements should be evaluated using an appropriate reference chart (e.g. Clarren et al.[26 27]).

Upper lip thinness and philtrum smoothness should be assessed using the appropriate UW Lip-Philtrum Guide (1. Caucasian or 2. African American), based on the patient's racial background. The UW Lip-Philtrum Guides include representative photographs of lip and philtrum combinations according to a 5-point Likert scale, with ranks 1-3 within normal limits and ranks 4 and 5 outside normal limits. Lip-Philtrum Guides specific to the Australian and Australian Aboriginal populations have not yet been

developed. Lip-Philtrum Guides specific to every racial group are unlikely to be required due to the lack of a homogenous phenotype for many races, the frequency of multiracial ancestry, and the small magnitude of differences involved.[5] Laminated UW Lip-Philtrum Guides are available from http://depts.washington.edu/fasdpn.

Assessment of the upper lip and philtrum must be performed with the lips gently closed, and with no smile. The UW Lip-Philtrum Guide should be held next to the patient's face, and the physician's eyes must be in the same plane as the patient's external auditory meatus and inferior border of the bony orbital rim to obtain a standard assessment of upper lip thinness.

Summary ranking of facial features is based on how many of the three facial features assessed were found to be present at the specified thresholds summarised below:

i. small palpebral fissure length (2 or more standard deviations below the mean),
ii. thin upper lip (rank 4 or 5 on the Lip-Philtrum Guide), and
iii. smooth philtrum (rank 4 or 5 on the Lip-Philtrum Guide).

The three characteristic FAS facial features must be assessed at the same age. If multiple occasions of assessment are available, select the time when the features are collectively most strongly expressed.[1]

Other dysmorphic features

Individual dysmorphic features can occur in multiple syndromes, and examination for features that differentiate alternate or co-existing syndromes and other disorders during the diagnostic assessment is essential. Other dysmorphic features consistent with alcohol exposure or other syndromes provide critical information for differential diagnosis and should be assessed during the clinical examination. Differential diagnosis should include consideration of conditions that have a clinical presentation that is similar to FAS. If a genetic disorder is suspected, or any uncertainty regarding differential diagnosis exists, review by a clinical geneticist is indicated.

Central Nervous System

Prenatal exposure to alcohol can have a range of effects on the developing brain and nervous system. Abnormalities associated with brain damage due to prenatal alcohol exposure can include structural, neurological and functional abnormalities. Functional abnormalities observed in individuals with prenatal alcohol exposure are not specific to that exposure and may be due to other causes. More severe or global CNS dysfunction is more likely to indicate underlying brain damage, but does not provide definite evidence of brain damage.[1] The diagnostic examination should assess for significant CNS structural abnormalities, hard neurological signs and significant dysfunction using methods described in the UW 4-Digit Diagnostic Code,[1] as summarised below.

Structural and neurological abnormalities

Evidence of severe brain damage, including clinically significant structural or neurological abnormalities, should be confirmed by physicians. Structural abnormalities may include microcephaly, defined as occipitofrontal circumference (OFC) of 2 or more standard deviations below the mean, or clinically significant structural abnormalities detected by brain imaging.[1] Magnetic resonance imaging (MRI) examination is recommended only when clinically indicated (for example, by the presence of non-familial microcephaly, localising neurological signs, a focal seizure disorder or macrocephaly). It is important to consider racial background when assessing OFC and to exclude microcephaly of familial origin.

Microcephaly due to other causes (e.g. chromosomal abnormalities, infection or exposure to other teratogens) should also be excluded.

Clinically significant neurological abnormalities include severe seizure disorders of presumed prenatal origin, or other significant hard neurological signs. Clear evidence of significant structural or neurological CNS abnormality does not eliminate the need for standard age-appropriate psychometric or developmental assessment.

Functional abnormalities

Functional CNS impairment is evaluated using a variety of standardised assessment methods that are appropriate for the age and cultural and linguistic background of the individual. Among patients who have been exposed to potentially harmful levels of alcohol, evidence of significant CNS dysfunction across multiple domains of function can provide evidence of problems that are likely to be due to underlying brain damage rather than due to adverse postnatal experiences.[1]

Information relevant to the assessment of CNS dysfunction in children should be obtained during an interview with the caregiver. Relevant information that should be sought includes the problems that led to referral, caregivers' assessments of the patient's main strengths and weaknesses, social and medical history, and caregivers' assessment of age-appropriate functional abilities. The assessing professional should seek information relevant to their assessment (e.g. planning, behavioural regulation, judgement, memory and learning, spatial skills, social skills and motor control).[1]

Evidence suggests that individuals with FASD may show impairment in the following areas:[12 28 29]

1. Neurocognitive function (intellectual ability, learning, memory, visual spatial reasoning, number processing)
2. Self-regulation (executive function, attention and arousal)
3. Adaptive function (motor function, sensory integration, social skills/social communication, independent living)

Evidence of significant CNS dysfunction must be based on findings from standardised validated psychometric assessments that are administered to the individual or based on information obtained from reliable informants and interpreted by qualified professionals (e.g. psychologists, psychiatrists, speech pathologists).[1] Evidence of significant CNS dysfunction requires identification of significant impairment in three or more different domains of functioning. Significant impairment is generally defined as performance that is 2 or more standard deviations below the mean on a standardised test, based on interpretation by a qualified professional.[1] Significant impairment in global functioning (which by definition includes impairment in multiple domains of functioning) also fulfils the criteria for CNS abnormality. This may be relevant in a young child in whom individual domains cannot be tested.

Review of psychometric test results by qualified and experienced professionals in the context of the characteristics of the tests used, the individual being examined, and the broader assessment process is critical to making an accurate diagnosis which considers quantitative and qualitative aspects of performance. Clinician judgement is crucial both in the selection of appropriate valid and reliable tests that examine domains that may be affected by prenatal alcohol exposure, and in the interpretation of these findings. This includes the use of judgement where there is overlap between domains, where test scores reflect deficits in multiple domains of functioning, or where test scores are borderline or indicate discrepancies in performance between subdomains. Identification of clinically significant dysfunction that is likely to be due to underlying brain damage rather than adverse postnatal experiences should be

based on evidence generated by standardised valid psychometric assessments and the judgement of experienced clinicians ideally within an interdisciplinary team.[1 2]

Although a broad range of functional domains are identified on the diagnostic form (cognition, memory, language, executive function, attention and activity level, adaptive behaviour, social skills/social communication, and motor function), the diagnostic process does not require assessment of all domains. Clinicians may select specific domains for assessment based on the specific needs of each individual. Ideally a comprehensive evaluation will provide information relevant to diagnosis, management and planning, and include identified strengths and needs. The neuropsychological assessment process should identify other psychiatric diagnoses where appropriate, and contribute to the differential diagnosis.

Comprehensive functional CNS examination is generally unable to be performed with children aged less than 5 years. Developmental assessments (e.g. the Bayley Scales of Infant Development) are not usually used as a source of evidence to support a classification of significant CNS dysfunction because developmental delay is not always predictive of brain dysfunction. However, evidence of severe global developmental delay in young children is highly predictive of significant brain dysfunction, and may be used as evidence of CNS dysfunction.[1] The behavioural diagnoses of Autism Spectrum Disorder and Attention Deficit Hyperactivity Disorder (ADHD) made based on standardised Diagnostic and Statistical Manual of Mental Disorders (DSM) or International Classification of Diseases (ICD) criteria are also considered to provide evidence of significant impairment in the domains of social skills/social communication (ASD) and attention (ADHD).

Other exposures

Impairment observed among individuals with FASD, including CNS dysfunction, may be associated with exposures other than alcohol. It is important to determine whether any observed impairments can be explained by other causes or events (e.g. prenatal complications, genetic factors, head injuries, early life trauma, problems with vision or hearing, or substance abuse by the patient). There may not be a singular explanation for the observed CNS dysfunction, and it is important that the diagnostic process consider the effects of other adverse prenatal and postnatal exposures.[1]

All relevant prenatal and postnatal exposures or events should be documented during the diagnostic assessment, and evaluated based on their likely severity of influence. Other exposures should be considered when determining the appropriate diagnosis and intervention plan.

Diagnosis and management plan

The ***Australian FASD Diagnostic Assessment Summary Form*** (Appendix A3) includes a summary of the diagnostic criteria for FASD, and can be used to summarise assessment findings and identify if a FASD diagnosis is appropriate. Information collected during the diagnostic assessment should be reviewed, ideally in an interdisciplinary context, to evaluate the strength of evidence to support a diagnosis of FAS, PFAS or ND-AE, to exclude other causes or conditions, and to assess the potential influence of other exposures and events. For individuals diagnosed with a FASD, a management plan which documents major findings, management needs and specific recommendations is required (***Australian FASD Management Plan Form:*** Appendix A4). Ideally the management plan is prepared by the interdisciplinary assessment team.

Resources

University of Washington 4-Digit Diagnostic Code, Lip-Philtrum Guide and palpebral fissure length charts (http://depts.washington.edu/fasdpn/htmls/diagnostic-tools.htm)
Canadian Guidelines for the diagnosis of FASD (www.cmaj.ca/content/172/5_suppl/S1.full.pdf+html)

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AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT FORM

PATIENT DETAILS

Name			
Sex	☐ female	☐ male	☐ other
Date of birth (DD/MM/YYYY)	/ /	Age at assessment:	
Racial background (select 1 or more)	☐ Aboriginal ☐ Caucasian ☐ Other (specify)	☐ African ☐ Pacific Islander	☐ Asian ☐ Torres Strait Islander
Name of person(s) accompanying patient			
Relationship to the patient			
Consent form completed	☐ no	☐ yes (attach)	

Form completed by	
Place of assessment	
Assessment date (DD/MM/YYYY)	/ /

GROWTH

Assess birth parameters and postnatal growth, and determine if any deficit ≤10th percentile exists that is unexplained by genetic potential, environmental influences (e.g. nutritional deficiency) or other known conditions (e.g. chronic illness).

Birth	Gestational age	Birth length		Birth weight	
Date	weeks	cm	percentile	grams	percentile

Growth reference chart used:

Postnatal		Height		Weight	
Date	Age	cm	percentile	kg	percentile

Growth reference chart used:

Parental height (if available)

Mother's height (cm)	Father's height (cm)	Sex-specific target height (cm)	Sex-specific target height (percentile)

Specify factors that may explain growth parameters: Are growth parameters explained by other known conditions? ☐ no ☐ yes

Growth summary

Was an unexplained deficit in heath or weight identified at any time? ☐ no ☐ height or weight ≤10th and >3rd percentile ☐ height or weight ≤3rd percentile

MATERNAL ALCOHOL USE

Assess evidence of maternal alcohol use prior to and during pregnancy. The definition of a standard drink should be explained prior to administering the AUDIT-C (Q1-3).

Reported alcohol use (if available)

Source of reported information on alcohol use: ☐ birth mother ☐ other (specify)

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

never [skip Q2+Q3]	monthly or less	2-4 times a month	2-3 times a week	4 or more times a week
☐0	☐1	☐2	☐3	☐4

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

1 or 2	3 or 4	5 or 6	7 to 9	10 or more
☐0	☐1	☐2	☐3	☐4

3. How often did the birth mother have 5 or more standard drinks on one occasion during this pregnancy?

never	less than monthly	monthly	weekly	daily or almost daily
☐0	☐1	☐2	☐3	☐4

AUDIT-C score during this pregnancy: (Q1+Q2+Q3)=

Alcohol use in early pregnancy: When did the birth mother realise that she was pregnant? (weeks)

Did the birth mother modify her drinking behaviour on confirmation of pregnancy? (specify - AUDIT-C may be used)

During which trimesters was alcohol consumed? ☐ 1st ☐ 2nd ☐ 3rd ☐ unknown ☐ none

Was consumption of 7 or more standard drinks per week reported at any time during pregnancy? ☐ no ☐ yes

Other evidence of exposure

Is there evidence that the birth mother has ever had a problem associated with alcohol?

☐ no ☐ yes (identify below, including source of information)

☐ alcohol dependency (specify)

☐ alcohol-related illness or hospitalisation (specify)

☐ alcohol-related injury (specify)

☐ alcohol-related offence (specify)

☐ other (specify)

Other information on alcohol exposure (e.g. medical records, court reports, other information on consumption patterns):
(specify evidence of exposure and indicate source of information)

Alcohol exposure summary

Prenatal alcohol exposure:
☐ none ☐ unknown ☐ confirmed ☐ confirmed-high risk

Reliability of information on alcohol exposure: ☐ unknown ☐ low ☐ high

AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT FORM

FACIAL FEATURES AND PHYSICAL EXAMINATION

Assess for the 3 characteristic facial features of Fetal Alcohol Syndrome: short palpebral fissure length (2 SD or more below the mean), smooth philtrum (rank 4 or 5 on the Lip-Philtrum guide), and thin upper lip (rank 4 or 5 on the Lip-Philtrum guide).

Palpebral Fissure Length (PFL)			Right PFL		Left PFL		Mean PFL	
Date	Age	Assessment method	mm	SD	mm	SD	mm	SD
		☐ direct measure ☐ photo analysis						
		☐ direct measure ☐ photo analysis						
		☐ direct measure ☐ photo analysis						

PFL reference chart used:

Philtrum

Date	Age	Assessment method	UW Lip-Philtrum Guide 5-point rank
		☐ direct measure ☐ photo analysis	
		☐ direct measure ☐ photo analysis	
		☐ direct measure ☐ photo analysis	

Upper lip

Date	Age	Assessment method	UW Lip-Philtrum Guide 5-point rank
		☐ direct measure ☐ photo analysis	
		☐ direct measure ☐ photo analysis	
		☐ direct measure ☐ photo analysis	

Lip-Philtrum Guide[†] used: ☐ 1. Caucasian ☐ 2. African American

Other dysmorphic features

☐ midface hypoplasia
☐ flat (low) nasal bridge
☐ anteverted nares
☐ cleft lip/palate
☐ long philtrum
☐ other (specify)
☐ ptosis
☐ epicanthal folds (non-racial)
☐ micrognathia
☐ relative prognathism
☐ railroad track ears
☐ hockeystick palmar crease
☐ camptodactyly
☐ clinodactyly of fifth digits

Other physical findings, syndromes or medical conditions

Hearing impairment: ☐ no ☐ yes (specify)
Vision impairment: ☐ no ☐ yes (specify)
Other findings:

Facial features summary

Number of characteristic FAS facial features (PFL 2 SD or more below the mean, philtrum rank 4 or 5, upper lip rank 4 or 5):
☐ 0 ☐ 1 ☐ 2 ☐ 3

[†]University of Washington Lip-Philtrum Guides: http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm

CENTRAL NERVOUS SYSTEM (CNS)

Assess for clinically significant structural or neurological abnormalities; or evidence of significant dysfunction on standardised psychometric assessment in 3 or more domains of function (impairment 2 SD or more below the mean).

STRUCTURAL CNS

Occipitofrontal Circumference (OFC)

Date	Age	OFC (cm)	Percentile*	Reference used
birth:				

*correct for gestational age when < 2 years old

Imaging

CNS imaging performed: ☐ no ☐ yes (specify image modality and date)

Specify any structural abnormalities observed:

Structural CNS summary

Evidence of clinically significant structural abnormalities?

☐ no ☐ OFC ≤3rd percentile ☐ other evidence of significant structural abnormalities

NEUROLOGICAL CNS

Assess evidence of seizure disorders or other abnormal hard neurological signs.

Seizure disorder findings

Seizure disorder present: ☐ no ☐ yes (specify)

If yes, is the seizure disorder explained by prior injury, infection, or metabolic or other disease?

☐ no ☐ yes

Other hard neurological signs

Other abnormal hard neurological signs present: ☐ no ☐ yes (specify)

If yes, are these signs explained by prior injury, infection, or metabolic or other disease?

☐ no ☐ yes (specify)

Neurological CNS summary

Evidence of clinically significant hard neurological signs of presumed prenatal origin that are unexplained by other causes?

☐ no ☐ yes

FUNCTIONAL CNS

Reported functional problems

Functional concerns identified: (e.g. by parent, caregiver, teacher, at work)

Mental health, behavioural or psychiatric diagnoses

FUNCTIONAL DOMAIN SUMMARIES – STANDARDISED ASSESSMENTS

Assess evidence of significant CNS dysfunction due to underlying brain damage. Required evidence includes significant impairment (2 SD or more below the mean) in 3 or more different domains of brain function based on standardised psychometric assessment by a qualified professional.

Cognition (or Global Development if < 5 years of age)

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Cognitive impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Memory

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Memory impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Language (expressive and receptive)

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Language impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Executive function

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Executive function impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Attention / activity level / sensory processing

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Attention impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Adaptive behaviour / social skills / social communication

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Adaptive impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Academic achievement

Test/subtest name	Date	Score	SD	Interpretation
Other information:				
Academic impairment: ☐ none ☐ some ☐ significant ☐ not assessed				

Motor

Test/subtest name	Date	Score	SD	Interpretation

Other information:

Motor impairment: ☐ none ☐ some ☐ significant ☐ not assessed

CNS summary

Evidence of clinically significant structural or neurological abnormality: ☐ no ☐ yes

Number of functional domains with evidence of significant impairment: ☐ none ☐ 1 ☐ 2 ☐ 3 or more

OTHER EXPOSURES

Assess evidence of adverse prenatal and postnatal exposures and events that may explain observed abnormalities.

Prenatal

Other prenatal exposures identified: (if yes, specify and indicate source of information)

☐ cigarettes (specify)

☐ marijuana (specify)

☐ heroin (specify)

☐ cocaine (specify)

☐ amphetamines (specify)

☐ other (specify)

Specify other risk factors and assess risk: (e.g. pregnancy complications, congenital infection, trauma, exposure to know teratogens, paternal or maternal intellectual impairment, maternal ill-health)

Other prenatal risk summary: ☐ no known risk ☐ unknown risk ☐ some risk ☐ high risk

Postnatal (e.g. history of abuse or neglect, serious head injury, other medical conditions that lead to brain damage, substance abuse)

Specify other physical or medical risk factors and assess risk:
Specify other psychosocial risk factors and assess risk:
Postnatal risk summary: ☐ no known risk ☐ unknown risk ☐ some risk ☐ high risk

DIAGNOSIS

Please refer to the Australian FASD Diagnostic Criteria Summary for derivation of the Australian FASD diagnostic categories. Record the diagnosis below.

☐ Fetal Alcohol Syndrome (FAS)
☐ Partial Fetal Alcohol Syndrome (PFAS)
☐ Neurodevelopmental Disorder–Alcohol Exposed (ND-AE)
☐ Other (specify)

AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT SUMMARY FORM

Patient name				Assessment date	
Date of birth		Age		Gestational age		Assessment clinic	
Racial background				Sex		Form completed by	

MATERNAL ALCOHOL USE

Prenatal alcohol exposure: ❑ none ❑ unknown ❑ confirmed
Trimesters exposed: ❑ none ❑ 1st ❑ 2nd ❑ 3rd
Reported 7+ drinks per week: ❑ no ❑ yes
Reported 5+ drinks/occasion: ❑ no ❑ yes
AUDIT-C score:
Other information:

Exposure:
❑ none
❑ unknown
❑ confirmed
❑ confirmed-high risk

GROWTH

	Height		Weight	
Date	cm	percentile	gm or kg	percentile

Growth deficit percentile rank:
❑ no deficit
❑ height or weight ≤ 10th and > 3rd
❑ height or weight ≤ 3rd

FACIAL FEATURES AND PHYSICAL EXAMINATION

	Right PFL		Left PFL		Mean PFL		Philtrum	Upper lip
Date	mm	SD	mm	SD	mm	SD	Rank	Rank

Lip-Philtrum Guide† used: ❑ 1. Caucasian ❑ 2. African American

Other dysmorphic features and abnormalities:

Facial feature summary:
❑ PFL 2 SD or more below the mean
❑ philtrum rank 4 or 5
❑ upper lip rank 4 or 5
Number of FAS facial features:
❑ 0 ❑ 1 ❑ 2 ❑ 3

CENTRAL NERVOUS SYSTEM - Structural and neurological

❑ microcephaly - OFC (cm):	percentile:	date measured:
❑ abnormal brain structure (specify)		
❑ seizure disorder (specify)		
❑ abnormal hard neurological signs (specify)		

CNS evidence summary:
❑ <3 domains significant
❑ 3+ domains significant
❑ structural/neurological

Functional

Domain, test name and findings	Score/SD	Date
1.		
2.		
3.		
4.		
5.		

OTHER PRENATAL AND POSTNATAL EXPOSURES (physical, psychosocial, environmental)

DIAGNOSIS

❑ Fetal Alcohol Syndrome (FAS)
❑ Partial Fetal Alcohol Syndrome (PFAS)
❑ Neurodevelopmental Disorder–Alcohol Exposed (ND-AE)
❑ Other (specify)

† University of Washington Lip-Philtrum Guides: http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm

AUSTRALIAN FASD DIAGNOSTIC CATEGORIES AND CRITERIA

Diagnostic criteria[#]	Diagnostic category		
	Fetal Alcohol Syndrome (FAS)	**Partial Fetal Alcohol Syndrome (PFAS)**	**Neurodevelopmental 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: • 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: • 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 ≤ 10th percentile adjusted for gestational age, or postnatal height or weight ≤ 10th percentile	No deficit required[*]	No deficit required[*]
Central Nervous System (CNS) abnormality	At least 1 of the following: • clinically significant structural abnormality (e.g. head circumference ≤ 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) [‡]		

[†]University of Washington Lip-Philtrum Guides: http://depts.washington.edu/fasdpn/htmls/lip-philtrum-guides.htm
[*]Not required for diagnosis but may be present
[#]Appropriate reference charts should be used, and other causes of growth deficit and CNS abnormality excluded
[‡]Assessment of dysfunction based on evidence from standard validated assessments instruments interpreted by qualified professionals

AUSTRALIAN FASD MANAGEMENT PLAN FORM

Assessment details Patient name	
Patient date of birth (DD/MM/YYYY)	/ / Age at assessment:
Date of assessment (DD/MM/YYYY)	/ /
Form completed by	

Diagnosis

Management needs	**Specific recommendations** (including person/agency responsible and time frame)

Progress review of management plan (DD/MM/YYYY): / /

AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT CONSENT FORM

Name of person undergoing diagnostic assessment	
Date of birth (Day/Month/Year)	/ /

Please tick the appropriate box

☐ I am legally responsible for myself

OR

I am legally responsible for the person named above and have the authority to consent to the diagnostic assessment because:

☐ I am his/her PARENT

☐ I am his/her LEGAL GUARDIAN

☐ I have read and understood the *Information for Patients and Parents/Carers on FASD Diagnostic Assessment*. Any questions I have asked have been answered to my satisfaction.

I, _____consent to this diagnostic assessment
Give Names Surname

Signature: _____

Date: _____ (Day/Month/Year)

I, _____have explained to the signatory above
Doctors full name

who stated that he/she understood the diagnostic assessment process and gave informed consent.

Signature: _____

Date: _____ (Day/Month/Year)

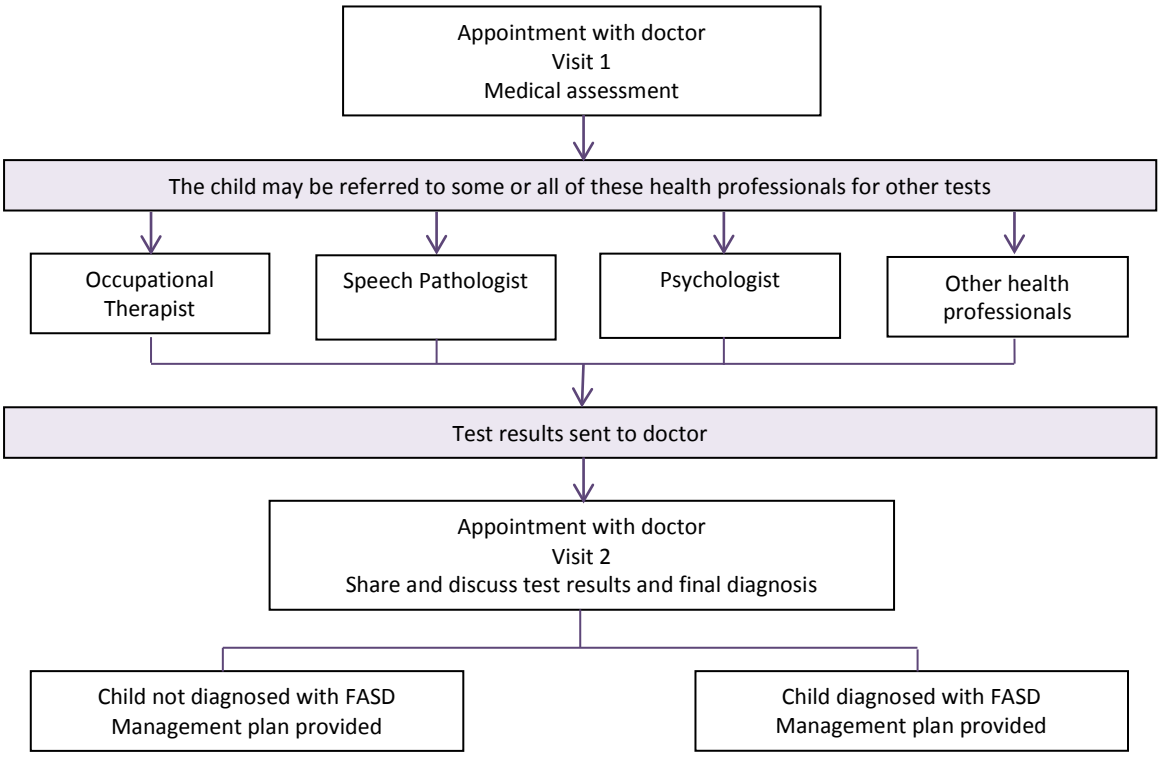
A copy of the signed consent form to be given to the signatory

INFORMATION ON FASD DIAGNOSTIC ASSESSMENT FOR PARENTS AND CARERS

Who is this information for?

Diagnostic assessment for Fetal Alcohol Spectrum Disorder (FASD) can be conducted with people of all ages. However diagnostic assessment is most commonly used with children under the age of 18 years. Ideally diagnostic assessment should occur as early as possible. The information in this document is for parents and carers. In this document the word 'child' refers to a person under the age of 18. However, the information could also be used to explain the FASD Diagnostic Assessment to a person of any age undergoing diagnostic assessment.

What is involved in getting a diagnosis?



What documents do I need?

At the beginning, the doctor will need to record some information about your child. As a parent or carer you may be asked to complete a form before you come to the appointment or to bring the information with you to the appointment. The following is a list of the type of information you may be asked to bring. You may not have all of this information but bring as much as you can.

- Birth records – date of birth, weight, length
- Medical history such as illnesses, surgery, vision or hearing problems
- Child health records – history of growth, weight, height
- School reports and any issues that have been raised by teachers or the school
- Photos of the child where you can see their face at different ages

Where does the diagnostic assessment happen and how long will it take?

At the first appointment the doctor will complete a medical assessment which will take about one hour. This will include testing hearing and vision, measuring height and weight and reviewing the documents you have brought to the appointment. During your appointment tell the doctor about the child's strengths and weaknesses, behaviour, any memory problems and how they relate to other people. Depending on the age of the child, let them talk about their own experiences. The doctor may take a photo of the child's face or look at the face and take measurements.

Your child may be referred to other health professionals who are skilled in doing different assessments. Make sure you have clear instructions on where each appointment is, the time of each appointment and what to do after all the assessments have been completed.

Occupational Therapist

The occupational therapist will assess motor skills (such as walking, running, tying shoe laces), sensory processing (how we receive, organise and understand visual and auditory messages) and visual perceptual skills (making sense of what we see). For a young child this may involve doing things with their hands, like drawing, writing letters, matching shapes, cutting with scissors, threading beads, asking about the things they like or don't like to play with because of the way they feel, taste, move or sound. This assessment may take an hour.

Speech Pathologist

A speech pathologist will assess understanding of language, use of language, verbal reasoning and use of speech sounds. For a young child this will involve talking with them and showing some pictures or toys, finding how many words they know, how well they can talk about things and how well they can understand words and questions. This assessment may take an hour.

Psychologist

The psychological assessment involves tests of memory, problem solving skills, academic abilities and cognitive abilities (how we think, remember and learn). To assess a child, a psychologist, who has had special training in how children learn and how the brain works, will assess what your child knows and test their memory and understanding. This will involve answering questions, and for a young child working with puzzles and blocks and doing some writing activities. This assessment may take 2 hours.

Other health professionals

A range of other health professionals could be consulted for their expertise, for example a geneticist or radiologist.

How much does the assessment cost?

Depending on your personal circumstances the cost will vary. In a public system the cost of each assessment may be covered but you will need to ask if there are any extra expenses. If you have a diagnostic assessment in the private system you will need to ask the clinic or doctor's practice about the cost of all the assessments and how much is covered by Medicare. If you have private health insurance contact them to find out how much you will be able to claim.

What happens after all the assessments?

Your child will be given another appointment with the doctor. You may like to ask a support person, friend or relative to accompany you to this appointment. The doctor will share and discuss the medical assessment and test results and final diagnosis which may be Fetal Alcohol Spectrum Disorder or any other diagnosis. You or your support person should ask questions and request a copy of the findings and diagnosis. Discuss with the doctor what the 'next steps' are and plan where to go for treatment and services. Also ask if you can phone the doctor with questions once you have had time to read the information and discuss the diagnosis with members of your family.

If you would like to talk to someone before, during or after the diagnostic assessment the National Organisation for Fetal Alcohol Syndrome and Related Disorders and the Russell Family Fetal Alcohol Disorders Association are Australian support groups that provide information, advocacy and support for families caring for people who have or are suspected of having Fetal Alcohol Spectrum Disorder.

Support Groups

- National Organisation for Fetal Alcohol Syndrome and Related Disorders (NOFASARD) http://www.nofasard.org/ or phone 1300 306 238
- Russell Family Fetal Alcohol Disorders Association (rffada) http://rffada.org/ or phone 1800 733 232
- If you are a foster carer you can also contact the foster care association in your state or territory

Why is diagnosis important?

To get to know the child better

A diagnostic assessment looks at all the things a child is good at and where they need help. It gives health professionals, parents, carers, family members, teachers and the child a better understanding of how to manage and or care for the child.

To access services that can help the child

A diagnosis may help you access services in the community that best meet the child's needs.

To answer your questions

A diagnostic assessment helps you understand more about the child. If you are wondering why the child has challenges in some areas of their life (for example, school, behaviour, memory) the diagnosis will help answer your questions.

To improve the quality of life

A diagnosis and management plan can contribute to positive long term outcomes for the child and their family.

Parents have said getting a diagnosis:

- was the catalyst that opened the door to meeting their child's needs
- brought relief and provided a reason for their child's difficulties
- removed the blame from them and the child and that alcohol's effect in pregnancy was to blame for the child's behaviour difficulties
- enabled them to find out more specific information about the disability
- gave them the knowledge they needed to be stronger advocates
- helped them understand that the child had brain differences and the child's behaviours were "normal" for them
- paved the way for trying different parenting approaches and to see the child as one who maybe "can't do" rather than one who "won't do"
- enabled them to change goals and set realistic expectations for the child

Children and young people and getting a diagnosis:

- "… I am the same person but have more of an idea why I do the things I do. My parents understand me better now."
- "… our past does not dictate our future."

Informed consent

Explanation of consent for the diagnostic assessment

- Informed consent is recommended in order for the diagnostic assessment to be completed.
- Consent can be withdrawn at any time.
- Informed consent can be withdrawn either verbally or in writing.
- Any information gathered before, during and after the diagnostic assessment will be treated as confidential.
- Information from the diagnostic assessment will only be shared with health professionals, and you as the child's parents or carers.
- Copies of any reports from the completed diagnostic assessment will be available to you.

Consent after the diagnostic assessment

- The recommendations from the diagnostic assessment should be implemented as appropriate between the child who has undergone the diagnostic assessment, their family and health professionals.
- For a child who is attending school you may be asked to give consent to sharing the diagnostic assessment results with people within the education system to enable the school to develop an appropriate plan for the child. This may include the teacher, principal, school psychologist and support services within the education department.

You will be provided with a copy of the Australian FASD Diagnostic Assessment Consent Form to review.

Information about Fetal Alcohol Spectrum Disorders

Information about Fetal Alcohol Spectrum Disorder is available on the following websites. There are many other websites that are not listed in this information sheet.

Australian websites

- National Organisation for Fetal Alcohol Syndrome and Related Disorders (NOFASARD) http://www.nofasard.org/ or phone 1300 306 238
- Russell Family Fetal Alcohol Disorders Association (rffada) http://rffada.org/ or phone 1800 rffada
- Living with Fetal Alcohol Spectrum Disorder – A Guide for Parents and Caregivers Drug Education Network http://beta.den2.handbuiltcreative.com/wp-content/uploads/2011/05/Living-with-FASD-A-Guide-for-Parents-and-Caregivers-VR-4-2011-2.pdf
- Telethon Institute for Child Health Research: Alcohol, Pregnancy and FASD http://alcoholpregnancy.childhealthresearch.org.au/
- Fetal Alcohol Spectrum Disorder Booklet: Information for People Working with Children and Families, Government of Western Australia, Department for Communities http://www.communities.wa.gov.au/parents/parentingresources/Documents/Foetal_Alcohol_Spectrum_Disorder_FASD_Booklet.pdf

International websites

- Asante Centre http://www.asantecentre.org/
- Finding Hope http://findinghope.knowledge.ca/home.html
- Parenting Children affected by Fetal Alcohol Syndrome: A Guide for Daily Living http://www.fasaware.co.uk/education_docs/daily_guide_for_living.pdf
- Strategies parents find helpful in raising their children with FASD http://come-over.to/FAS/PDF/TorontoStrategiesParents.pdf
- FASD Strategies not Solutions http://www.faslink.org/strategies_not_solutions.pdf
- Parenting Guidelines for Families of Children with Fetal Alcohol Syndrome/Fetal Alcohol Effects http://www.von.ca/fasd/
- Young adults share their experiences living with FASD (Conference Presentation 26 October 2011) http://www.youtube.com/watch?v=KBaAS2CNxbA

INFORMATION FOR CLINICIANS: ISSUES THAT PATIENTS AND THEIR PARENTS OR CARERS MAY EXPERIENCE DURING THE FASD DIAGNOSTIC ASSESSMENT

The effects of alcohol on the fetus are not widely known. While there are many reasons why people use alcohol, overwhelmingly the majority of birth mothers do not intentionally seek to harm their children. It is important that any language used by clinicians explains that any harm is caused by alcohol rather than the mother's behaviour and avoids *blaming the mother*. The more appropriate language to explain Fetal Alcohol Spectrum Disorder (FASD) is *"when alcohol was consumed during pregnancy"* or *"when the fetus is exposed to alcohol during pregnancy"*. It is important to offer non-judgemental support and advice. An early diagnosis and well-structured management and treatment plans can greatly improve the health outcomes and life of children with FASD and their families.

Respect is paramount to successful treatment. It is a vital tool in the elimination of discrimination and stigma and is pivotal to creating an environment where the issue of prenatal alcohol exposure can be discussed.

Adopt a consulting style that enables the person and their parents/carers to participate as partners in all decisions about their healthcare and take fully into account their race, culture, and any specific needs. People with FASD should have a comprehensive care plan that is agreed between them and their parents or carers, and their care providers.

The strategy for treatment should be individualised according to the degree of severity within the syndrome; other medications and comorbidity; the lifestyle and preferences of the family and/or carers.

Listen to the concerns raised by the parents or carers

Many people will have tried numerous avenues to obtain a diagnosis. For the person and their parents or carers this may result in them feeling frustrated, disempowered and not being believed. They may have also experienced health professionals as unwilling or not confident to raise the issue of fetal alcohol exposure as a possible cause. The person and their parents/carers may experience grief, loss, anger and guilt and require validation that these are normal feelings.

Encourage the person and their parents or carers to talk to a counsellor or contact a support group that provides information, advocacy and support for people living with FASD and families caring for people living with FASD.

- National Organisation for Fetal Alcohol Syndrome and Related Disorders (NOFASARD) http://www.nofasard.org/ or phone 1300 306 238
- Russell Family Fetal Alcohol Disorders Association (rffada) http://rffada.org/ or phone 1800 733 232

Speaking to a person undergoing diagnostic assessment for FASD

- Make eye contact with the person and use their name
- Keep instructions brief and use language that is not ambiguous
- Ask simple and single questions needing one answer – closed questions
- Don't speak too quickly, use repetition and ensure the person has understood the instructions and what is required of them
- The use of visual clues can be useful
- Don't assume that because the person is able to speak well that he or she can also understand what you are saying and follow through with suggestions or advice

Speaking to the parents or carers

The diagnostic assessment process is a particularly sensitive and emotive time for the person and their parents or carers, especially for birth parents. They may like to ask a support person, friend or relative to accompany them to the appointment.

Before the diagnostic assessment process

- Use clear language
- Explain the assessment process and any medical terminology
- Discuss the Information on FASD Diagnostic Assessment for Parents and Carers and provide a copy (Appendix A6)
- Discuss the Australian FASD Diagnostic Assessment Consent Form (Appendix A7) and gain informed consent for the assessment and provide a copy
- Some parents may themselves be affected by fetal alcohol exposure – be aware of the possibility of intergenerational alcohol harm

After the diagnostic assessment process

- Discuss the content of the reports from the occupational therapist, speech pathologist, psychologist or other health professionals with the person and their parents/carers and provide a copy of each report.
- Provide a definite referral and 'next steps' plan and ensure they are appropriate for the diagnosis whether FASD or any other diagnosis.
- Provide some written information on the diagnosis and management plan so the person and their parents or carers can take it away and read it at a later time and discuss it with other people.
- For a child discuss how this information will be important to share with their school. Parents or carers will need to provide consent for any reports to be sent directly to the school; however the parent or carer may take their copy of the reports to the school to develop an appropriate plan and access services through the education system.
- Allow the person, parents, carers or their support person to ask questions during the appointment and provide contact details for follow up communication if required.

AUSTRALIAN FASD CRITERIA FOR REFERRAL FOR DIAGNOSTIC ASSESSMENT

The effective management of Fetal Alcohol Spectrum Disorders (FASD) in Australia requires appropriate methods for the identification and referral of individuals for diagnostic assessment. The need for an individual to be assessed for FASD may be identified in a range of contexts, and there is a lack of evidence to support formal screening for FASD in the general population. Until suitable formal screening tests are developed, we recommend evaluation by a primary health care provider using the following criteria for referral to an appropriate medical practitioner for a diagnostic assessment.

Criteria for referral

Review patient history and clinical features that may be consistent with a FASD. If any one of the following five criteria for referral is met, a diagnostic assessment is recommended:

1. Confirmed moderate or high level[†] prenatal alcohol exposure
2. 2 or more characteristic FAS facial anomalies
3. 1 facial anomaly and growth deficit and 1 or more CNS abnormalities
4. Microcephaly and any confirmed prenatal alcohol exposure
5. 2 or more CNS abnormalities and any confirmed prenatal alcohol exposure

[†]defined as exposure at any time during pregnancy to: 7+ standard drinks per week or 5+ standard drinks per occasion; or strong clinical suspicion of high level prenatal alcohol exposure

CNS-central nervous system; FAS-fetal alcohol syndrome

Assessment methods

Assessment of growth deficit, head circumference, and characteristic FAS facial anomalies in the referral context should use standardised assessment methods as detailed in the Guide to the Australian FASD Diagnostic Instrument (Appendix A1).

The assessment of CNS abnormalities in the referral context may use, but does not require, evidence of abnormality based on formal standardised psychometric assessment.

- Individual, parent or caregiver report (including from appropriate health or education professionals) of significant dysfunction or difficulties that are not able to be explained by other causes or conditions can be used to establish evidence of suspected significant CNS abnormalities for the purpose of referral.
- Areas of CNS abnormality that may be affected include cognition, memory, language, executive function, attention, adaptive behaviour, social skills and social communication, academic achievement, motor function and other areas.