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

Australian FASD Collaboration

Final Report: Volume 2

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CONTENTS

APPENDIX B: AUSTRALIAN FASD COLLABORATION MEMBERS

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One hundred and three health professionals completed part or all of the two rounds of the Delphi process. We are extremely grateful for their valuable contribution to this project. The following people agreed to the inclusion of their names in this final report.

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APPENDIX D: SYSTEMATIC LITERATURE REVIEW (FULL VERSION)

CONTENTS (FOR THE FULL LITERATURE REVIEW)

LIST OF TABLES (FOR THE FULL LITERATURE REVIEW)

List of Appendices

Index of Abbreviations

ARBD	Alcohol-Related Birth Defects
ARND	Alcohol-Related Neurodevelopmental Disorder
CDC	Centers for Disease Control and Prevention
CNS	Central Nervous System
FAS	Fetal Alcohol Syndrome
FASD	Fetal Alcohol Spectrum Disorders
IOM	Institute of Medicine
PFAS	Partial Fetal Alcohol Syndrome

INTRODUCTION

Alcohol is a potent teratogen, and prenatal exposure to alcohol during pregnancy may damage the developing brain and other major organs in the fetus (Stratton et al., 1996). Not every fetus exposed to alcohol will be affected in the same way because the timing, dose and pattern of maternal drinking and other socio-behavioural and biological factors may influence the severity of the outcome (Department of Health Western Australia, 2010).

Exposure to alcohol during pregnancy can result in a spectrum of life-long disorders (Manning & Eugene Hoyme, 2007). The term Fetal Alcohol Spectrum Disorders (FASD) is not a medical diagnosis (S. J. Astley et al., 2009a); rather, it is used as an umbrella term to describe the spectrum of disorders caused by alcohol exposure during pregnancy (Department of Health Western Australia, 2010; May et al., 2010).

OBJECTIVE

The purpose of this systematic review is to inform the development of an evidence-based instrument that can be used to improve the postnatal identification and/or diagnosis of FASD in Australia, thus enabling earlier intervention and management to improve health outcomes, increase functioning of persons diagnosed with a disorder from the FASD continuum, and improve the quality of life for individuals and families affected by FASD.

RATIONALE

A number of screening and diagnostic instruments have been developed overseas (S J Astley, 2004; BMA Board of Science, 2007; Chudley et al., 2005; Hoyme et al., 2005; National Center on Birth Defects and Developmental Disabilities, 2004; Stratton, et al., 1996). There is a need to develop a nationally applicable, evidence-based screening and diagnostic instrument for FASD for use in Australia. Acceptance and implementation of such an instrument would inform health professional training, service development and prevention programs, and through early and accurate diagnosis, improve the quality of life for those with FASD and their families. Standardisation of data collection would ensure data were comparable throughout Australia.

In Australia, a large proportion of women of child-bearing age consume alcohol, often at high levels and over half of pregnant women (58.7%) drink alcohol during pregnancy (Colvin et al., 2007). It has been suggested that FASD are under-diagnosed and under-reported in Australia. This may be due to the lack of a standardised screening and diagnostic instrument (Elliott, Payne et al. 2008) and due to the

implications of and possible stigma associated with such diagnoses (Bertrand et al., 2005; E. Elliott et al., 2008). Furthermore, fewer than half of Australian health professionals routinely asked women about alcohol consumption in pregnancy or routinely provided information to pregnant women about the effects of alcohol use in pregnancy (E. Elliott et al., 2006; Payne et al., 2005). Less than 20% of Australian health professionals knew the four essential criteria for the diagnosis of FAS and over half were concerned about stigmatising the child or the family with a diagnosis of FAS. A small proportion (less than 5%) felt very prepared to deal with FAS (E. Elliott, et al., 2006; Payne, et al., 2005).

Early diagnosis of FASD is important to allow for early health and educational interventions that will improve outcomes for the child (Caprara et al., 2007). Research shows that a lack of an early diagnosis is strongly correlated to adverse outcomes, and that the longer the delay in diagnosis, the greater the odds of adverse outcomes (Streissguth et al., 2004). Stratton and colleagues (1996) explain that: "A medical diagnosis serves several major purposes: to facilitate communication among clinicians; to facilitate communication between clinician and patient (including, in this instance, the parents of patients); to assist in the study of pathophysiology and etiology; and to guide treatment" p. 64 (Stratton, et al., 1996). Early diagnosis also provides opportunities for referral of the mother for medical attention.

METHODS

RESEARCH AIM

A systematic review of literature was conducted to identify existing postnatal screening or diagnostic criteria and guidelines for the diagnosis of disorders within the FASD continuum. The current review builds on the postnatal screening and diagnostic literature identified in the report by the Health Services Assessment Collaboration in New Zealand (L. Elliott et al., 2008).

SEARCH STRATEGY

In order to identify additional literature published since the New Zealand Health Services Assessment Collaboration review (L. Elliott, et al., 2008), the same search strategy was employed to search the literature published between 2008 and 30th September 2010 was searched. In addition, the Steering Group of the FASD Collaboration developed a further list of search terms and databases to be searched in order to expand the results. The databases were searched for these terms from the inception date of each database through to the 30th September 2010. A number of Internet sources relevant to the screening and diagnosis of FASD were also included in the search. Further information was sourced

from the reference lists of literature identified in the search and from key informants. Key informants included members of the FASD Collaboration Steering Group.

Keyword search matrices comprising search terms and databases or Internet sources were used to record the number of retrieved articles for each search. The matrices are located in Appendix 1 (Full Literature Review)

The review includes findings from the New Zealand systematic review conducted by Elliott and colleagues (L. Elliott, et al., 2008) and considers the results of the Murdoch Childrens Research Institute report on asking about alcohol use during pregnancy (Muggli et al., 2010) and the Department of Health (WA) Child and Youth Network Fetal Alcohol Spectrum Disorder Model of Care (Department of Health Western Australia, 2010).

SEARCH METHOD

The criteria for inclusion for this review included postnatal screening or diagnostic criteria, guidelines or instruments and rigorously conducted original research papers referring to postnatal screening or diagnostic criteria, guidelines or instruments. The search was limited further to human studies and articles published in English.

The Research Officer scanned abstracts of retrieved articles and those deemed relevant for further review were retained. These documents were saved in a PDF format with all citations imported into an Endnote library. One of the Lead Investigators reviewed the retained abstracts to determine the literature relevant for inclusion in the review. Excluded articles included duplicates and those that did not meet the study's inclusion criteria. For example, studies that addressed only prenatal screening were excluded.

The final selection of articles was distributed among a sub-group of the Steering Group. The sub-group performed a data extraction for each study by entering information into a data extraction form. Two different data extraction forms were developed; one for screening and diagnostic instruments or guidelines and review articles, and the other for original research (please see Appendix 3). The sub-group critically reviewed each of the studies for overall quality and relevance. Further documents were eliminated from the review following the critical appraisal. Articles were excluded due to poor or weak methodology, and if they were not relevant to the postnatal screening or diagnosis of disorders within the FASD spectrum (Appendix 6).

The literature findings are presented in the next section of this report under the headings of: screening and referral guidelines; diagnostic criteria and guidelines; and screening methods. Data extraction forms for literature included in this review are located in Appendix 4.

FINDINGS

BACKGROUND

The Health Services Assessment Collaboration of New Zealand undertook a systematic review of the literature on the prevention, diagnosis and management of FASD. A review of top-level strategies was conducted and included literature published from 1966 to July 2008 (L. Elliott, et al., 2008). The results included the following published postnatal screening or diagnostic criteria and guidelines and one key review article.

Diagnostic Criteria and Guidelines:

- Institute of Medicine Diagnostic Criteria for FASD (Stratton, et al., 1996)
- Diagnostic Guide for FASD: The 4-Digit Diagnostic Code (S J Astley, 2004)
- Updated Institute of Medicine Criteria for FASD (Hoyme, et al., 2005)
- FASD: Canadian Guidelines for Diagnosis (Chudley, et al., 2005)
- FAS: Guidelines for Referral and Diagnosis (National Center on Birth Defects and Developmental Disabilities, 2004)
- FASD: A Guide for Healthcare Professionals (BMA Board of Science, 2007)

Key review article:

- International survey of diagnostic services for children with Fetal Alcohol Spectrum Disorders (Peadon et al., 2008)

Elliott and colleagues (2008) acknowledge that postnatal screening can identify individuals who may have one of the FASD and that positive results from screening should be followed by a diagnostic assessment. Their review suggested that it was not possible to identify strategies for screening or diagnosis of FASD for New Zealand as the postnatal screening and diagnostic literature did not evaluate the accuracy of the diagnostic criteria. They conclude that there is no international consensus and no evidence that any one criterion is the most appropriate (L. Elliott, et al., 2008).

The Murdoch Childrens Research Institute conducted a review of literature to demonstrate the need for a comprehensive measure for alcohol use in pregnancy. The report recommended that a clear

definition of low, moderate and high risk levels of maternal alcohol intake should be used in a research setting when investigating prenatal alcohol consumption along with a pictorial guide that illustrates a standard drink across a range of alcoholic beverages (Muggli, et al., 2010). The report suggested that it is important to record drinking patterns and timing of the alcohol exposure and to identify potential factors (confounders, modifiers and mediators) that can affect the measure of association between alcohol exposure and outcomes (Muggli, et al., 2010). Their research found that women were willing to answer questions about alcohol use in pregnancy, but that women with alcohol problems may under-report actual intake (Muggli, et al., 2010). Existing antenatal screening instruments were examined and the authors found that they were most useful for identifying high risk drinking. They also recommended that, in a clinical setting, women should be screened for alcohol consumption with a validated instrument that determines the quantity of alcohol consumed (Muggli, et al., 2010).

The Western Australian Department of Health released a 'Model of Care' document for FASD (Department of Health Western Australia, 2010). The model prioritises the use of prevention strategies to reduce the prevalence of FASD, recognising there is no cure for the condition. A number of recommendations were documented in the report and cover issues pertaining to primary, secondary and tertiary prevention; universal and targeted screening; clinical pathways; diagnosis and service delivery in metropolitan and rural and remote areas; workforce professional development, training and education; and monitoring, evaluation and surveillance (Department of Health Western Australia, 2010). The model recommends universal screening for alcohol consumption for women of child-bearing age and during pregnancy; and for new-borns or children who are at risk of prenatal alcohol exposure. It is suggested that a multi-disciplinary diagnostic service be developed within the Child Development Service along with pathways to other relevant services and agencies (Department of Health Western Australia, 2010).

SCREENING AND REFERRAL GUIDELINES

FAS SCREEN

Burd and colleagues (1999) explain that a screening tool should be used at a population level in order to identify individuals who are likely to have FAS. FAS Screen was developed as a rapid 15-minute, 32 item screening test for 4 to 18 year olds (Burd et al., 1999; Poitra et al., 2003). The screening test records the age, gender and ethnicity of an individual and includes items for measuring growth impairment, neurologic dysfunction and facial features.

Table 1 lists the items within each of these domains. Measurements for height, weight and head circumference are recorded if they are below the 5th percentile, although the authors note that this will result in inclusion of individuals with growth parameters in the lower end of population norms but without FAS. A 'yes' or 'no' option is provided for each category and a score is assigned for a 'yes' response. If a score totals 20 or above, referral is recommended for further assessment (Burd, et al., 1999).

Table 1 FAS Screen Items

	Score
Growth, head and face	
Height <5%	10
Weight <5%	10
Head circumference <5%	10
Ears stick out (Protruding auricles)	4
Skin folds near inner eye (Epicanthal folds)	5
Drooping of eyelids (Ptosis)	4
Cross-eyes, one or both eyes (Strabismus)	3
Flat midface/cheeks (Hypoplastic maxilla)	7
Flat/low nose between eyes (Low nasal bridge)	2
Upturned nose	5
Groove between lip & nose absent or shallow (Flat philtrum)	5
Thin upper lip	4
Cleft lip or cleft of roof of mouth (Present or repaired)	4
Neck and back	
Short, broad neck	4
Curvature of the spine (Scoliosis)	1
Spina bifida (History of neural tube defect)	4
Arms and hands	
Fingers, elbows (Limited joint mobility)	4
Permanently curved, small fingers, especially pinkies (Clinomicrodactyly)	1
Deep or accentuated palmar creases	4
Small nails/nail beds (Hypoplastic nails)	1
Tremulous, poor finger agility (Fine motor dysfunction)	1
Chest	
Sunken chest (Pectus Excavatum)	3
Chest sticks out (Pectus Carinatum) optional	1
History of heart murmur or any heart defect	4
Skin	
Raised red birthmarks (Capillary Hemangiomas)	4
Greater than normal body hair, hair also on forehead and back (Hirsutism)	1
Development	
Mild to moderate mental retardation (IQ < 70)	10

Speech and language delays	2
Hearing problems	1
Vision problems	1
Attention concentration problems	2
Hyperactivity	5

(Burd, et al., 1999)

The sensitivity and specificity of the screening instrument was obtained by screening 1013 children aged 3 to 14 years from schools in North Dakota. The sensitivity of the instrument was calculated to be 100%; specificity was 94.1%; the positive predictive value was 9.2% and the negative predictive value was 100% (Burd, et al., 1999). The FAS Screen was designed for use in community-based screening programs and has not been evaluated for its application in the clinical setting (Burd et al., 2000).

A review conducted by the Public Health Agency of Canada highlights the advantages and disadvantages of this screening tool (Goh & Rosenbaum, n.d.). The advantages include its low cost allowing schools to complete it without requiring additional financial, logistical or technical support. However, this method does not consider neurobehavioral deficits and front-line healthcare providers may not be confident in assessing dysmorphic features (Goh & Rosenbaum, n.d.).

YOUTH PROBATION OFFICERS' GUIDE TO FASD SCREENING AND REFERRAL

The Department of Justice Canada, funded the development of the Youth Probation Officers' Guide to FASD Screening and Referral as a result of disproportionately high numbers of individuals with FASD entering the justice system (Conry & Asante, 2010). The guide was developed by the Asante Centre for Fetal Alcohol Syndrome and designed to increase the capacity of probation officers in Canada to identify youth requiring referral for FASD assessment. The Asante Centre emphasises that these guidelines are not sufficient to confirm a diagnosis, rather the screening tool is used to identify individuals who are likely to have a particular condition so that a comprehensive, diagnostic assessment can follow (Conry & Asante, 2010).

The screening tool comprises items that are linked to specific criteria for making a diagnosis. The information required for each item is generally available to a probation officer and does not require special expertise to ascertain (Conry & Asante, 2010). Background information is collected, including information about legal guardian (birth parent, adoptive parent, social worker or other) and the person with whom the individual resides (birth parent, adoptive parent, foster parent, group home, custody centre or other). The screening checklist (Table 2) includes items pertaining to social and personal

factors. If one social factor and at least two personal factors, or no social factors and at least three personal factors are recorded, an individual is referred for further assessment.

Table 2 Screening Checklist for Youth Probation Officers

A) **Social Factors** are those that may identify a youth at-risk for FASD. That is, these factors may increase the probability that the youth could have FASD:

- Youth is adopted
- Youth currently, or previously, was in foster care or involved with child protection services
- Youth has a sibling with a documented diagnosis of FASD
- There is documentation that the youth is suspected of having FASD
- Youth's mother has known history of alcoholism or prenatal alcohol use

B) **Personal Factors** are those that have been associated with (but not necessarily unique to) FASD:

- Developmental delay in early childhood (speech/language therapy, occupational therapy, infant development or child development services prior to school entry)
- Learning difficulties (learning assistance, modified program or experienced school failure or drop-out)
- Growth deficiency (appears short compared to peers, or of a low weight for age)
- Diagnosis of ADHD
- Mental health diagnosis (anxiety, depression, Oppositional Defiant Disorder, Conduct Disorder)

(Conry & Asante, 2010)

If an individual is referred for further assessment, the Asante Centre recommends that probation officers compile a record of detailed information from past medical records and other sources, such as the birth mother, physician's/midwife's prenatal and birth records, maternal grandparents/aunts, social workers' records, father's or mother's partners (Conry & Asante, 2010).

NATIONAL SCREENING TOOL KIT FOR CHILDREN AND YOUTH IDENTIFIED AND POTENTIALLY AFFECTED BY FASD

A Steering Committee convened by the Canadian Association of Paediatric Health Centres, in collaboration with experts and providers from Canada and the United States developed the 'National Screening Tool Kit for Children and Youth Identified and Potentially Affected by Fetal Alcohol Spectrum Disorder (FASD)' (Canadian Association of Paediatric Health Centres, 2010). The Steering Committee identified and evaluated FASD screening tools and methods used in Canada and developed guidelines based on these evaluations. The identification and evaluation processes included: a survey of diagnostic clinics in Canada; critical review of literature; establishment of a 'National Advisory'; workshops for researchers and health-care providers; piloting of tools; and a process for future tool evaluation. The

criteria used to evaluate identified tools included: sensitivity; specificity; positive and negative predictive values; and practical applicability (ease of use, accessibility, cost, expertise required, cultural appropriateness, and interpretation of results) (Canadian Association of Paediatric Health Centres, 2010). The Took Kit comprises a number of screening tool options to account for the differences in ages, stages and settings that can influence screening requirements (Table 3).

Table 3 National Screening Tool Kit Screening Tools

Tool	Type of screen	Population screened	Setting	Sector
Neurobehavioural Screening Tool (NST)	Maladaptive behaviour	6-18 years	Elementary & secondary schools, primary care, and children's mental health	Health Social Services Education
Meconium FAEE testing	Prenatal exposure	New-borns At-risk mothers	Hospital/home	Health
Maternal Drinking Guide	Prenatal exposure	At-risk women	Primary care, prenatal care, women's mental health, specialized child health	Health Social Services
Medicine Wheel Student Index Medicine Wheel Developmental History	Maladaptive Behaviour Prenatal exposure	4-14 years At-risk mothers	Elementary & middle school Home, counselling services	Education Health Social Services
FASD Screening & Referral Form for Youth Probation Officers	Maladaptive behaviour	Youth	Youth Justice System	Justice

(Canadian Association of Paediatric Health Centres, 2010)

The Tool Kit lists the benefits and limitations of each of the screening tools shown in Table 3.

- The Steering Committee found the *Neurobehavioural Screening Tool* to be a simple checklist that can be administered to a parent or carer by health or social services professionals; and that it can identify children who may be affected by FASD and differentiates between Attention Deficit Hyperactivity Disorder and Oppositional Defiant Disorder/Conduct Disorder. The screening tool takes less than five minutes to complete and is free of charge for use in Canada. Limitations of this method include the potential for confounders such as age, gender, socioeconomic status; Intelligence Quotient is not measured and the tool has not been tested in a large population.
- *Meconium FAEE testing* of new-borns can identify fetal alcohol exposure during pregnancy; improve early diagnosis and intervention; and identify high risk pregnancies. However, this

method is only effective for screening for maternal alcohol consumption more than 12 weeks post conception. Furthermore there is potential for misuse of positive screen results by courts and social services agencies; and it is relatively expensive if not implemented o a wide-scale.

- The *Maternal Drinking Guide* is used to screening women for high risk alcohol use during pregnancy and has been validated as an effective method of determining maternal alcohol use. The questions can be easily asked as part of an overall health assessment and can provide opportunity for education, harm reduction and referral. Limitations include a lack of time for health professionals to complete the screening and lack of services for referral and follow-up. This method has not been validated for retrospective collection of alcohol consumption during pregnancy.
- The *Medicine Wheel Tools* were developed as a culturally sensitive assessment or screening tool for use within the Aboriginal school and community system in Canada. The tools have been tested with First Nations people in Canada and have been adapted for use within Inuit cultures. They fit within traditional practices and can track specific behaviours over time. However the training needs and resources required for implementation have not been assessed and the tools require further validation. A lack of services may limit referral for full assessment.
- The *Youth Probation Officers' Guide to FASD Screening and Referral* has been discussed above. The Steering Committee suggests this guide is beneficial for addressing needs of a high risk group. It comprises a referral form that is easy to use, a case management form that is practical and useful and clear criteria for further assessment and referral. Limitations of this tool include difficulty in accessing maternal history, time constraints for probation officers, limited assessment and diagnostic services and the need for validation in other jurisdictions (Canadian Association of Paediatric Health Centres, 2010).

FASD: A GUIDE FOR HEALTHCARE PROFESSIONALS

This document was prepared by the British Medical Association Board of Science to raise awareness of FASD, particularly among healthcare professionals and organisations within the public health arena (BMA Board of Science, 2007). The report suggests that maternal alcohol consumption should be monitored, and although there is no definitive test that can accurately identify alcohol use during pregnancy, the Board recommend a number of potential tests including biomarkers (fatty acid ethyl esters) and the T-ACE and TWEAK screening questionnaires (Appendix 2). They recommend that routine screening should be considered as part of antenatal assessment (BMA Board of Science, 2007).
The British Medical Association Board of Science suggests that health departments in the UK should provide guidance for healthcare professionals on identification, referral and diagnosis for the full range of FASD; and ensure provision of appropriate diagnostic and referral services with adequate funding for

development, training and maintenance of multidisciplinary diagnostic teams (BMA Board of Science, 2007).

DIAGNOSTIC CRITERIA AND REFERRAL GUIDELINES

INSTITUTE OF MEDICINE DIAGNOSTIC CRITERIA

In 1996, the Institute of Medicine Diagnostic Criteria were developed in consultation with a panel of experts (Stratton, et al., 1996). They were based on a review of children with clinical abnormalities and confirmed alcohol exposure in utero (E. Elliott & Peadon, 2009). The panel developed a systematic approach for defining diagnostic categories that included Fetal Alcohol Syndrome (FAS) with and without confirmed alcohol exposure, partial FAS (PFAS), alcohol-related birth defects (ARBD) and alcohol-related neurodevelopmental disorder (ARND).

Table 4 presents the diagnostic criteria for FAS and alcohol-related effects as delineated by the Institute of Medicine. Confirmed maternal alcohol exposure is defined as "A pattern of excessive intake characterized by substantial, regular intake or heavy episodic drinking" (Stratton, et al., 1996). This can include evidence of frequent occurrences of intoxication; experiencing tolerance or withdrawal, social problems or legal problems as a result of drinking; engaging in physically hazardous behaviour or developing alcohol-related medical problems (Stratton, et al., 1996). Lower levels of consumption or variable patterns of alcohol use are included in the criteria for their association with ARBD and/or ARND (Stratton, et al., 1996).

Individuals diagnosed with FAS, with or without confirmed maternal alcohol exposure, are identified as having a characteristic pattern of minor facial anomalies (short palpebral fissures, a thin upper lip, and flat philtrum and mid-face); at least one form of growth abnormality (low birth weight for gestational age, decelerating weight over time not due to nutrition, disproportional low weight to height); and at least one central nervous system (CNS) neurodevelopmental abnormality (decreased cranial size at birth, structural brain anomalies, neurological hard or soft signs or neurodevelopmental delay). A diagnosis of partial FAS includes confirmed maternal alcohol exposure; evidence of some characteristic facial anomalies; and either evidence of growth retardation, CNS neurodevelopmental abnormality, or a complex pattern of behaviour or cognitive abnormalities consistent with developmental level that cannot be attributed to family or environmental origins. In these criteria, alcohol-related effects comprise the categories of ARBD and ARND. They refer to clinical conditions when there is a history of

maternal alcohol exposure and abnormalities are present in individuals who do not fulfill the diagnostic criteria for FAS (Stratton, et al., 1996).

Table 4 IOM Diagnostic Criteria for Fetal Alcohol Syndrome and Alcohol-Related Effects

Fetal Alcohol Syndrome

1. FAS with confirmed maternal alcohol exposure[a]

A. Confirmed maternal alcohol exposure[a]

B. Evidence of a characteristic pattern of facial anomalies that includes features such as short palpebral fissures and abnormalities in the maxillary region (thin upper lip, flattened philtrum and flat midface)

C. Evidence of growth retardation, as in at least one of the following:
- low birth weight for gestational age
- decelerating weight over time not due to nutrition
- disproportional low weight to height

D. Evidence of CNS neurodevelopmental abnormalities, as in at least one of the following:
- decreased cranial size at birth
- structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia)
- neurological hard or soft signs (as age appropriate), such as impaired fine motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination

2. FAS without confirmed maternal alcohol exposure

B, C, and D as above

3. Partial FAS with confirmed maternal alcohol exposure

A. Confirmed maternal alcohol exposure[a]

B. Evidence of some components of the pattern of characteristic facial anomalies

Either C or D or E

C. Evidence of growth retardation, as in at least one of the following:
- low birth weight for gestational age
- decelerating weight over time not due to nutrition
- disproportional low weight to height

D. Evidence of CNS neurodevelopmental abnormalities, as in at least one of the following:
- decreased cranial size at birth
- structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia)
- neurological hard or soft signs (as age appropriate), such as impaired fin motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination

E. Evidence of a complex pattern of behaviour or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone, such as learning difficulties; deficits in school performance; poor impulse control; problems in social perception; deficits in higher level receptive and expressive language; poor capacity for abstraction or metacognition; specific deficits in mathematical skills; or problems with memory, attention, or judgement

Alcohol-Related Effects

Clinical conditions in which there is a history of maternal alcohol exposure,[a,b] and where clinical or animal research has linked maternal alcohol ingestion to an observed outcome.

There are two categories which may co-occur. If both diagnoses are present, then both diagnoses should be rendered:

4. Alcohol-related birth defects (ARBD)

Cardiac: atrial septal defects; ventricular septal defects; aberrant great vessels; tetralogy of Fallot

Skeletal: hypoplastic nails; shortened fifth digits; radioulnar synostosis; flexion contractures; camptodactyly; clinodactyly; pectus excavatum and carinatum; Klippel-Feil syndrome; hemivertebrae; scoliosis

Renal: aplastic, dysplastic or hypoplastic kidneys, horseshoe kidneys, ureteral duplications, hydronephrosis

Ocular: strabismus, refractive problems secondary to small globes, renal vascular anomalies

Auditory: conductive hearing loss, neurosensory hearing loss

Other: virtually every malformation has been described in some patient with FAS. The etiologic specificity of most of these anomalies to alcohol teratogenesis remains uncertain

5. Alcohol-related neurodevelopmental disorder (ARND)

A. Evidence of CNS neurodevelopmental abnormalities, as in any one of the following:
- decreased cranial size at birth
- structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia)
- neurological hard or soft signs (as age appropriate), such as impaired fine motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination

AND/OR:

B. Evidence of a complex pattern of behaviour or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone, such as learning difficulties; deficits in school performance; poor impulse control; problems in social perception; deficits in higher level receptive and expressive language; poor capacity for abstraction or metacognition; specific deficits in mathematical skills; or problems with memory, attention, or judgement

[a]A pattern of excessive intake characterised by substantial, regular intake or heavy episodic drinking. Evidence of this pattern may include frequent episodes of intoxication, development of tolerance or withdrawal, social problems related to drinking, legal problems related to drinking, engaging in physically hazardous behaviour while drinking, or alcohol-related medical problems such as hepatic disease.

[b]As further research is completed and as, or if, lower quantities or variable patterns of alcohol use are associated with ARBD or ARND, these patterns of alcohol use should be incorporated into the diagnostic criteria.

(Stratton, et al., 1996)

Hoyme and colleagues (2005) reported a number of concerns with the Institute of Medicine Diagnostic Criteria. They suggest the criteria were vague, without specific objective parameters for items in each diagnostic category (Hoyme, et al., 2005). They explain that the degree of growth deficiency and the exact facial features required for each category are not defined; the specific behavioural/cognitive phenotype is not characterised; there are no guidelines for assessment of the behavioural or cognitive difficulties; family and genetic history is not sufficiently addressed; and that ARBD and ARND are not defined in a practical manner to make them useful in a clinical setting (Hoyme, et al., 2005).

The 4-Digit Diagnostic Code was developed by the University of Washington in 1997 to overcome limitations arising from the gestalt approach that does not assure diagnostic accuracy and precision when diagnosing prenatal alcohol exposure (S J Astley, 2004; S. J. Astley & Clarren, 1995, 2001). The guide has been updated following its application to over 2,000 patients, with advice from clinical teams and the advancement of medical research and other diagnostic guidelines (S J Astley, 2004). Unlike the Institute of Medicine Diagnostic Criteria for FASD, this method specifies the number and degree of abnormalities that should be present to define a diagnosis (L. Elliott, et al., 2008) and allows for a diagnosis of FAS or other diagnoses within the FASD continuum to be made (S J Astley, 2004; L. Elliott, et al., 2008). The principal author suggests this diagnostic approach is logical and easy to use (S J Astley, 2004).

The 4-Digit Diagnostic Code applies specific case definitions and objective and quantitative measurement scales to increase the accuracy of diagnosis (S J Astley, 2004; S. J. Astley & Clarren, 2000). The four digits represent the diagnostic features of FASD being: growth deficiency, the FAS facial phenotype, CNS abnormalities, and gestational exposure to alcohol (S J Astley, 2004). The magnitude of each feature is ranked on a 4-point Likert scale with regard to level of severity. A score of 1 reflects absence of a feature and a score of 4 represents a strong presence of a feature (Table 5). Final ranking scores can range from 1111 (meaning there is an absence of growth abnormalities and characteristic facial features, unlikely CNS damage and no risk of prenatal alcohol exposure) to 4444 (being the most severe ranking showing severe growth deficiency, the full facial phenotype, definite CNS damage and confirmed high risk levels of prenatal alcohol exposure).

Table 5 4-Digit Diagnostic Code Criteria for FASD

Rank	Growth deficiency	FAS facial phenotype	CNS damage or dysfunction	Gestational exposure to alcohol
4	**Significant** Height and weight below 3rd percentile	**Severe** All 3 features: Palpebral fissure length 2 or more standard deviations below the mean; upper lip rank 4 or 5 and philtrum rank 4 or 5	**Definite** Structural or neurologic evidence 2 or more standard deviations below the mean	**High risk** Confirmed exposure to high levels
3	**Moderate** Height and weight below 10th percentile	**Moderate** Generally 2 of the 3 features	**Probable** Significant dysfunction across 3 or more domains	**Some risk** Confirmed exposure. Level of exposure unknown or less than rank 4

2	**Mild** Height or weight below the 10th percentile	**Mild** Generally 1 of the 3 features	**Possible** Evidence of dysfunction, but less than rank 3	**Unknown** Exposure not confirmed present or absent
1	**None** Height and weight at or above 10th percentile	**Absent** None of the 3 features	**Unlikely** No structural, neurologic or functional evidence of impairment	**No risk** Confirmed absence of exposure from conception to birth

(S J Astley, 2004)

There are a total of 256 possible outcomes using this method and 22 diagnostic categories, eight of which are within the FASD continuum (S J Astley, 2004). The clinical diagnostic categories (Table 6) are used in varying combinations to make the 22 diagnostic categories.

Table 6 4-Digit Clinical Diagnostic Categories

Sentinel Physical findings	This term is used when a patient presents with growth deficiency at the Rank 3 or 4 level and/or presents with the FAS facial phenotype at the Rank 3 or 4 level. Other physical findings (major or minor anomalies) may be detected instead of or in addition to these sentinel findings that may suggest alternate or additional conditions.
Static Encephalopathy	This term is used when the patient presents with significant structural, neurological, and/or functional abnormalities that strongly support the presence of underlying CNS damage at the Rank 3 and/or Rank 4 levels. The term does not define or suggest any specific pattern of structural, neurological or functional abnormality.
Neurobehavioral Disorder	This term is used when the patient presents with cognitive/behavioural dysfunction at the Rank 2 level and no evidence of structural, neurological or functional abnormalities at the Rank 3 or Rank 4 levels.
Alcohol (Exposed, Not Exposed, Exposure Unknown	These terms are used to reflect prenatal alcohol exposure and its potential risk to the unborn child. Alcohol exposure is reported independently of outcome(s) and does not imply that a causal association exists between the exposure and outcome(s).
Fetal Alcohol Syndrome (alcohol exposed)	This term is used to refer to patients who present with one of twelve 4-Digit Diagnostic Code combinations reflecting growth deficiency; the full FAS facial phenotype; significant structural, neurological, and/or functional CNS abnormalities; and confirmed prenatal alcohol exposure.
Fetal Alcohol Syndrome (alcohol exposure unknown)	A diagnosis of FAS can be rendered when prenatal alcohol exposure is unknown but only when the outcomes (growth, face and CNS) are at the severe end of the spectrum to maintain the specificity of these outcomes to prenatal alcohol exposure.
Partial Fetal Alcohol Syndrome (alcohol exposed)	This term is used for patients who present with static encephalopathy, most (but not all) of the growth and/or facial features of FAS, and have a confirmed history of prenatal alcohol exposure.
Fetal alcohol Syndrome Phenocopy (no alcohol exposure)	This term is used for patients who meet the growth, face and CNS criteria for FAS, but have a confirmed absence of alcohol exposure during gestation (no known cases of this type to date).

(S J Astley, 2004)

Growth measurements using the 4-Digit Diagnostic Code require height and weight to be adjusted for age and gender, and separated into prenatal and postnatal growth. Facial features are measured by either a direct measurement (palpebral fissure length is measured using a clear plastic ruler) or on a digital facial photograph using the FAS Facial Photographic Analysis Software (S. Astley, 2003). The Lip-Philtrum Guide (Caucasian Lip-Philtrum Guide or African American Lip-Philtrum Guide) is used to identify lip thinness and philtrum smoothness. Evidence of structural CNS damage includes head circumference less than the 3rd percentile and/or significant brain abnormalities on neuroimaging. Evidence of neurological dysfunction includes impairment in any of the domains of executive function, memory, cognition, social/adaptive skills, academic achievement, language, motor, attention and activity level. When alcohol exposure is known, it is ranked according to quantity, timing, frequency and certainty of exposure during pregnancy (S J Astley, 2004).

The University of Washington group recommends that a diagnostic assessment be carried out by a multidisciplinary team of professionals including a physician, psychologist, speech-language pathologist and occupational therapist, in order to achieve an accurate, global assessment of function (S J Astley, 2004).

UPDATED INSTITUTE OF MEDICINE CRITERIA

Hoyme and colleagues (2005) acknowledge that the 4-Digit Diagnostic Code (S J Astley, 2004) is accurate in placing patients into a specific diagnostic category, however they suggest the method is confusing and impracticable for use in the clinical, as opposed to the research setting. They report that the diagnostic code has similar shortfalls to those the Institute of Medicine Diagnostic Criteria (Stratton, et al., 1996), being that family and genetic history are not adequately accounted for (Hoyme, et al., 2005). They propose a revision and clarification of the 1996 Institute of Medicine Diagnostic Criteria in order to make them more applicable for clinical practice and to increase their reliability and validity. They explain that an ideal classification system would accurately diagnose individuals by defining diagnostic categories, minimising false-positive and false-negative outcomes, accounting for genetic and family history, applying a multidisciplinary approach and using practical terms that could be easily used in clinical settings (Hoyme, et al., 2005). The importance of exclusion of other conditions is also stressed, when applying a diagnosis within the FASD continuum.

The clarifications (Table 7) require that a patient diagnosed with FAS (with or without confirmed prenatal alcohol exposure) must show abnormalities across all three domains: characteristic facial features, growth and CNS. For a diagnosis of partial FAS, a patient must display characteristic facial features and abnormalities in one of the other domains of growth or CNS structure or function (Hoyme,

et al., 2005). The updated criteria specify that maternal alcohol exposure must be documented in order to allocate a diagnosis of either ARBD or ARND. ARBD is defined as an individual with normal facial features, growth and development and who has one or more specific birth defects outside the CNS. A diagnosis of ARND applies to an individual with normal growth and structural development, but who show a characteristic pattern of behavioural or cognitive abnormalities (Hoyme, et al., 2005).

Table 7 Updated Institute of Medicine Criteria

I. FAS With Confirmed Maternal Alcohol Exposure (requires all features of A–D)

A. Confirmed maternal alcohol exposure

B. Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following:

1. short palpebral fissures ($\leq 10^{th}$ percentile)
2. Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide)
3. Smooth philtrum (score 4 or 5 with the lip/philtrum guide)

C. Evidence of prenatal and/or postnatal growth retardation

1. Height and/or weight $\leq 10^{th}$ percentile, corrected for racial norms, if possible

D. Evidence of deficient brain growth and/or abnormal morphogenesis, including ≥ 1 of the following:

1. Structural brain abnormalities
2. Head circumference $\leq 10^{th}$ percentile

II. FAS Without Confirmed Maternal Alcohol Exposure

IB, IC, and ID as above

III. Partial FAS With Confirmed Maternal Alcohol Exposure (requires all features, A-C)

A. Confirmed maternal alcohol exposure

B. Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following:

1. Short palpebral fissures ($\leq 10^{th}$ percentile)
2. Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide)
3. Smooth philtrum (score 4 or 5 with the lip/philtrum guide)

C. One of the following other characteristics:

1. Evidence of prenatal and/or postnatal growth retardation
   a. Height and/or weight $\leq 10^{th}$ percentile corrected for racial norms, if possible
2. Evidence of deficient brain growth or abnormal morphogenesis, including ≥ 1 of the following:
   a. Structural brain abnormalities
   b. Head circumference $\leq 10^{th}$ percentile
3. Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background, or environment alone
   a. This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behavior (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction)

IV. Partial FAS Without confirmed Maternal Alcohol Exposure

IIIB and IIIC, as above

V. ARBD (requires all features, A-C)

A. Confirmed maternal alcohol exposure

B. Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following:

1. Short palpebral fissures ($\leq 10^{th}$ percentile)
2. Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide)
3. Smooth philtrum (score 4 or 5 with the lip/ philtrum guide)

C. Congenital structural defects in ≥ 1 of the following categories, including malformation and dysplasias (if the patient displays minor anomalies only, ≥ 2 must be present): *cardiac:* atrial septal defects, aberrant great vessels, ventricular septal defects, conotruncal heart defects; *skeletal*: radioulnar synostosis, vertebral segmentation defects, large joint contractures, scoliosis; *renal*: aplastic/hypoplastic/dysplastic kidneys, ''horseshoe'' kidneys/ureteral duplications; *eyes:* strabismus, ptosis, retinal vascular anomalies, optic nerve hypoplasia; *ears:* conductive hearing loss, neurosensory hearing loss; *minor anomalies:* hypoplastic nails, short fifth digits, clinodactyly of fifth fingers, pectus carinatum/excavatum, camptodactyly, ''hockey stick'' palmar creases, refractive errors, ''railroad track'' ears

VI. ARND (requires both A and B)

A. Confirmed maternal alcohol exposure

B. At least 1 of the following:

1. Evidence of deficient brain growth or abnormal morphogenesis, including ≥ 1 of the following:
    a. Structural brain abnormalities
    b. Head circumference $\leq 10^{th}$ percentile
2. Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background, or environment alone
    a. This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behaviour (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction)

In the proposed diagnostic criteria, the following considerations apply. Each of the categories assumes that genetic and medical assessment has ruled out a phenocopy, including other genetic and malformation syndromes. Confirmed maternal alcohol exposure is defined as a pattern of excessive intake characterized by substantial regular intake or heavy episodic drinking. Evidence of this pattern may include frequent episodes of intoxication, development of tolerance or withdrawal, social problems related to drinking, legal problems related to drinking, engaging in physically hazardous behavior while drinking, or alcohol-related medical problems such as hepatic disease. Confirmation may be from maternal interview or reliable collateral sources.

(Hoyme, et al., 2005)

The revised classification system was tested with a large multiracial international cohort of children prenatally exposed to alcohol, but there was no unexposed comparison group. The authors report that the system was easily applied within the clinical setting with reproducible results (Hoyme, et al., 2005;

Manning & Eugene Hoyme, 2007). Hoyme and colleagues (2005) acknowledge that a lack of population norms across multiracial populations is a limitation of this method when values are used to plot growth and facial features. The authors recommend development of normative height and weight curves across racial groups (Hoyme, et al., 2005).

Astley (2006) reviewed the Updated Institute of Medicine Diagnostic Criteria (Stratton, et al., 1996) and noted that while the system addresses the full spectrum of FASD, it differs considerably from the 4-Digit Diagnostic Code (S J Astley, 2004), the FAS: Guidelines for Referral and Diagnosis (National Center on Birth Defects and Developmental Disabilities, 2004), and the FASD: Canadian Guidelines for Diagnosis (Chudley, et al., 2005). It specifies different and sometimes less stringent criteria for the diagnosis of FAS, for example by requiring only structural, not functional CNS abnormalities to be present; two of the three characteristic facial features (as opposed to all three); and a head circumference measured at or below the 10th percentile (rather than the 3rd percentile) (S. J. Astley, 2006).

FIRST NATIONS AND INUIT HEALTH COMMITTEE POSITION STATEMENTS ON FAS

The Canadian Paediatric Society First Nations and Inuit Health Committee developed a list of position statements on FAS covering prevention, diagnosis, early identification and management for health care professionals (First Nations and Inuit Health Committee, 2002). The committee recommends that a FAS diagnosis be considered if there is confirmed prenatal alcohol exposure, with current or previous growth deficiency, presence of characteristic facial features and neurodevelopmental abnormalities. They provide a list of 'age-related diagnostic criteria' for FAS and/or atypical FAS (Table 8), and recommend the use of the 4-Digit Diagnostic Code (S J Astley, 2004) for making the diagnosis of FAS (First Nations and Inuit Health Committee, 2002). The committee highlights the importance of identifying at-risk individuals within a culturally appropriate context and recommends that all women presenting to primary care physicians, midwives or nurse practitioners be asked about their drinking habits.

Table 8 Age-related Diagnostic Criteria for FAS and/or Atypical FAS

Infants
History of prenatal alcohol exposure
Facial abnormalities
Growth retardation – height, weight, head circumference
Hypotonia, increased irritability
Jitteriness, tremulousness, weak suck
Difficulty 'habituating', getting used to stimulation

Preschool
History of alcohol exposure, growth retardation, facial abnormalities
Friendly, talkative and alert
Temper tantrums and difficulty making transitions
Hyperactive; may be over sensitive to touch or over-stimulation
Apparent skill levels may appear to be higher than their tested levels of ability
Attention deficits, developmental delays – speech, fine motor difficulties
Middle childhood
History of alcohol exposure, growth retardation, facial abnormalities
Hyperactivity, attention deficit, impulsiveness
Poor abstract thinking
Inability to foresee consequences of actions
Lack of organisational skills
Inappropriate behaviour: overly affectionate – does not discriminate between family and strangers; lack of inhibitions; communication problems – lack of social skills to make and keep friends, unresponsive to social clues, uses behaviour as communication; difficulty making transitions
Academic problems – reading and mathematics
Behaviour problems – 'stretched toddler'
Adolescent and adult
History of alcohol exposure, growth retardation, facial abnormalities
Intelligence Quotient – average to mildly retarded with wide range; continued school difficulties
Difficulty with adaptive and living skills
Attention deficits, poor judgement, impulsivity lead to problems with employment, stable living and the law
Serious life adjustment problems – depression, alcoholism, crime, pregnancy and suicide

(First Nations and Inuit Health Committee, 2002)

FASD: CANADIAN GUIDELINES FOR DIAGNOSIS

The FASD: Canadian Guidelines for Diagnosis (Chudley, et al., 2005) were developed by a subcommittee of the Public Health Agency of Canada's National Advisory Committee on Fetal Alcohol Spectrum Disorders, following extensive consultation with Canadian and American professionals experienced in the diagnosis of FASD. Further input was sought from individuals, professional organisations and various levels of government. The guidelines combine elements of both the Institute of Medicine Diagnostic Criteria for FASD and the 4-Digit Diagnostic Code (E. Elliott & Peadon, 2009).

Importance is given to the use of a multidisciplinary team for the accurate diagnosis and treatment of individuals with a disorder in the FASD continuum. Such a team may include trained and experienced professionals including a nurse or social worker, physician trained in FASD diagnosis, psychologist, occupational therapist and a speech pathologist. Additional members could include childcare workers,

mental health workers, parents or caregivers, probation officers, psychiatrists, teachers, geneticists, dysmorphologists or cultural interpreters (Chudley, et al., 2005).

The guidelines are organised under six key areas: screening and referral; physical examination and differential diagnosis; neurobehavioural assessment; treatment and follow-up; maternal alcohol history in pregnancy; and diagnostic criteria for FAS, partial FAS and ARND (Chudley, et al., 2005). It is recommended that all pregnant and post-partum women be screened for alcohol use with a validated screening tool; and that background information including birth and pregnancy records, adoption records, medical and hospital records, academic records, developmental and psychological assessments and family history should be obtained to support a comprehensive assessment of an individual. In addition, all patients should undergo physical and neurologic examinations to exclude other disorders (Chudley, et al., 2005). The diagnostic criteria for FAS, partial FAS and ARND are listed in Table 9. These criteria are more stringent than those published previously as they require evidence of impairment of three or more CNS domains (E. Elliott & Peadon, 2009).

Table 9 Canadian Diagnostic Criteria for FAS, partial FAS and ARND

The criteria for the diagnosis of fetal alcohol syndrome, after excluding other diagnoses, are: A. Evidence of prenatal or postnatal growth impairment, as in at least 1 of the following: a. Birth weight or birth length at or below the 10th percentile for gestational age. b. Height or weight at or below the 10th percentile for age. c. Disproportionately low weight-to-height ratio (= 10th percentile). B. Simultaneous presentation of all 3 of the following facial anomalies at any age: a. Short palpebral fissure length (2 or more standard deviations below the mean). b. Smooth or flattened philtrum (rank 4 or 5 on the lip-philtrum guide). c. Thin upper lip (rank 4 or 5 on the lip-philtrum guide). C. Evidence of impairment in 3 or more of the following central nervous system domains: hard and soft neurologic signs; brain structure; cognition; communication; academic achievement; memory; executive functioning and abstract reasoning; attention deficit/hyperactivity; adaptive behaviour, social skills, social communication. D. Confirmed (or unconfirmed) maternal alcohol exposure.
The diagnostic criteria for partial fetal alcohol syndrome, after excluding other diagnoses, are: A. Simultaneous presentation of 2 of the following facial anomalies at any age: a. Short palpebral fissure length (2 or more standard deviations below the mean). b. Smooth or flattened philtrum (rank 4 or 5 on the lip-philtrum guide). c. Thin upper lip (rank 4 or 5 on the lip-philtrum guide). B. Evidence of impairment in 3 or more of the following central nervous system domains: hard and soft neurologic signs; brain structure; cognition; communication; academic achievement; memory; executive functioning and abstract reasoning; attention deficit/hyperactivity; adaptive behaviour, social skills, social communication. C. Confirmed maternal alcohol exposure.

The diagnostic criteria for alcohol-related neurodevelopmental disorder, after excluding other diagnoses, are: A. Evidence of impairment in 3 or more of the following central nervous system domains: hard and soft neurologic signs; brain structure; cognition; communication; academic achievement; memory; executive functioning and abstract reasoning; attention deficit/hyperactivity; adaptive behaviour, social skills, social communication. B. Confirmed maternal alcohol exposure.
Alcohol-related birth defects (ARBD) The term ARBD should not be used as an umbrella or diagnostic term, for the spectrum of alcohol effects. ARBD constitutes a list of congenital anomalies, including malformations and dysplasias and should be used with caution.

(Chudley, et al., 2005)

The FASD: Canadian Guidelines for Diagnosis (Chudley, et al., 2005) and The 4-Digit Diagnostic Code (S J Astley, 2004) both cover the full spectrum of diagnostic outcomes for FASD and require that abnormal measurements be two or more standard deviations below the mean (S. J. Astley, 2006). In addition, the guidelines recommend "harmonisation" of the Institute of Medicine Diagnostic Criteria for FASD and the 4-Digit Diagnostic Code approaches. It is suggested that the 4-Digit Diagnostic Code be used to describe, assess and measure alcohol exposure, growth, characteristic facial features and brain damage and the Institute of Medicine terminology to describe the diagnosis (Chudley, et al., 2005).

FAS: GUIDELINES FOR REFERRAL AND DIAGNOSIS

A committee of experts was convened by the Centers for Disease Control and Prevention (CDC) to develop guidelines for the diagnosis of FAS and other disorders resulting from prenatal alcohol exposure (National Center on Birth Defects and Developmental Disabilities, 2004). The Scientific Working Group comprised professionals with expertise in FAS research, diagnosis and treatment. Criteria were developed for FAS only, as the committee decided there was a lack of evidence to support the development of reliable diagnostic criteria for the rest of the FASD spectrum (E. Elliott & Peadon, 2009; National Center on Birth Defects and Developmental Disabilities, 2004). The working group sought to harmonise the guidelines with other diagnostic systems and provide standard diagnostic criteria for FAS to allow for consistency in diagnosis. The guidelines highlight a need for further research to develop criteria for the other disorders within the FASD spectrum (National Center on Birth Defects and Developmental Disabilities, 2004).

The Scientific Working Group developed a framework for FAS diagnosis and services, to inform the development of the guidelines. The framework identified various sources and settings from which an individual could be identified as having a potential problem (parents, teachers, schools, social service professionals, social workers, foster care agencies and health care providers such as paediatricians). A

referral is made when one or more features of FAS are identified, such as developmental problems, facial abnormalities, growth delay and maternal alcohol use. Once referred, it is recommended that further assessment be completed by a multidisciplinary team to confirm the diagnosis. A confirmed FAS diagnosis would be followed by an intervention plan and support from appropriate services. Individuals who do not meet the FAS referral criteria should be monitored for changes in health over time (National Center on Birth Defects and Developmental Disabilities, 2004). A summary of the CDC diagnostic criteria is presented in Table 10.

Table 10 Summary of CDC Diagnostic Criteria for FAS

Facial dysmorphia

Based on racial norms, individual exhibits all three characteristic facial features:

- Smooth philtrum (University of Washington Lip-Philtrum Guide rank 4 or 5)
- Thin vermillion border (University of Washington Lip-Philtrum Guide rank 4 or 5)
- Small palpebral fissures – measured as $\leq 10^{th}$ percentile

Growth problems

Confirmed prenatal or postnatal height or weight, or both, at or below the 10^{th} percentile, documented at any one point in time (adjusted for age, sex, gestational age, and race or ethnicity).

Central Nervous System abnormalities

I. Structural

1) Head circumference (OFC) at or below the 10^{th} percentile adjusted for age and sex.
2) Clinically significant brain abnormalities observable through imaging.

II. Neurological

Neurological problems not due to a postnatal insult or fever, or other soft neurological signs outside normal limits.

III. Functional

Performance substantially below that expected for an individual's age, schooling, or circumstances, as evidenced by:

1) Global cognitive or intellectual deficits representing multiple domains of deficit (or significant developmental delay in younger children) with performance below the 3^{rd} percentile (2 standard deviations below the mean for standardised testing)

or

2) Functional deficits below the 16^{th} percentile (1 standard deviation below the mean for standardised testing) in at least three of the following domains:

a) cognitive or developmental deficits or discrepancies
b) executive functioning deficits
c) motor functioning delays
d) problems with attention hyperactivity
e) social skills
f) other, such as sensory problems, pragmatic language problems, memory deficits, etc.

Maternal Alcohol Exposure

I. Confirmed prenatal alcohol exposure

II. Unknown prenatal alcohol exposure

Criteria for diagnosis

Requires all three of the following:

1. Documentation of all three facial abnormalities (smooth philtrum, thin vermilion border, small palpebral fissures)
2. Documentation of growth deficits
3. Documentation of CNS abnormality

(National Center on Birth Defects and Developmental Disabilities, 2004)

The Scientific Working Group recommend an individual be referred for a full FAS evaluation if there is a confirmed, high risk level of prenatal alcohol use (seven or more drinks per week or three or more drinks on multiple occasions, or both) (National Center on Birth Defects and Developmental Disabilities, 2004). This requires documentation of alcohol consumption patterns from clinical observation, self-report, reports from a reliable informant, medical records, or other social, legal or medical problems related to drinking during the pregnancy (Bertrand, et al., 2005). The guidelines acknowledge the difficulty in identifying prenatal alcohol use if a child is in foster or adoptive care, or if a mother is feeling threatened by possible stigmatisation attached with alcohol use during pregnancy (Bertrand, et al., 2005). If prenatal alcohol exposure is unknown, an individual should be referred if there is any report of concern by a parent or caregiver; when all three characteristic facial features are present; one or more facial features are present and there are deficits in height or weight or both; one or more facial features are present in addition to one or more CNS abnormalities; or one or more facial features are present along with growth deficits and one or more CNS abnormalities (National Center on Birth Defects and Developmental Disabilities, 2004). The guidelines identify a number of stages at which a differential diagnosis should be considered, including when assessing for dysmorphic features, growth retardation, and CNS abnormalities (National Center on Birth Defects and Developmental Disabilities, 2004).

A report by Bertrand and colleagues (2005) summaries the guidelines developed by the Scientific Working Group. In addition to the criteria presented in Table 8, the report suggests that prenatal alcohol exposure should be considered for people experiencing, or who have experienced one or more of the following:

- premature maternal death related to alcohol use (either disease or trauma)
- living with an alcoholic parent
- current or previous abuse or neglect
- current or previous involvement with child protective service agencies

- a history of transient care giving situations
- foster or adoptive placements (including kinship care)

(Bertrand, et al., 2005)

ALBERTA PARTNERSHIP GUIDELINE FOR THE DIAGNOSIS OF FAS

This guideline for the diagnosis of FAS was developed by a working group and included information from a province-wide survey of physicians (Alberta Partnership on Fetal Alcohol Syndrome, 2003). The guideline was intended to assist healthcare professionals to recognise disorders associated with fetal alcohol exposure; promote early and accurate diagnoses; prevent further disabilities through early diagnosis; and prevent the occurrence of FAS in future children of affected families by providing support (Alberta Partnership on Fetal Alcohol Syndrome, 2003).

The guideline follows the Institute of Medicine Diagnostic Criteria for FASD (Stratton, et al., 1996) and comprises recommendations for referring and diagnosing individuals with FAS:

1) A standard diagnosis should include a history of maternal alcohol consumption; prenatal and/or postnatal growth retardation; neurodevelopmental and behavioural characteristics and characteristic facial features.
2) Primary care providers should refer any individual suspected to have FAS to an appropriate specialist for further assessment.
3) Following a positive diagnosis, intervention measures with a multidisciplinary team can improve the outcome for an individual and information and support should be provided to the family or caregivers.

Further guidance is documented for establishing history of alcohol consumption during pregnancy, identifying physical and neurological features and characteristics, neurodevelopmental and behavioural characteristics and applying a differential diagnosis (Alberta Partnership on Fetal Alcohol Syndrome, 2003).

SCREENING METHODS

Well conducted, rigorous research that has evaluated or refined screening or diagnostic elements of FASD have been included in this review to provide further context to existing screening and diagnostic criteria, guidelines and instruments.

Goh and colleagues (2008) explain that measuring growth deficiencies as a screening tool for FASD is only useful when used in combination with other screening methods (Goh et al., 2008). The Wisconsin Fetal Alcohol Syndrome Screening Project tested a multi-source, multi-stage, active case ascertainment screening approach to promote earlier identification of children with FAS (Weiss et al., 2004). The initial study population (n=56,247) were infants born in 1998 and 1999 from 22 hospitals to mothers from urban, suburban and rural households in Wisconsin. Growth measurements were used to screen infants for FAS. Information from birth records were used to identify cases that were small for gestational age (birth weight below the 10th percentile) adjusted for gestational age and gender (n=3,291). The 10th percentile cut-offs were adjusted at 200g less to allow for expected lower birth weight among African American infants. The remaining study population of 3,291 were measured for birth head circumference less than gestational age-specific 10th percentile (n=615). The next stage of screening was carried out by graduate nursing students when the patient population had reached 2 to 3 years of age and included measurements of growth (weight, height and head circumference), facial features and development. One hundred and seventy seven infants participated in this screen, with the remaining 438 lost to follow-up. Further assessment was carried out applying the Institute of Medicine Diagnostic Criteria and 13 children met the criteria for a FAS diagnosis. However, children with normal weight or head circumference at birth, but with subsequent below normal growth patterns were not identified through this screening and a large number of infants were lost to follow-up. The high loss to follow-up children were those at greater risk for FAS and, growth impairment and developmental delays (Weiss, et al., 2004). The paper recommends this simple screening approach to initiate surveillance for children at risk for FAS and suggest that better documentation of prenatal alcohol exposure and FAS characteristics at birth will result in less under-reporting and promote early identification (Weiss, et al., 2004).

In addition to identifying children with FAS, collecting growth data helps to identify growth norms and standardise observations (May et al., 2007). A study conducted in the Western Cape Province of South Africa applied a two-tiered population-based screening method to students in their first year of school (n=1013) to identify growth norms for this particular population, standardise observations and set cut-off criteria for future screening, and to identify children with FAS and PFAS. The first tier of screening measured height, weight and head circumference (May, et al., 2007). If head circumference and/or both height and weight were at or below the 10th percentile, the child was referred for further assessment. The second tier involved examination for facial dysmorphology, intelligence and behavioural characteristics, and maternal interviews in order to make a diagnosis. This method of

screening identified 55 cases of FAS and 18 cases of PFAS in this region with high alcohol consumption (May, et al., 2007).

CHARACTERISTIC FACIAL FEATURES

Astley and Clarren (1995) previously evaluated the use of the characteristic facial features of smooth philtrum, thin upper lip and short palpebral fissures to identify FAS. The study was conducted at the Center of Human Development and Disabilities FAS Clinic at the University of Washington. The clinic serves a racially mixed population who are diagnosed by one dysmorphologist experienced in diagnosing FAS. The study population comprised all patients aged between two months to 10 years that were evaluated at the Clinic between January 1993 and January 1995 (n=194). On the sample of 194 children from several racial backgrounds (Caucasian, African, American, American Indian, Alaskan Native, Asian, Hispanic and Mexican American) sensitivity was found to be 100% and specificity was 89% (S. J. Astley & Clarren, 1995). The authors suggested that assessment of diagnostic inter-rater agreement between trained dysmorphologists and testing in other clinic populations was needed to assess the tool's external validity. Goh and colleagues (2008) report that facial screening is non-invasive and relatively low cost, however it does not identify the majority of individuals with FASD who do not present with facial characteristics (Goh, et al., 2008).

Facial photographic screening has been used to identify the FAS facial phenotype with a photograph (S. J. Astley et al., 2002). This tool was tested in a foster care setting and performed with very high accuracy based on seven screen-positive children and 590 screen-negative children. The positive predictive value for FAS was 85.7%; the negative predictive value was 100%; sensitivity was measured at 100%; specificity at 99.8% and overall accuracy of the tool was 99.8% (S. J. Astley, et al., 2002). The authors recommended this FAS screening tool as cost-effective and accurate when used in a high risk population. Avner and colleagues (2006) validated the computer-assisted method of measuring facial characteristics with digital photographs by comparing the results with manual measurements (ruler and lip-philtrum guide). A small sample of 40 children was studied. The computer assisted method demonstrated 100% sensitivity and 64% specificity. However, when assessing children younger than four years, the bias in measurement was greater as the palpebral fissure length was underestimated. This method may be useful for identifying patients from remote areas where access to a trained physician may be limited (Avner et al., 2006).

An automated method for screening FAS using 3D facial image analysis has also been reported. In a diagnostic case-control study, Fang and colleagues (2008) found the method had good sensitivity and specificity when used within the same ethnic group (88.2% sensitivity and 100% specificity in Caucasian

Finnish population; and 91.7% sensitivity and 90% specificity in Cape Coloured population form South Africa). This technology is more expensive than other methods (Fang et al., 2008).

CENTRAL NERVOUS SYSTEM - NEUROLOGICAL

The Fetal Alcohol Diagnostic Program (FADP) is a community-based, family-focused diagnostic program in Minnesota (Lang, 2006). The FADP specified ten brain domains within the central nervous system parameters, in order to effectively evaluate, diagnose and provide appropriate intervention recommendations for clients and their families. The brain domains as identified by the Program include: achievement, adaptation, attention, cognition, executive functioning, language, memory, motor, sensory/soft neurological, and social communication (Lang, 2006).

In 2007, the Canada Northwest FASD Research Network collaborated with diagnostic clinics to recommend the best psychometric tools to use when evaluating individuals with FASD (Canada Northwest FASD Research Network, 2007a). Thirty psychologists involved in assessing individuals for FASD, from 14 clinics in the Canada Northwest region participated in the first phase to reach consensus on the psychometric tools to be used in diagnosis (Canada Northwest FASD Research Network, 2007a). In order to work towards a consensus, criteria were established to reflect a typical case scenario of FASD and included an individual aged between 4 and 18 years; with an Intelligence Quotient between 70 and 100; who speaks English adequately; has no sensory deficits; and has experience in life (Canada Northwest FASD Research Network, 2007a).

Information was collected to identify which tools were currently being used. Across the 14 clinics in this study, 16 different tools were being used to assess cognition, 19 for academic achievement, 3 for memory, 25 for attention and hyperactivity, 26 for executive functioning and 18 to assess adaptive behaviour (Canada Northwest FASD Research Network, 2007a). The working group reviewed and discussed the tools and reached an agreement on the tools that should be used in an assessment, shown in Table 11.

Table 11 Consensus on Psychometric Tools – Phase 1

Cognition	
WPPSI-III: Wechsler Preschool and Primary Scale of Intelligence Alternate: DAS: Differential Ability Scales	For age groups 4-6 years
WISC-IV: Wechsler Intelligence Scale for Children	6-16 years
WAI-III: Wechsler Adult Intelligence Scale	≥16 years

Academic achievement	
BBCS-R: Bracken Basic Concept Scale-Revised School Readiness Composite	4-6 years
Alternate: DAS: Differential Ability Scales	
Math: WIAT-II: Wechsler Individual Achievement Test	6-16 years
Reading: WIAT-II: Wechsler Individual Achievement Test	
Spelling: WIAT-II: Wechsler Individual Achievement Test	
Written Expression (story only): TOWL-3: Test of Written Language	
Alternate: WJ-R to WJ III: Woodcock-Johnson Test of Achievement	
WRAT-4: Wide Range Achievement Test	≥16 years
Alternate: WJ-R to WJ III: Woodcock-Johnson Test of Achievement	
Memory	
NEPSY Learning and Memory	4-6 years
WRAML2: Wide Range Assessment of Memory and Learning	
WRAML2: Wide Range Assessment of Memory and Learning	6-16 years
Supp: CAVLT: Children's Auditory Verbal Learning Test, OR	
CVLT-C: California Verbal Learning Test-Children's Version	
WRAML 1-2: Wide Range Assessment of Memory and learning	≥16 years
Supp: RAVLT: Rey Auditory Verbal Learning Test, OR	
CVLT-II: California Verbal Learning Test 2nd Ed	
Executive functioning and abstract reasoning	
BRIEF-P: Behaviour Inventory of Executive Function, Preschool Version	4-6 years
NEPSY: Attention and Executive Functioning	
NEPSY II: Second edition for ≤6	
BRIEF: Behaviour Rating Inventory of Executive Function	6-16 years
RCFT: Rey Complex Figure Test	
WISC-IV Digit Span Backwards and Letter-Number Sequencing	
D-KEFS: Delis-Kaplan Executive Function System (≥8). Subsets: verbal, fluency, design fluency, color-word interference, sorting	
Children's Color Trials Test	
WRAML-2 Verbal and Symbolic Working Memory	
BRIEF: Behavior Rating Inventory of Executive Function	≥16 years
RCFT: Rey Complex Figure Test	
WISC-IV Digit Span Backwards and Letter-Number Sequencing	
DKEFS: Delis-Kaplan Executive Function System (≥8). Subsets: verbal, fluency, design fluency, color-word interference, sorting	
Color Trails Test	
WRAML-2 Verbal and Symbolic Working Memory	

Attention and hyperactivity BASC-2: Behavior Assessment System for Children	 4-6 years, 6-16 years, ≥16 years
Adaptive behavior ABAS-II Adaptive Behavior Assessment System VABS-II Vineland Adaptive Behavior Scale Choice of measure depends on situation: - limitation of time - parent literacy - age of child - need for an interview rather than a questionnaire ABAS Adaptive Behavior Assessment System VABS Vineland Adaptive Behavior Scale Choice of measure depends on situation: - limitation of time - parent literacy - age of child - need for an interview rather than a questionnaire	 4-6 years 6-16 years, ≥16 years

(Canada Northwest FASD Research Network, 2007b)

The second phase of the collaboration involved a group of 45 specialists comprising speech and language pathologists, occupational therapists and paediatricians who were involved in assessing individuals with FASD (Canada Northwest FASD Research Network, 2007b). The participants were representing 21 clinics from five different provinces and territories. A survey was used to collect information about the tools currently being used in the clinics. The criteria applied in the first phase were also used in this second phase. These tools were discussed and consensus was sought on the best tools to use in an assessment (Table 12).

Table 12 Consensus on Psychometric Tools – Phase 2

Psychometric Tools for Neurological Signs (Sensory Motor) — Occupational Therapists The Peabody Developmental Motor Scales–Second Edition (PDMS-2)	 4-11 years Selected as the most age appropriate motor-based assessment providing information on visual motor integration, manual dexterity and gross motor skills.
The Bruininks-Oseretsky Test of Motor Proficiency–Second Edition (BOT-2) (full form) BOT-2 (short form)	5-18 years Most appropriate overall motor assessment providing information on both fine and gross motor skills Also considered an acceptable option for those situations where time constraints or the client's attention span might limit

	the amount of time allotted to complete the assessment.
Short Sensory Profile (SSP) Sensory Profile Caregiver Questionnaire could be used for supplementary information	4 years
Short Sensory Profile Adult Adolescent Self-Questionnaire, with the option of having a caregiver assist the adolescent when filling out the questionnaire	5-10 years 11/12 – 18 years
Psychometric Tools for Communication: Receptive and Expressive – Speech—Language Pathologists Core Language: CELF P-2 Narrative Language (Bus Story) Expressive language: PLAI-II Receptive language: CELF P-2 Pragmatics checklist	4-6 years
CELF-4, TNL TOPS-2Elementary Pragmatics Profile	6-11 years
CELF-4 for core language, receptive and expressive TOPS – Adolescent or TOPS Elementary, Pragmatics Profile of CELF-4 and Word Definitions of CELF-4 for 12 year olds CASL for inferred and non-literal language	12+ years
Supplemental subtest TOWK was considered helpful for multiple contexts Frog Story – Mental State Reasoning	
Physician-Administered Measures At the time of this report there was no standard battery of tests designed for use by paediatricians. The group provided a list of the various tasks and associated median timelines that were part of paediatrician role in FASD diagnosis. Six paediatricians conferred and detailed the physician role, goals for the medical examination and specific tasks and tools recommended for the achievement of goals. They outlined the tasks and tools as: – History analysis – Current function of the child and how this has changed over time is obtained by past and present documentation from school and caregivers. The tool could be the Caregiver Interview from the DPN Manual – Health determinants that impact development and function – Physical exam – Mental status – Formulation of diagnosis – Development of intervention strategies and support systems after diagnosis – Longitudinal follow up	

(Canada Northwest FASD Research Network, 2007b)

DEVELOPMENTAL DELAY

Individuals with FASD may present with symptoms of developmental delay in early childhood. There are several screening instruments which detect developmental delay (Hamilton, 2006). Two screening instruments are based on parental report and have been used within Australia, the PEDS and the Ages and Stages Questionnaire (Hamilton, 2006). In addition other screening instruments include direct observation of the child, such as the Brigance and the Batelle Developmental Inventory Screening test (Hamilton, 2006). However, the screening instruments do not point to the underlying cause of developmental delay and further assessment is needed to differentiate FASD from other developmental disabilities.

CENTRAL NERVOUS SYSTEM - STRUCTURAL

A review of structural anomalies by brain region conducted by Spadoni and colleagues (2007) confirmed that brain-imaging techniques could be used to identify structural damage to the brain caused by prenatal alcohol exposure. Reduced brain size, damage to the corpus callosum, cerebellar vermis, basal ganglia and orbito-frontal and parietal brain regions were identified among FASD individuals (Spadoni et al., 2007).

Magnetic resonance technology can be used as a non-invasive method for assessment of neuroabnormalities (S. J. Astley et al., 2009b). A comprehensive magnetic resonance study of children with FASD was conducted by Astley and colleagues (2009) to assess whether participants with FASD demonstrated impaired working memory as measured by performance on the *N-back* assessment of working memory, and functional Magnetic Resonance Imaging scanning of targeted brain regions. The study was conducted with 65 children aged 8-15.9 years who had been diagnosed with either FAS or PFAS, Static encephalopathy-alcohol exposed, or Neurobehavioral disorder-alcohol exposed at the Washington State FAS DPN clinic (Susan J. Astley et al., 2009). A group of 16 healthy controls with no antenatal alcohol exposure were matched for age, gender and ethnicity. The study provided evidence that children across the FASD spectrum show significant working memory deficits that are correlated with abnormalities in activation in brain areas and that functional Magnetic Resonance Imaging is developing as an important assessment tool to consider in FASD evaluation (Susan J. Astley, et al., 2009).

Sowell and colleagues (2008) evaluated white matter integrity in individuals with FASD using a combination of diffusion tensor and T1-weighted Magnetic Resonance Imaging. Seventeen subjects with FASD, aged 7 to 15 years were compared with 19 age and gender matched controls. An experienced clinician examined the cases, and the controls were screened for neurological impairments, psychiatric illness, and history of learning disability or developmental delay. Lower fractional anisotropy (FA) was observed in individuals with FASD relative to controls in the right temporal lobe, bilaterally in the posterior cingulate and in regions of the lateral splenium of the corpus callosum. The results suggest that this region of white matter is particularly susceptible to damage from prenatal alcohol exposure and that disruption of splenial fibres in this group in associated with poorer visuomotor integration. It was reported that the left hemisphere may also be related to disorganisation of reduced myelin, but not to the same extent as found in the right hemisphere (Sowell et al., 2008).

MECONIUM

Meconium is the first faecal matter of a new-born and is only available for testing within approximately 72 hours after birth (Goh & Rosenbaum, n.d.). Fatty Acid Ethyl Esters (FAEE) are a by-product of alcohol that can be found in meconium (Canadian Association of Paediatric Health Centres, 2010). It is considered a non-invasive and easy method of screening for maternal alcohol use (Goh & Rosenbaum, n.d.), that can improve early diagnosis, early intervention and identification of high risk pregnancies (Canadian Association of Paediatric Health Centres, 2010). However, there is only a limited amount of time in which meconium can be collected and it is only able to detect prenatal alcohol exposure in the second and third trimesters of pregnancy (Goh & Rosenbaum, n.d.). Further to this, it is a relatively expensive test unless it is used at a wider-scale level (Canadian Association of Paediatric Health Centres, 2010).

Gifford and colleagues (2010) conducted a review to assess the benefits of universal meconium screening for prenatal alcohol exposure. The literature search was conducted using online databases. In order to determine cost-effectiveness, monetary values were recalculated from their value in US$ for 2006 using consumer price index inflation estimates. Estimates included administrative costs, materials and cost of professional analysis of the test. State regulated new-born screening programs in the US require lab tests and a physician-parent reporting process, which means that administrative and reporting costs of adding a meconium screening are approximately 20% of the cost of the program for additional facilities, transport, personnel training and specimen collection (Gifford et al., 2010). A number of data gaps have been identified by the authors that affect ability to accurately estimate costs and benefits of universal screening, these include: cost of including meconium screening in established new-born screening systems; number of women who would voluntarily participate in interventions;

long-term effectiveness of each intervention; development of a second-stage screen that reduces false positives; impact of gestational age on the sensitivity/specificity of the meconium test; refinement of test to indicate level of drinking; ability to identify social drinkers; inclusion of FASD children into primary research; relation between binge drinking during pregnancy and alcohol dependence; strategy to reduce false negatives; cross-country generalizability; and effect of multiple drug use on effectiveness of intervention (Gifford, et al., 2010). The paper concludes that universal meconium analysis of new-borns and subsequent intervention for identified mothers could be cost-effective in reducing the incidence of FASD. Estimated savings, dependant on the type of intervention, were calculated to range from $97 to $6 per every dollar spent. These estimated savings are conservative and do not consider the long-term benefits such as reduced secondary disability as a result of early diagnosis and increased quality of life. The review notes that further thought should be made to consider the possibility of separation of mother and child as a result of positive test results (Gifford, et al., 2010).

SUMMARY OF FINDINGS

The FASD Collaboration recognises the importance and significance of prenatal screening; however the scope of this review was to identify literature on postnatal screening and diagnostic guidelines and instruments to inform the development of a postnatal screening and diagnostic instrument for Australia. A number of papers recommend population-based screening should be implemented (Department of Health Western Australia, 2010; Gifford, et al., 2010; Weiss, et al., 2004) along with targeted screening (for example, children in foster care or in the justice system) (Conry & Asante, 2010; Goh, et al., 2008) provided there are appropriate services for referral and follow-up (Canadian Association of Paediatric Health Centres, 2010; Gifford, et al., 2010; Weiss, et al., 2004), to better reach individuals with undiagnosed disorders within the FASD spectrum.

The literature search did not locate any standardised screening or diagnostic instruments developed in Australia. This review identified a number of screening and diagnostic instruments that have been developed in other countries, including the Institute of Medicine Diagnostic Criteria, the University of Washington 4-Digit Diagnostic Code for FASD, the Hoyme Updated Institute of Medicine Criteria, the Canadian Guidelines for Diagnosis and the Centers for Disease Control Guidelines for Referral and Diagnosis of FAS. Criteria for FAS and partial FAS are well defined but the complexity of outcomes associated with fetal alcohol exposure makes it difficult to accurately define other categories of disorders within the spectrum. The key criteria for a FAS diagnosis are relatively consistent: prenatal alcohol exposure, growth abnormalities, characteristic facial features and neurological, functional and structural 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 continuum.

A number of research papers identified in the systematic search were relevant for inclusion and 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 racial norms of facial characteristics to be identified for Australia. Brain domains within the central nervous system parameters were identified for effectively 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 child has, so that tailored and specific treatment and support can be provided.

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APPENDICES FOR THE FULL LITERATURE REVIEW

Keyword Search Matrix – online databases (update)

2008 – 30 September 2010

This search is a continuation of that completed by Elliott et al (2008) – using the same search terms and databases from January 2008 to 30 September 2010.

Database	Date searched	Search #	Search terms	Citations
EMBASE	Jan 2008 – 30 Sep 2010	1	('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR ('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR 'fetal alcohol syndrome' OR 'fetal alcohol spectrum disorder' OR 'fetal alcohol spectrum disorder' OR fasd	627
		2	('meta analysis'/exp OR 'meta analysis') OR ('systematic review'/exp OR 'systematic review') OR 'pooled analysis' OR ('review'/exp OR 'review') OR ('meta analysis'/exp OR 'meta analysis') OR systemat* OR pool*	354,743
		3	#1 AND #2	**129** 1 duplicate 3 for further review
MEDLINE	Jan 2008 – 30 Sep 2010	1	('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR ('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR 'fetal alcohol syndrome' OR 'fetal alcohol spectrum disorder' OR 'fetal alcohol spectrum disorder' OR fasd	364
		2	('meta analysis'/exp OR 'meta analysis') OR ('systematic review'/exp OR 'systematic review') OR 'pooled analysis' OR ('review'/exp OR 'review') OR ('meta analysis'/exp OR 'meta analysis') OR systemat* OR pool*	264,096
		3	#1 AND #2	**73** 2 duplicates 7 for further review
Cochrane Library	Jan 2008 –	1	fetal alcohol spectrum disorder OR fetal alcohol spectrum disorder	**25**

	30 Sep 2010		OR fetal alcohol syndrome OR fetal alcohol syndrome	2 for further review
Health Technology Assessment Database	Jan 2008 – 30 Sep 2010	1	'fetal alcohol spectrum disorder' OR 'fetal alcohol syndrome'	**5** 0 articles retained
Total citations identified				14
Total citations after removal of duplicates				9
Total citations deemed relevant for review by Lead Investigator				**8**

Keyword Search Matrix – online databases (AD additional databases)

Inception of database – 30 September 2010

This search is using the same search terms that were used by Elliott et al (2008) on a number of new databases. The search will include literature from 1996 when the first diagnostic criteria for FAS was published.

Database	Date searched	Search #	Search terms	Citations
CINAHL	Jan 1980 – 30 Sep 2010	1	('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR ('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR 'fetal alcohol syndrome' OR 'fetal alcohol spectrum disorder' OR 'fetal alcohol spectrum disorder' OR fasd	765
		2	('meta analysis'/exp OR 'meta analysis') OR ('systematic review'/exp OR 'systematic review') OR 'pooled analysis' OR ('review'/exp OR 'review') OR ('meta analysis'/exp OR 'meta analysis') OR systemat* OR pool*	187638
		3	#1 AND #2	**67** 1 duplicate 5 for further review
PsycInfo	1806 – 30 Sep 2010	1	('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR ('fetal alcohol syndrome'/exp OR 'fetal alcohol syndrome') OR 'fetal alcohol syndrome' OR 'fetal alcohol spectrum disorder' OR 'fetal alcohol spectrum disorder' OR fasd	1072
		2	('meta analysis'/exp OR 'meta analysis') OR ('systematic review'/exp OR 'systematic review') OR 'pooled analysis' OR ('review'/exp OR 'review') OR ('meta analysis'/exp OR 'meta analysis') OR systemat* OR pool*	280127
		3	#1 AND #2	**154** 1 duplicate 2 for further review

Web of Science	30 Sep 2010		#1 AND #2	**213** 6 duplicates 5 for further review
DARE	1 Sept 2010		fetal alcohol spectrum disorder OR fetal alcohol spectrum disorder OR fetal alcohol syndrome OR fetal alcohol syndrome	**1** 0 articles retained
Total citations identified				20
Total citations after removal of duplicates				12
Total citations deemed relevant for review by Lead Investigator				**9**

Keyword Search Matrix – online databases (AS additional search terms)

Inception of database –30 September 2010

This search is being conducted in addition to the search completed by Elliott et al (2008). It includes additional search terms over a number of databases.

Database	Date searched	Search #	Search terms	Citations
EMBASE	1988 – 30 Sep 2010	1	('fetal alcohol effects'/exp OR 'fetal alcohol effects') OR 'fetal alcohol effects' OR 'fetal alcohol disorders' OR fae OR ('partial fetal alcohol syndrome'/exp OR 'partial fetal alcohol syndrome') OR 'partial fetal alcohol syndrome' OR 'prenatal alcohol exposure' OR pfas	1374
		2	('alcohol related birth defects'/exp OR 'alcohol related birth defects) OR 'alcohol related birth defects' OR 'prenatal alcohol exposure' OR arbd OR ('alcohol related neuro developmental disorder'/exp OR 'alcohol related neuro developmental disorder) OR 'alcohol related neuro developmental disorder' OR arnd	811
		3	#1 OR #2	1456
		4	screen$ OR diagnos$	1999671
		5	#3 AND #4	**201** 13 duplicates 24 for further review
MEDLINE	Inception – 30 Sep 2010	1	('fetal alcohol effects'/exp OR 'fetal alcohol effects') OR 'fetal alcohol effects' OR 'fetal alcohol disorders' OR fae OR ('partial fetal alcohol syndrome'/exp OR 'partial fetal alcohol syndrome') OR 'partial fetal alcohol syndrome' OR 'prenatal alcohol exposure' OR pfas	1314
		2	('alcohol related birth defects'/exp OR 'alcohol related birth defects) OR 'alcohol related birth defects' OR 'prenatal alcohol exposure' OR arbd OR ('alcohol related neuro developmental disorder'/exp OR 'alcohol related neuro developmental disorder) OR 'alcohol related	726

			neuro developmental disorder' OR arnd	
		3	#1 OR #2	1395
		4	screen$ OR diagnos$	1834389
		5	#3 AND #4	**238** 25 duplicates 14 for further review
Database	**Date searched**	**Search #**	**Search terms**	**Citations**
CINAHL	Inception – 30 Sep 2010	1	('fetal alcohol effects'/exp OR 'fetal alcohol effects') OR 'fetal alcohol effects' OR 'fetal alcohol disorders' OR fae OR ('partial fetal alcohol syndrome'/exp OR 'partial fetal alcohol syndrome') OR 'partial fetal alcohol syndrome' OR 'prenatal alcohol exposure' OR pfas	272
		2	('alcohol related birth defects'/exp OR 'alcohol related birth defects) OR 'alcohol related birth defects' OR 'prenatal alcohol exposure' OR arbd OR ('alcohol related neuro developmental disorder'/exp OR 'alcohol related neuro developmental disorder) OR 'alcohol related neuro developmental disorder' OR arnd	167
		3	#1 OR #2	288
		4	screen$ OR diagnos$	5791
		5	#3 AND #4	**4** 0 retained
PsycInfo	1806 – 30 Sep 2010	1	('fetal alcohol effects'/exp OR 'fetal alcohol effects') OR 'fetal alcohol effects' OR 'fetal alcohol disorders' OR fae OR ('partial fetal alcohol syndrome'/exp OR 'partial fetal alcohol syndrome') OR 'partial fetal alcohol syndrome' OR 'prenatal alcohol exposure' OR pfas	694
		2	('alcohol related birth defects'/exp OR 'alcohol related birth defects) OR 'alcohol related birth defects' OR 'prenatal alcohol exposure' OR arbd OR ('alcohol related neuro developmental disorder'/exp OR 'alcohol related neuro developmental disorder) OR 'alcohol related	548

			neuro developmental disorder' OR arnd	
		3	#1 OR #2	730
		4	screen$ OR diagnos$	223952
		5	#3 AND #4	**148** 19 duplicates 6 for further review
DARE	15 Sep 2010	1	fetal alcohol effects OR partial fetal alcohol syndrome OR alcohol related birth defects OR alcohol related neuro developmental disorder	**3** 0 retained
Cochrane Library	14 Sep 2010		fetal alcohol effects OR partial fetal alcohol syndrome OR alcohol related birth defects OR alcohol related neuro developmental disorder	**32** 1 duplicate 0 retained
Health Technology Assessment Database	15 Sep 2010		'fetal alcohol effects' OR 'partial fetal alcohol syndrome' OR 'alcohol related birth defects' OR 'alcohol related neuro developmental disorder'	0 0 retained
Web of Science	30 Sep 2010		Topic=(('fetal alcohol effects'/exp OR 'fetal alcohol effects') OR 'fetal alcohol effects' OR 'fetal alcohol disorders' OR fae OR ('partial fetal alcohol syndrome'/exp OR 'partial fetal alcohol syndrome') OR 'partial fetal alcohol syndrome' OR 'prenatal alcohol exposure' OR pfas OR ('alcohol related birth defects'/exp OR 'alcohol related birth defects) OR 'alcohol related birth defects' OR 'prenatal alcohol exposure' OR arbd OR ('alcohol related neuro developmental disorder'/exp OR 'alcohol related neuro developmental disorder) OR 'alcohol related neuro developmental disorder' OR arnd) AND Topic=(screen$ OR diagnos$) Refined by: Document Type=(ARTICLE OR REVIEW) AND Languages=(ENGLISH)	**52** 6 duplicates 1 for further review
Total citations identified				109
Total citations after removal of duplicates				45
Total citations deemed relevant for review by Lead Investigator				**42**

Keyword Search Matrix - Organisations

Search terms: Fetal alcohol spectrum disorders OR fetal alcohol syndrome OR fetal alcohol effects OR partial fetal alcohol syndrome OR alcohol related birth defects OR alcohol related neuro-developmental disorder

Source	Date searched	Citations
National Institute for Health and Clinical Excellence (NICE)	15 Sept 2010	0
World Health Organization (WHO)	16 Sept 2010	1 for review
Centers for Disease Control and Prevention (CDC)	16 Sept 2010	2 for review
Research Society on Alcoholism	16 Sept 2010	0
The Fetal Alcohol Spectrum Disorders Center for Excellence	16 Sept 2010	2 duplicates
FAS Diagnostic and Prevention Network http://depts.washington.edu/fasdpn/index.htm	16 Sept 2010	15 duplicates
The University of Washington Fetal Alcohol & Drug Unit	16 Sept 2010	2 duplicates
The Canadian Centre on Substance Abuse (CCSA)	17 Sept	1 duplicate
Canadian Foundation on Fetal Alcohol Research (CCFAR)	17 Sept	0
Canadian Association of Paediatric Hospitals (CAPH)	17 Sept	2 duplicates
Public Health Agency of Canada (PHAC)	17 Sept	2 duplicates
Canadian NW Research Network	17 Sept	1 for review 1 duplicate
Canadian NW FASD Partnership	17 Sept	0
Asante Centre	17 Sept	2 for review
Motherisk	17 Sept	1 duplicate
Interagency Coordinating Committee on Fetal Alcohol Spectrum Disorders (ICCFASD)	17 Sept	0
NeuroDevNet.ca	17 Sept	0
National Institute on Alcohol Abuse and Alcoholism	16 Sept	0
National Health and Medical Research Council (NHMRC)	17 Sept	0
Drug and Alcohol Office (DAO)	17 Sept	0
Department of Health and Ageing	17 Sept	1 for review
Department of Health – WA, VIC, NSW, ACT, TAS, QLD, NT		2 for review
http://www.nofas.org/resource/directory.aspx	17 Sept	0

Institute of Health Economics	20 Sept	1 for review
NOFASARD	16 Sept	1 for review 3 duplicates
FAS Link	20 Sept	1 for review
Total citations deemed relevant for review by Lead Investigator	TOTAL	12

Number of papers identified from reference list s = 24

Number of papers identified through key informants = 15

Appendix 2 T-ACE and TWEAK Screening Questionnaires (Full Literature Review)

T-ACE Screening Questionnaire

1. Tolerance (T) – how many drinks does it take to make you feel high?
2. Annoyance (A) – have people annoyed you by criticising your drinking?
3. Cut down (C) – have you ever felt you ought to cut down on your drinking?
4. Eye-opener (E) – have you ever had a drink first thing in the morning to steady your nerves or get rid of a hangover?

A single point is given for an affirmative answer to the A, C and E questions, and two points are given when a pregnant woman indicates a tolerance of more than two drinks to feel high. A total score of two or more on the test is suggestive of harmful drinking patterns during pregnancy.

TWEAK Screening Questionnaire

TWEAK for populations with high levels of binge drinking:

1. Tolerance (T) – how many drinks does it take before the alcohol makes you fall asleep or pas out?

Record number of drinks__(a positive score is six or more drinks) OR
If you never drink until you pass out, what is the largest number of drinks that you have?

Record number of drinks__(a positive score is six or more drinks)

2. Worried (W) – have your friends or relatives worried or complained about your drinking in the past year?
3. Eye opener (E) – do you sometimes take a drink in the morning when you first get up?
4. Amnesia (A) – are there times when you drink and you can't remember what you said or did?
5. Cut down (K) – do you sometimes feel the need to cut down on your drinking?

TWEAK for populations with low levels of binge drinking:

1. Tolerance (T) – how many drinks does it take before you begin to feel the first effects of alcohol?

Record number of drinks__(a positive score is three or more drinks)

2. Worried (W) – have your friends or relatives worried or complained about your drinking in the past year?
3. Eye opener (E) – do you sometimes take a drink in the morning when you first get up?
4. Amnesia (A) – are there times when you drink and you can't remember what you said or did?
5. Cut down (K) – do you sometimes feel the need to cut down on your drinking?

For each version, a positive response to question T or W yields two points each, and an affirmative reply to question E, A or K scores one point each. A total score of two or more points on the TWEAK test is suggestive of harmful drinking patterns during pregnancy.

Appendix 3 Data Extraction Form Templates (Full Literature Review)

DATA EXTRACTION FORM - Research

Citation	Full reference
Publication status	Published, unpublished journal, report, webpage, other
Source	Database, internet, key informant, other
Country of origin/region	
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	I = a systematic review of level II studies II = a study of test accuracy with: an independent, blinded comparison with a valid reference standard, among consecutive persons with a defined clinical presentation III-1 = a study of accuracy with: an independent, blinded comparison with a valid reference standard, among non-consecutive persons with a defined clinical presentation III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence III-3 = diagnostic case-control study IV = study of diagnostic yield (no reference standard)
Study type/design	Systematic review, study of test accuracy, comparison, diagnostic case-control, diagnostic yield
Research question/aim	
Intervention/Instrument (Elliott et al., 2008)	Any strategy that aims to screen or diagnose an individual at risk of a diagnosis within the FASD continuum
Instruments mentioned	
Patient population (Elliott et al., 2008)	Individuals who may have a diagnosis within the FASD continuum
Comparator (Elliott et al., 2008)	Any comparator
Outcomes/ Recommendations (Elliott et al., 2008)	Sensitivity and specificity of screening and/or diagnosis
Evaluation	Process, impact, outcome
Notes	Any other information deemed relevant for inclusion in the review

DATA EXTRACTION FORM – Review

Citation	Full reference
Publication status	Published, unpublished journal, report, webpage, other
Source	Database, internet, key informant, other
Country of origin/region	Country
Study type/design	Review
Research question/aim	
Interventions/Instruments (Elliott et al., 2008)	Any strategy that aims to screen or diagnose an individual at risk of a diagnosis within the FASD continuum
Diagnostic Criteria	Criteria
Patient population/s (Elliott et al., 2008)	Individuals who may have a diagnosis within the FASD continuum
Outcomes/ Recommendations (Elliott et al., 2008)	
Notes	Any other information deemed relevant for inclusion in the review

Appendix 4 Data Extraction Forms: Postnatal Screening and Diagnosis (Full Literature Review)

Citation	**(Stratton et al., 1996)** **Stratton K, Howe C, Battaglia F. Fetal alcohol syndrome: diagnosis, epidemiology, prevention, and treatment. Washington: Institute of Medicine and National Academy Press; 1996.**
Publication status	Published Book
Source	Elliott Review
Country of origin/region	USA
Instrument/s	Diagnostic criteria for FAS and Alcohol-Related Effects
Diagnostic Criteria	FAS: • Maternal confirmed alcohol exposure • FAS without confirmed maternal alcohol exposure • Partial FAS with confirmed maternal alcohol exposure Alcohol-Related Effects (history of maternal alcohol exposure) • Alcohol-related birth defects • Alcohol-related neurodevelopmental disorder See attached JP Appendix 5 (Stratton_from_Elliott)
Outcomes/ Recommendations (Elliott et al., 2008)	Recommend research to evaluate utility, reliability, and validity of this scheme for classification and diagnosis ARBD and ARND were not intended as diagnoses for individuals – they refer to a range of abnormalities that occur in those exposed to alcohol in utero who do not have FAS. They are not appropriate for use in clinical settings.
Notes	The IOM diagnostic criteria for FAS and Alcohol-Related Effects have been included in the Elliott (2008) review

Citation	**(Manning & Eugene Hoyme, 2007)** **Manning MA, Eugene Hoyme H. Fetal alcohol spectrum disorders: A practical clinical approach to diagnosis. Neurosci Biobehav Rev. 2007;31 (2):230-8.**
Publication status	Published **(Review)** Journal
Source	Online Database
Country of origin/region	US
Instrument/s	Revised Institute of Medicine criteria for FASD
Diagnostic Criteria	Revised IOM criteria for diagnosis of FASD (Hoyme et al., 2005) I. FAS With Confirmed Maternal Alcohol Exposure (requires all features of A–D) (A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide)

	(3) Smooth philtrum (score 4 or 5 with the lip/philtrum guide) (C) Evidence of prenatal and/or postnatal growth retardation (1) Height and/or weight p10%, corrected for racial norms, if possible (D) Evidence of deficient brain growth and/or abnormal morphogenesis, including 1 or more of the following: (1) Structural brain abnormalities (2) Head circumference p10% II. FAS Without Confirmed Maternal Alcohol Exposure IB, IC, and ID as above III. Partial FAS With Confirmed Maternal Alcohol Exposure (requires all features, A-C) (A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide) (3) Smooth philtrum (score 4 or 5 with the lip/philtrum guide) (C) One of the following other characteristics: (1) Evidence of prenatal and/or postnatal growth retardation (a) Height and/or weight p10% corrected for racial norms, if possible (2) Evidence of deficient brain growth or abnormal morphogenesis, including 1 or more of the following: (a) Structural brain abnormalities (b) Head circumference p10% (3) Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background, or environment alone (a) This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behavior (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction) IV. Partial FAS Without confirmed Maternal Alcohol Exposure IIIB and IIIC, as above V. ARBD (requires all features, A-C) (A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p 10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide) (3) Smooth philtrum (score 4 or 5 with the lip/ philtrum guide) (C) Congenital structural defects in 1 or more of the following categories, including malformation and dysplasias (if the patient displays minor anomalies only, X 2 must be present): cardiac: atrial septal defects, aberrant great vessels, ventricular septal defects, conotruncal heart defects; skeletal: radioulnar synostosis, vertebral segmentation defects, large joint contractures, scoliosis; renal: aplastic/hypoplastic/dysplastic kidneys, ‘‘horseshoe’’

	kidneys/ureteral duplications; eyes: strabismus, ptosis, retinal vascular anomalies, optic nerve hypoplasia; ears: conductive hearing loss, neurosensory hearing loss; minor anomalies: hypoplastic nails, short fifth digits, clinodactyly of fifth fingers, pectus carinatum/excavatum, camptodactyly, ''hockey stick'' palmar creases, refractive errors, ''railroad track'' ears VI. ARND (requires both A and B) (A) Confirmed maternal alcohol exposure (B) At least 1 of the following: (1) Evidence of deficient brain growth or abnormal morphogenesis, including 1 or more of the following: (a) Structural brain abnormalities (b) Head circumference p10% (2) Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background, or environment alone (a) This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behaviour (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction)
Outcomes/ Recommendations (Elliott et al., 2008)	The Revised IOM Diagnostic Classification System (Hoyme et al., 2005) has an advantage over the Canadian system in that it has been tested in a large multiracial international cohort of children and found to be straightforward to use with reproducible results. In addition to stressing a multidisciplinary approach to evaluating alcohol exposed children and adults, this system also emphasizes the importance of considering the full differential diagnosis of genetic and teratogenic causes of developmental disabilities before a designation within the FASD spectrum is made.
Notes	

Citation	**(S. J. Astley et al., 2009a)** **Astley, S. J., H. C. Olson, et al. (2009). "Neuropyschological and behavioral outcomes from a comprehensive magnetic resonance study of children with fetal alcohol spectrum disorders." Canadian Journal of Clinical Pharmacology 16(1): e178-201.**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	II = a study of test accuracy with: an independent, blinded comparison with a valid reference standard, among consecutive persons with a defined clinical presentation
Study type/design	Study of test accuracy
Research question/aim	Were three distinct FASD subgroups (FAS/PFAS, Static Encephalopathy/Alcohol Exposed, Neurobehavioural Disorder/Alcohol Exposed) able to be established

	using neurobehavioural testing? How were these subgroups defined?
Intervention/Instrument (Elliott et al., 2008)	4-Digit diagnostic code Neuropsychological test battery
Instruments mentioned	4-Digit diagnostic code Neuropsychological test battery (assessments of: Soft Neurological Signs, General Intellectual Function, Academic Achievement, Visuospatial Skills, Visual Memory, and Organization, Executive Function, Verbal Memory, Attention, Receptive and Expressive Language, Behavior Problems and Social Competence, Caregiver Report of Behaviors Related to Executive Function, Psychiatric Conditions). MRI, MRS, fMRI
Patient population (Elliott et al., 2008)	65 children aged 8-15.9 years who had been diagnosed with either FAS/PFAS, Static encephalopathy-alcohol exposed, or neurobehavioural disorder-alcohol exposed at the Washington State FAS DPN clinic. 16 healthy controls with no antenatal alcohol exposure matched for age, gender and ethnicity.
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	The three subgroups (ND/AE, SE/AE and FAS/PFAS) reflected a linear continuum of increasing neuropsychological impairment and physical abnormality, representing the full continuum of FASD. Behavioral and psychiatric disorders were comparably prevalent across the three FASD groups, and significantly more prevalent than among the Controls. All three FASD subgroups had comparably high levels of prenatal alcohol exposure.
Evaluation	
Notes	
Citation	**(S J Astley, 2004)** **Astley, S. J. (2004). Diagnostic Guide for Fetal Alcohol Spectrum Disorders: The 4-Digit Diagnostic Code. Seattle, University of Washington Publication Services**
Publication status	Published Diagnostic Guide
Source	Key Informant
Country of origin/region	USA
Instrument	A diagnostic code that ranks the degree of abnormality of growth, the CNS and facial features; and antenatal alcohol exposure An objective measure with clearly defined case definitions of growth, face, CNS and alcohol exposure
Diagnostic Criteria	Criteria (1) growth deficiency, (2) the FAS facial phenotype, (3) CNS abnormalities, and (4) prenatal alcohol exposure

	Detailed diagnostic criteria are presented in 22 different clinical diagnostic categories
Notes	Highly detailed assessment strategies with specific assessment domains and case definitions Descriptions of the various different FASD diagnostic terminology is provided, with justification for the terminology used in the 4-digit diagnostic code Fetal Alcohol Syndrome Tutor™ has been created by the University of Washington FAS DPN to instruct healthcare professionals, through video, computer animation, and photographic examples, on how to screen and diagnose FASD

Citation	**(BMA Board of Science, 2007)** **BMA Board of Science. Fetal alcohol spectrum disorders: A guide for healthcare professionals: British Medical Association 2007.**
Publication status	Published Guidelines
Source	Elliott Review
Country of origin/region	UK
Instrument/s	Antenatal screening for alcohol exposure
Diagnostic Criteria	Review compares diagnosis using revised IOM, IOM and 4 digit code
Notes	The paper is really an argument for developing protocols in the UK for antenatal screening for alcohol exposure and for referral pathways for assessment and diagnosis of children with suspected FAS; it does not discuss the advantages/disadvantages of various diagnostic methods.

Citation	**(Chudley et al., 2005)** **Chudley AE, Conry J, Cook JL, Loock C, Rosales T, LeBlanc N. Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis. Can Med Assoc J. 2005 Mar;172(Suppl):S1-S21.**
Publication status	Published Journal
Source	Elliott Review
Country of origin/region	Canada
Instrument/s	The approach identified in the 4-Digit Diagnostic Code should be used to describe, assess and measure objectively alcohol exposure, growth, facial features and brain damage. The terminology in the IOM criteria should be used to describe the diagnosis. Palpebral fissures difficult to measure accurately without training and commented about callipers and plastic rulers to measure this. Because callipers are not a common tool in most medical clinics, the use of a clear flexible plastic ruler was recommended. Plot the result on an appropriate nomogram chart to determine the percentile of standard deviation for each eye.

Diagnostic Criteria	Diagnosis: harmonizing the IOM and 4-Digit Diagnostic Code. Although the approaches are different, the underlying, fundamental criteria of the IOM and the 4-Digit Diagnostic Code are similar. **Growth** should be monitored to detect deficiency – defined as at or , 10th percentile. Need to consider confound variables such as parental size, genetic potential and associated conditions. **Facial features**: the 3 characteristics facial features include: short palpebral fissures (at or below the 3rd percentile), smooth or flattened philtrum (4 or 5 on the 5 point scale Likert scale), thin vermilion border of the upper lip, 4 or 5 on the 5-point Likert scale of the lip-philtrum guide (developed by Astley and Clarren). **Neurobehavioural assessment:** Assess the following domains: i. hard and soft neurologic signs, ii. brain structure, iii. cognition, iv. communication (receptive and expressive), v. academic achievement, vi. memory, vii. executive functioning and abstract reasoning, viii. attention deficit / hyperactivity and ix. adaptive behaviour, social skills, social communication. They describe the criteria for FAS, partial FAS and ARND using the harmonization of IOM and 4-dignit Diagnostic Code. The ARBD category has limited utility in diagnosis, but recognise that alcohol is teratogenic and may be responsible for birth defects if exposure occurs during critical periods of development. Included recommendations for screening and referral
Outcomes/ Recommendations (Elliott et al., 2008)	Diagnosis: Recommended harmonizing the IOM and 4-Digit Diagnostic Code. Although the approaches are different, the underlying, fundamental criteria of the IOM and the 4-Digit Diagnostic Code are similar. Sensitivity and specificity of FASD screening and FASD diagnosis Because of the complexity and the range of expression of dysfunction related to PAE, a multi-disciplinary team is essential for an accurate and comprehensive diagnosis and treatment recommendations. The team can be geographic, regional or virtual; it can also accept referrals from distant communities and carry out an evaluation using telemedicine. Recommended team: Coordinator for case management (eg nurse, social worker), Physician specifically trained in FASD diagnosis, Psychologist, OT, Speech-language pathologist. Highlighted the importance of differential diagnosis and gave a list of 9 conditions and described the overlapping and differentiating features with FAS. A general physical and neurologic examination, including appropriate measurements of growth and head size, assessment of characteristic findings and documentation of anomalies is required to exclude the presence of other genetic disorders of multifactorial disorders that could lead to features mimicking FAS or partial FAS. Knowledge of exposure history will decrease the possibility of misdiagnosing FASD.
Notes	Because of limited capacity and expertise and the need to involve several professionals in a comprehensive multi-disciplinary diagnostic evaluation, only a fraction of those affected currently receive a diagnosis. Highlighted the role of telemedicine until regionally based diagnostic teams are established. Areas for research: Growth and facial anthropometric data are needed for the specific population, as sensitivity and specificity of the assessment will be lowered without the use of appropriate norms. More longitudinal research is needed to correlate changes in these characteristic physical findings in adolescents and adults diagnosed with FAS or partial FAS. Lip-philtrum guides were developed for use in Caucasian and African-American populations, but no standards are currently available for other populations.

Citation	**(Hoyme et al., 2005)** **Hoyme HE, May PA, Kalberg WO, Kodituwakku P, Gossage JP, Trujillo PM, et al. A practical clinical approach to diagnosis of fetal alcohol spectrum disorders: Clarification of the 1996 institute of medicine criteria. Pediatrics. 2005 Jan;115 (1):39-47.**
Publication status	Published Journal
Source	Elliott Review
Country of origin/region	US
Research question/aim	To present specific clarifications of the 1996 IOM criteria for the diagnosis for FASD, to facilitate practical application in a clinical setting
Instrument/s	Hoyme Updated Institute of Medicine Criteria 2005 - In the proposed clarifications of the IOM criteria, children with FAS (with or without confirmed maternal alcohol exposure) must have abnormalities in all domains, ie, facial dysmorphic features, growth, and grain growth or structure. In the partial FAS category (with or without confirmed maternal alcohol exposure), children must display typical facial dysmorphic features and abnormalities in 1 of the other domains (growth or CNS or function). - For the proposed clarifications of the 2 diagnoses characterised as alcohol-related effects, maternal alcohol exposure must be documented.
Patient population	- Study participants were identified with active case-ascertainment methods, from multiple sources. - American subjects were from 6 Native American communities and 1 urban population each in South and North Dakota - South African subjects were from 1 community in the wine producing region of the Western Cape - Of the 1500 children evaluated, 164 with a potential FASD diagnosis were identified, 72 Native American children and 92 South African children
Comparator	
Outcomes/ Recommendations (Elliott et al., 2008)	- There are several problems with the IOM criteria for the diagnosis of FAS and alcohol-related effects as they were formulated in 1996. They are vague with no specific parameters being set forth for diagnosis in each category; neither the degree of growth deficiency nor the exact facial dysmorphic features required for each category are defined. Assessment of the family and genetic history of each affected child is not addressed adequately; and ARBD and ARND are not practically defined in a clinical sense. - Washington criteria/4-digit code: the myriad of diagnostic categories is confusing and the system is impractical for routine use in clinical practice. Much emphasis is placed on encephalopathy and neurobehavioural disorder – these findings are not specifically defined and are not unique to the prenatal effects of alcohol on fetal development. Genetic background of the child is not adequately integrated into the criteria. Potential for over diagnosis. - The authors proposed revision and clarification of the 1996 IOM criteria for diagnosis of FASD. Data from this large multiracial cohort of children prenatally exposed to alcohol indicate that this method can be applied easily in clinical practice, thus improving care for affected children and leading to improved precision of clinical and population-based research in FASD. - Simple IQ tests are inadequate to differentiate children with ARND from those with developmental disabilities resulting from other causes. There is an emerging consensus that children with ARND are markedly impaired in executive functioning , however, these children perform in the normal range

	with relatively simple tests.
Evaluation	- These revisions correct the vagueness of the original IOM criteria by defining the degree of growth deficiency and specifying the minor physical anomalies required to make diagnoses in the FASD continuum. - ARBD and ARND are specifically defined - The diagnostic approach is multidisciplinary and uses the input of physicians, psychologists, educational diagnosticians, and skilled maternal interviewers in categorising the disabilities. - The approach is evidence based and uses data from previous animal and human studies - Application to our extensive database of children prenatally exposed to alcohol demonstrates that the method is rigorous and accurate - The suggested method is not based only on prenatal alcohol exposure but stresses diagnosis based on elimination of known genetic and malformation syndromes and inclusion based on the pattern of deficits observed among children prenatally exposed to alcohol. - A weakness is that normative values currently used for growth and facial morphologic features are based largely on white populations. - There is a need for development of normal height and weight curves and anthropometric data for palpebral fissure lengths for other races.
Notes	- FASD must always be a diagnosis of exclusion. Many genetic and malformation syndromes have some of the clinical characteristics of FAS.

Citation	**(National Center on Birth Defects and Developmental Disabilities, 2004)** **National Center on Birth Defects and Developmental Disabilities. Fetal alcohol syndrome: guidelines for referral and diagnosis: Centre for Disease Control 2004.**
Publication status	Published Report
Source	Elliott Review
Country of origin/region	USA
Aim	Develop guidelines for the diagnosis of FAS and other negative birth outcomes resulting from prenatal exposure to alcohol
Instrument/s	CDC Fetal Alcohol Syndrome: Guidelines for Referral and Diagnosis
Diagnostic Criteria	<u>Facial dysmorphia</u> - Smooth philtrum – measured as 4 or 5 on Lip-Philtrum Guide - Thin vermillion border – measured as 4 or 5 on Lip-Philtrum Guide - Small palpebral fissures – measured as $\leq 10^{th}$ percentile according to age and racial norms <u>Growth problems</u> – confirmed prenatal or postnatal height or weight, or both, at or below the 10^{th} percentile, documented at any one point in time (adjusted for age, sex, gestational age, and race or ethnicity). <u>CNS abnormalities</u> - Structural 3) Head circumference (OFC) at or below the 10^{th} percentile adjusted fro age and sex. 4) Clinically significant brain abnormalities observable through imaging. - Neurological

	Neurological problems not due to a postnatal insult or fever, or other soft neurological signs outside normal limits. - Functional Performance substantially below that expected for an individual's age, schooling, or circumstances, as evidenced by: 3) Global cognitive or intellectual deficits representing multiple domains of deficit (or significant developmental delay in younger children) with performance below the 3rd percentile (2 standard deviations below the mean for standardised testing). OR, 4) Functional deficits below the 16th percentile (1 standard deviation below the mean for standardised testing) in at least three of the following domains: a) cognitive or developmental deficits or discrepancies b) executive functioning deficits c) motor functioning delays d) problems with attention hyperactivity e) social skills f) other, such as sensory problems, pragmatic language problems, memory deficits, etc. <u>Maternal Alcohol Exposure</u> 1) Confirmed prenatal alcohol exposure 2) Unknown prenatal alcohol exposure <u>Criteria for diagnosis</u> requires all three of the following: - Documentation of all three facial abnormalities (smooth philtrum, thin vermillion border, small palpebral fissures) - Documentation of growth deficits - Documentation of CNS abnormality
Outcomes/ Recommendations (Elliott et al., 2008)	A primary goal of these guidelines is to provide standard diagnostic criteria for FAS so that consistency in the diagnosis can be established for clinicians, scientists, and service providers. There is a great need to acquire science-based information that will facilitate diagnostic criteria for additional related disorders, such as ARND.
Notes	FAS diagnosis is based on clinical examination of features, but not all children with FAS look or act the same. Lack of knowledge and misconceptions among primary care providers. Lack of diagnostic criteria to distinguish FAS from other alcohol-related conditions. Initial recognition that a child or older individual has a potential problem can come from many sources. Often, parents notice differences between a child and his or her siblings. School systems and day-care staff interact with a large number of children and often recognize when someone is having difficulty. Social service professionals frequently recognize children and individuals having difficulties and needing evaluation. Healthcare providers (particularly paediatricians) often are the first to screen for and detect problems.

Citation	**(Bertrand et al., 2005)** **Bertrand J, Floyd RL, Weber MK. Guidelines for identifying and referring persons with fetal alcohol syndrome. MMWR: Morbidity & Mortality Weekly Report. 2005;54(RR-11):1-14.**
Publication status	Published (Guidelines) Journal
Source	Online Database
Country of origin/region	USA
Research question/aim	Summarises the guidelines drafted as a result of the Scientific Working Group's deliberations. CDC update and refine diagnostic and referral criteria for FAS. Recommendations for when and how to refer a person suspected of having problems related to prenatal alcohol exposure and assesses existing practices for creating supportive environments that might prevent long-term adverse consequences associated with FAS
Instrument/s	Guidelines for identifying and referring persons with FAS
Diagnostic Criteria	**Diagnosis:** - Key indicator for FAS is the set of characteristic facial features - Lack of confirmation of alcohol use during pregnancy should not preclude a diagnosis of FAS if all other criteria are present. - The diagnosis should be classified on the basis of available history as confirmed prenatal alcohol exposure or unknown prenatal alcohol exposure. - Prenatal exposure to alcohol alone is not sufficient to warrant a diagnosis of FAS. A diagnosis requires documentation of 1) three specific facial abnormalities, 2) growth deficit, and 3) CNS abnormalities. - (figure provided: Characteristics for diagnosing FAS – CDC guidelines) - Other additional features can be present – in addition to the key facial dysmorphic features – maxillary hypoplasisa is often noted for persons with FAS (Ebrahim et al, 1998). Features often change with age or development. After puberty characteristic facial features associated with FAS can become more difficult to detect (Connor e tal, 1999), however the key features remain constant for the majority of persons with FAS (Mattson et al, 1998; Coles, 1993). - Changes in growth pattern across development also lead to variability in presentation. For certain affected persons, growth problems might occur at a younger age but not be present at the time of the diagnostic evaluation. The clinician should be certain that the child was not nutritionally deprived at the single point in time when the growth deficit was present. - Diagnostic criteria for CNS abnormality require documentation of one of three types of deficits or abnormalities (structural, neurologic and functional). - In order to capture the full spectrum of effects adequately, two levels of functional deficits are consistent with the criteria for a CNS abnormality: 1) performance below the third percentile (two standard deviations below the mean) on a measure of global cognitive functioning or 2) performance <16th percentile (one standard deviation below the mean) on standardised measures of three functional domains. - A process of differential diagnosis is essential in making an accurate FAS diagnosis. Certain syndromes have single overlapping features with FAS. Both environmental and genetic bases for growth retardation should be considered for differential diagnosis when considering a FAS diagnosis. Differential diagnosis of CNS abnormalities involves not only ruling out other disorders but also specifying simultaneously occurring disorders. Disrupted home

	environments or other external factors can produce functional deficits in multiple domains that overlap those affected by FAS. In making differential diagnosis, the clinician should evaluate CNS abnormalities in conjunction with dysmorphia and laboratory findings. To assist with differential diagnosis between FAS and environmental causes for CNS abnormalities, clinicians should obtain a complete, detailed history for the person and family members. **Referral:** - If prenatal alcohol exposure is unknown, a child or person should be referred for a full FAS evaluation when alcohol abuse (seven or more alcoholic drinks person week or three or more alcohol drinks on multiple occasions, or both) is confirmed. - If prenatal alcohol exposure is unknown, an individual should be referred for full FAS evaluation when: a parent or caregiver reports that a child might have FAS; all three facial features are present; one or more facial features are present in addition to growth deficits in height, weight or both or one or more facial features are present and one or more CNS abnormalities; or one or more facial features are presenting with growth deficits and one or more CNS abnormalities. - Social and family history factors might indicate a need for referral: premature maternal death related to alcohol use; living wit an alcoholic parent; current or previous abuse or neglect; current or previous involvement with child PSAs; a history of transient care giving situations; or having been in foster or adoptive care.
Notes	- Identifying CNS abnormalities resulting from prenatal alcohol exposure can be the most difficult aspect of a FAS diagnosis because of the heterogeneity of expression for these deficits across persons. - These guidelines represent a consensus of opinion from persons with expertise in relevant scientific and clinical fields, with input from service professionals and families affected by FAS.

Citation	**(L. Elliott et al., 2008)** **Elliott, L., K. Coleman, et al. (2008). Fetal Alcohol Spectrum Disorders (FASD): systematic reviews of prevention, diagnosis and management, Health Services Assessment Collaboration. 1(9).**
Publication status	Published (Review) Report
Source	Key Informant
Country of origin/region	New Zealand
Research question/aim	Postnatal screening/diagnosis: review of top-level strategies from existing systematic reviews and clinical practice guidelines; brief narrative discussion of non-systematic by high quality, comprehensive reviews.
Instrument/s	Results were organised under three sections: a summary of any identified diagnostic criteria; a summary of any identified diagnosis guidelines; and a summary of any key review articles. Literature describing postnatal diagnostic criteria: ▪ Institute of Medicine ▪ 4-Digit Diagnostic Code ▪ Hoyme Updated Institute of Medicine Criteria

	Postnatal screening or referral guidelines: ▪ Canadian FASD referral Guidelines ▪ Center for Disease Control FAS Referral Guidelines Postnatal diagnostic guidelines: ▪ Canadian Guidelines 2005 ▪ Center for Disease Control Guidelines 2004 (FAS) ▪ British Medical Association Guidelines 2007
Diagnostic Criteria	Postnatal screening is used to identify individuals who may have FASD. Individuals who are positive after postnatal screening should be referred for a full FASD diagnosis. A screening strategy should be broad and identify all individuals who may potentially have FASD. A full diagnostic evaluation should only be performed by a trained specialist, and often requires a multi-disciplinary team. Institute of Medicine: five diagnostic categories: FAS with and without a confirmed history of alcohol exposure, partial FAS, alcohol-related birth defects (ARBD) and alcohol-related neurodevelopmental disorder (ARND). Screening guidelines recommended that screening should occur based on identification of facial feature, known exposure to alcohol or learning and/or behavioural difficulties. Diagnostic approaches included evaluating maternal prenatal alcohol exposure, characteristic facial abnormalities, growth retardations and CNS abnormalities.
Notes	Outcome Overall there was limited high level evidence available for postnatal screening and diagnosis and management of FASD. Therefore it was not possible to identify the best method for implementation in New Zealand. The 4-Digit diagnostic code was the most commonly used criteria worldwide. <u>Postnatal screening:</u> ▪ Screening of mothers to trigger referral of children considered likely to have FASD to a paediatrician for formal diagnosis (asking mothers retrospective questions about alcohol consumption during that child's pregnancy). ▪ Screening of children to trigger referral of children considered to be at risk of FASD to a paediatrician for formal diagnosis of FASD (through the health system, education system, mental health system, judicial system or social services). - The 4-Digit Diagnostic Code was developed in response to concerns that guidelines such as those developed by the Institute of Medicine were not sufficiently specific to assure diagnostic accuracy or precision. - Hoyme et al state that the Institute of Medicine criteria is vague, with no specific parameters being set forth for diagnosis in each category. The degree of growth deficiency, facial dysmorphic features, behavioural and cognitive deficits are not clearly defined. - The pattern and severity of outcome is dependent on the timing, frequency and quantity of prenatal alcohol exposure and is frequently confounded by other adverse prenatal and postnatal exposure events.

Citation	**(Burd et al., 1999)** **Burd, L., C. Cox, et al. (1999). "The FAS Screen: a rapid screening tool for fetal alcohol syndrome." Addiction Biology 4(3): 329-336.**
Publication status	Published Journal
Source	Reference list
Country of origin/region	USA
Level of evidence	IV = study of diagnostic yield (no reference standard)
Study type/design	diagnostic yield
Research question/aim	To develop a brief screening tool for use in population-based settings to improve the identification of children with FAS
Intervention/Instrument (Elliott et al., 2008)	FAS Screen - Designed for use in community settings (public schools, preschool programs) and clinical settings by both paraprofessional and medically trained personnel. - Screening for 4-18 year olds - Rapid test – 15 minutes or less
Instruments mentioned	N/A
Patient population (Elliott et al., 2008)	1013 children aged 3-14 years 6 sites in North Dakota (schools)
Comparator (Elliott et al., 2008)	N/A
Outcomes/ Recommendations (Elliott et al., 2008)	Sensitivity (How good is this test at picking up people who have the condition?) = 100% Specificity (How good is this test at correctly excluding people without the condition?) = 94.1% Positive predictive value (If a person tests positive, what is the probability they have the condition?) = 9.2% Negative predictive value (If a person tests negative, what is the probability that they do not have the condition?) = 100% The tool is rapid; has adequate performance characteristics, and the test is cost effective.
Evaluation	- Costs of screening were calculated using costs of training, staff time, staff travel and printing costs for the screening tool. The cost per case included the costs of screening, and scheduling the clinics and the diagnostic evaluation which includes a meeting with the parents to discuss the results with a copy of the report sent to the child's physician. The cost of screening was $13 per child and the cost per case of FAS identified was $4100. ($US) - As the screening continues the cost of screening declines to about $11 per case.
Notes	- The development of a rapid screening tool suitable for use in population based screening would be helpful in case finding and facilitate early identification. - Early identification may improve adolescent and adult outcomes for persons with FAS by providing needed access to intervention services early in life (Burd & Wentz, 1997). - Completing all steps (screen, diagnostic assessments and meeting with parents) to the end of the process with children whose parents are active

	alcoholics was challenging.

Citation	**(Poitra et al., 2003)** **Poitra BA, Marion S, Dionne M, Wilkie E, Dauphinais P, Wilkie-Pepion M, et al. A school-based screening program for fetal alcohol syndrome. Neurotoxicol Teratol. 2003 Nov-Dec;25(6):725-9.**
Publication status	Published Journal
Source	Online Database
Country of origin/region	US
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	IV = study of diagnostic yield (no reference standard)
Study type/design	Study of test accuracy
Research question/aim	The purpose of this screening project was to examine the feasibility of screening for FAS in community settings. This project was felt to be a useful opportunity to accomplish three goals: (1) to examine the development of a gate to identify children who have development or behavioural disorders from prenatal alcohol exposure; (2) to estimate prevalence rates of FAS and to capture some cases of partial FAS; and (3) to examine the epidemiologic performance characteristics of the FAS Screen when applied by community personal in a community setting.
Intervention/Instrument (Elliott et al., 2008)	FAS Screen: a 32 item screening test, a rapid screening tool for community-based screening of FAS. The goal is to screen out low-risk children and identify a high risk population. The FAS Screen in a community setting typically screens out as low risk about 94–96% of children The sensitivity in the norming sample was 100%, the specificity was 94%, the positive predictive value was 92%, and the accuracy was 94%. The screening project is supported by the school. The cost of diagnosis is billed to insurance or medical assistance. Some children are charged on a sliding fee scale. No child is refused due to inability to pay. The diagnostic clinics are held one to two times per year. Children who had scores above the cut-off or during the screening and miss the clinic appointment are then seen at one of two regional referral centers either 50 or 200 miles away in other identical genetic dysmorphology clinics.
Instruments mentioned	FAS screen
Patient population (Elliott et al., 2008)	1384 kindergarten students
Comparator (Elliott et al., 2008)	No comparator
Outcomes/ Recommendations (Elliott et al., 2008)	Annual prevalence of positive screen in those children who were screened ranged from 3.2% to 8.3% over the 9 years. 11% of these were diagnosed with FAS or partial FAS
Evaluation	Sensitivity of screening tool was 100% and specificity was 95%.

Notes	

Citation	**(Burd et al., 2000)** **Burd L, Cox C, Fjelstad K, McCulloch K. Screening for fetal alcohol syndrome: is it feasible and necessary? Addict Biol. 2000 Apr 1;5(2):127-39**
Publication status	Published (Review) Journal
Source	Online Database
Country of origin/region	USA
Research question/aim	Review of screening processes for FAS
Instrument/s	Screening tools for FAS - Screening tool developed by Astley & Clarren - FASSCREEN
Diagnostic Criteria	- Screening tool developed by Astley & Clarren uses photographs of patients to screen for FAS. This tool utilizes facial features in photographs and computer-guided measurements to identify the essential dysmorphic features of children 2 months ± 10 years of age to screen for FAS. - FASSCREEN was designed for use in community- based screening programs. It is rapid, takes about 10 minutes per child, and can be administered by paraprofessionals after a 4-hour training session.
Evaluation	- **Astley & Clarren photographic screening**: A dysmorphologist, using a *gestalt* approach, correlates the computerized data with clinical diagnosis to calculate sensitivity and specificity of the screening tool. This method has been found to have a specificity of 100% and sensitivity of 100% in clinical settings (42, 43). Results from population-based screening tests have not yet been published. Other potential limitations of this screening tool include access to the computer equipment, inadequate quality of photographs, corrective lenses on the children which may distort facial anatomy and an age limit, as the facial features of children over the age of 10 become less distinct. - **FASSCREEN** was evaluated in a population of 1500 children in North Dakota. The FASSCREEN has an estimated sensitivity of 100% (44). The specificity of the test was 94%. The negative predictive value was 100%. The positive predictive value of the test was 9% (44). This test typically excludes 94% of those children screened as not having FAS and will correctly identify nearly all people with FAS. A positive screening score indicates that 9% of the people who scored above 20 and are seen for examination will have FAS. Reliability studies are currently under way utilizing the FASSCREEN in other populations to determine the reliability of the screening test. The FASSCREEN has not been evaluated in a clinical setting. The performance characteristics of screening tests will vary in different populations. The FASSCREEN was developed and the performance characteristics were examined in North Dakota.
Notes	- Different screening tools will need to be paired with different ascertainment and diagnostic strategies depending on the location of the screening (screening in a birth defects clinic vs. screening in a classroom of children with mental retardations vs. screening new-borns). - The Astley & Clarren screening tool appears to be very useful in the clinic population. - The FASSCREEN tool has shown success in a population-based setting.

Citation	**(Goh & Rosenbaum, n.d.)** **Goh YI, Rosenbaum C. FASD screening tool development project: FASD screening in children and youth: a review of the literature: Canadian Association of Paediatric Health Centres n.d.**
Publication status	Published Report
Source	Internet
Country of origin/region	Canada
Instrument/s	1. Maternal screening in pregnancy excluded 2. Facial phenotype ▪ Sensitivity estimated at 99-100%, specificity 64-99%, Positive predictive value 85.7%, Negative predictive value 100% ▪ Possible underestimation of palpebral fissure length, especially under age 4 years 3. Screening Checklist (Burd) ▪ Reported sensitivity 100%, specificity 94-95%, Positive predictive value 92%, accuracy (?) 94-95% 4. Antenatal ultrasound ▪ Small for gestation age is too non-specific ▪ Abnormality of the splenium of the corpus callosum only looked at in one small study 5. EEG – not validated 6. MRI (MRI, functional MRI and MR spectroscopy (MRS) – not validated 7. Diffusion Tensor Imaging – not validated 8. Meconium Fatty Acid Ethyl Esters ▪ Non-invasive and objective ▪ Limited window of collection and requires specific handling ▪ Only detects alcohol exposure after 12-14 weeks 9. Hair ▪ Not validated ▪ Forms from 20 weeks' gestation ▪ Can be collected up to 3 months 10. Cord blood ▪ AST, ALT, GGT and CDT measured – not effective screen 11. Fetal Alcohol Behavior Scale – needs more validation 12. CBCL ▪ Modification proposed as a screening tool with 2 step approach ▪ Not been replicated in a large sample size nor across different populations 13. Ocular motor testing (assesses executive function) ▪ Saccadic reaction times – children with FASD had prolonged reaction times, excessive direction error and no express saccades compared to controls ▪ Needs validation 14. ALARM ▪ Justice system screening of adaptive behaviour, language, reasoning and memory.

	▪ No validation 15. FAS Indicator Tool – not validated 16. Review of justice system inmate records ▪ IQ, height, weight, facial features
Diagnostic Criteria	Criteria
Notes	Recommendation is: further research is needed. Summarises a number of screening tools – useful to support research papers referring to screening tools.

Citation	**(Conry & Asante, 2010)** **Conry, J. and K. Asante (2010). Youth probation officers' guide to FASD screening and referral. British Columbia, The Asante Centre for Fetal Alcohol Syndrome**
Publication status	Published (Guidelines) Report
Source	Internet
Country of origin/region	Canada
Research question/aim	
Instrument/s	The FASD Screening Tool and Referral Form for Youth Probation Officers was developed to be used as part of a referral process for an FASD diagnostic assessment in the Youth Justice FASD Program at the Asante Centre. The rating scores are not on a continuous scale with cut-off points representing a greater or lesser probability of the youth Having FASD. It is a screening and referral from for a more formal assessment.
Diagnostic Criteria	- Diagnosed by a multidisciplinary team consisting of a paediatrician (or other specialist experienced in dysmorphology, genetic conditions and developmental disabilities), psychology, speech and language pathologist and other health professionals who can interpret the assessment findings from their respective disciplines. - The screening tool items should be based on information that is generally available to the probation officer - Gathering the information should not be time-consuming for the probation officer - Information requested should be fairly general, and not require special expertise on the part of the probation officer - The information in the items should be linked to specific criteria for making an FASD diagnosis A) Social Factors are those that may identify a youth at-risk for FASD. That is, these factors may increase the probability that the youth could have FASD: - Youth is adopted - Youth currently, or previously, was in foster care or involved with child protection services - Youth has a sibling with a documented diagnosis of FASD - There is documentation that the youth is suspected of having FASD - Youth's mother has known history of alcoholism or prenatal alcohol use B) Personal Factors are those that have been associated with (but not necessarily unique to) FASD.

	- Developmental delay in early childhood (speech/language therapy, occupational therapy, infant development or child development services prior to school entry) - Learning difficulties (learning assistance, modified program or experienced school failure or drop-out) - Growth deficiency (appears short compared to peers, or of a low weight for age) - Diagnosis of ADHD - Mental health diagnosis (anxiety, depression, Oppositional Defiant Disorder, Conduct Disorder) The youth should be referred for assessment if he/she had - 1 social factor PLUS at least 2 personal factors, OR - No social factors PLUS at least 3 personal factors - The - Where there is a probability that a client's problems may be related to prenatal alcohol exposure, the officer should endeavour to gather information from the client's past medical records and other sources (birth mother, physician's/midwife's prenatal and birth records, maternal grandparents/aunts, social workers' records, father's or mother's partners). - Personal factors can be obtained from family members, social workers, previous reports and school records.
Outcomes/ Recommendations	- Screening should not be done if there is no follow-up with a full diagnostic assessment. - While the screening indicators in the screening items are quite general, they are linked to the specific criteria for making an FASD diagnosis. -
Notes	- The purpose of screening is to identify individuals who are likely to have a particular condition so that a comprehensive, diagnostic assessment can follow. - Screening for FASD is of little value and can be harmful without a referral for a comprehensive FASD assessment.

Citation	**(Canadian Association of Paediatric Health Centres, 2010)** **Canadian Association of Paediatric Health Centres (2010). National screening tool kit for children and youth identified and potentially affected by Fetal Alcohol Spectrum Disorder. Canada, Canadian Association of Paediatric Health Centres.**
Publication status	Published Report
Source	Key Informant
Country of origin/region	Canada
Research question/aim	Survey and critically evaluate FASD screening tools and methods in use in Canada for referral to or acceptance into diagnostic clinics Evaluate practical values (sensitivity, specificity, and predictive values) of these tools Develop practical guidelines (the Tool Kit) based on the identified and evaluated tools

Instrument/s	Neurobehavioural Screening Tool Meconium testing Maternal Drinking Guide Medicine Wheel Tools FASD Screening and Referral Form for Youth Probation Officers
Diagnostic Criteria	
Notes	In-depth process was developed to identify, evaluate and develop FASD screening tools for children and youth, providing an effective methodology for on-going tool assessment and development. Process included: Survey of diagnostic clinics in Canada Critical review of the literature Establishment of a National Advisory Workshops of researchers and frontline providers Piloting of tools A process for future tool evaluation

Citation	**(E. Elliott & Peadon, 2009)** **Elliott E, Peadon E. Fetal alcohol syndrome. British Medical Journal: Epocrates Online; 2009 [cited 2010 24 September]; Available from: https://online.epocrates.com/u/29111141/Fetal+alcohol+syndrome/Summary/Highlights.**
Publication status	Published Online content
Source	Key informant
Country of origin/region	Australia
Instrument/s	Screening: • screening tests: o Question pregnant women at their 1st prenatal visit about their alcohol consumption [amount, frequency, pattern of intake] currently and in the 3 months before pregnancy. Screening tools available – TWEAK, T-ACE, AUDIT identify hazardous rather than low-level drinking o Screen asymptomatic children at population level to identify children with FASDs • Biomarkers: o Fatty acid ethyl esters (FAEE) in maternal hair and meconium • Prenatal ultrasonography: o Fetal growth parameters • Facial photography: o Digital facial photography o Stereo image matching • FASD checklist – 32 item: o Used to screen children in kindergarten for FASD, includes facial features, musculoskeletal anomalies, development

	Diagnosis: • Institute of Medicine criteria for FAS, • FASD 4-digit diagnostic code, • Centers for Disease Control and Prevention criteria for FAS, • Clarification of Institute of Medicine criteria for FAS, • FASD: Canadian guidelines for diagnosis)
Diagnostic Criteria	• Institute of Medicine criteria for FAS, • FASD 4-digit diagnostic code, • Centers for Disease Control and Prevention criteria for FAS, • Clarification of Institute of Medicine criteria for FAS, • FASD: Canadian guidelines for diagnosis)
Outcomes/ Recommendations (Elliott et al., 2008)	Authors report a FAS Screen Checklist of 32 items (ref Poitra, Marion, Dionne, 2003) with: • Sensitivity 100% • Specificity 94% • Positive predictive value 92%
Notes	This monograph outlines FAS in terms of: • Key highlights, • Definition, • ICD classification, • Epidemiology, • Etiology, • Pathophysiology, • Diagnostic approach (history, physical exam, investigations, referral, barriers to diagnosis), • Risk factors, • History and exam (key diagnostic factors, other diagnostic factors), • Diagnostic tests (first to order, other tests to consider, emerging tests), • Differential diagnosis, • Diagnostic criteria (Institute of Medicine criteria for FAS, FASD 4-digit diagnostic code, Centers for Disease Control and Prevention criteria for FAS, Clarification of Institute of Medicine criteria for FAS, FASD: Canadian guidelines for diagnosis), • Screening (tests, biomarkers, prenatal ultrasonography, facial photography, FASD checklist), • Treatment approach (individual assessment, academic or learning difficulties, social-skills deficits, behavioural problems, externalising or attention problems, attention deficit hyperactivity disorder), • Treatment options, • Emerging therapies, • Primary prevention, • Secondary prevention, • Prognosis, • Monitoring, • Patient instructions, • Complications, • Guidelines

Citation	**(S. J. Astley & Clarren, 1995)** **Astley, S. J. and S. K. Clarren (1995). "A fetal alcohol syndrome screening tool." Alcoholism: Clinical & Experimental Research 19(6): 1565-71**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	IV = study of diagnostic yield (no reference standard)
Study type/design	diagnostic yield
Research question/aim	Is there a distinct facial phenotype for FAS? If so, which features are most discriminatory for FAS?
Intervention/Instrument (Elliott et al., 2008)	Gestalt approach to diagnosis
Instruments mentioned	'Gestalt' diagnostic process
Patient population (Elliott et al., 2008)	194 children 0.2 - 10 years of age referred to the University of Washington FAS diagnostic clinic for assessment (ie high risk for alcohol exposure)
Comparator (Elliott et al., 2008)	Clinical judgement of a single dysmorphologist
Outcomes/ Recommendations (Elliott et al., 2008)	Children with and without FAS can be differentiated based on three features: Palpebral fissure length, philtrum smoothness and thinness of upper lip. Screening for these three facial features may provide a cost effective FAS screening tool
Evaluation	Assessment of diagnostic inter-rater agreement between trained dysmorphologists and testing in other clinic populations will be needed to assess the tool's external validity. Useful screening tool for facial dysmorphology – with further research required.
Notes	

Citation	**(S. J. Astley & Clarren, 2001)** **Astley, S. J. and S. K. Clarren (2001). "Measuring the facial phenotype of individuals with prenatal alcohol exposure: correlations with brain dysfunction." Alcohol and Alcoholism 36(2): 147-59.**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)

Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence
Study type/design	Study of test accuracy
Research question/aim	To demonstrate the use of two measures of the magnitude of the FAS facial phenotype (the 4-digit diagnostic code and the D-score) To correlate these two measures with brain structure and function
Intervention/Instrument (Elliott et al., 2008)	4-digit diagnostic code
Instruments mentioned	Gestalt diagnostic method D-score of magnitude of the FAS phenotype
Patient population (Elliott et al., 2008)	952 patients with confirmed antenatal alcohol exposure (4-digit code 3 or 4) assessed at the WA FAS DPN clinic
Comparator (Elliott et al., 2008)	Gestalt diagnostic method
Outcomes/ Recommendations (Elliott et al., 2008)	4-digit diagnostic code is a more objective, quantitative measure of the FAS phenotype than the Gestalt method Supportive evidence that midline defects can predict brain dysfunction
Evaluation	
Notes	

Citation	**(S. J. Astley & Clarren, 2000)** **Astley, S. J. and S. K. Clarren (2000). "Diagnosing the full spectrum of fetal alcohol-exposed individuals: Introducing the 4-digit diagnostic code." Alcohol and Alcoholism 35 (4): 400-410.**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence
Study type/design	Comparison Study of test accuracy
Research question/aim	
Intervention/Instrument (Elliott et al., 2008)	4-digit diagnostic code Gestalt method of diagnosis

Instruments mentioned	Instrument 4-digit diagnostic code Gestalt diagnostic method
Patient population (Elliott et al., 2008)	1014 patients 0-51 years old diagnosed in the Washington University FAS DPN
Comparator (Elliott et al., 2008)	Gestalt diagnostic method
Outcomes/ Recommendations (Elliott et al., 2008)	The 4-digit diagnostic code is more specific than the Gestalt diagnostic approach Inter- and intra-rater reliability between the two authors was 100% Inter-and intra-rater reliability between each of the 6 satellite diagnostic clinics and the University of Washington clinic was 94% for all four digits, and 100% for the diagnostic category
Evaluation	
Notes	

Citation	**(S. J. Astley, 2006)** **Astley, S. J. (2006). "Comparison of the 4-digit diagnostic code and the Hoyme diagnostic guidelines for fetal alcohol spectrum disorders." Pediatrics 118 (4): 1532-1545.**
Publication status	Published Journal
Source	Online database
Country of origin/region	USA
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence
Study type/design	Comparison
Research question/aim	Comparing the 4-Digit Diagnostic Code and Hoyme FASD diagnostic guidelines 1. To assess the specificity of the Hoyme FAS facial phenotype for the Hoyme FAS diagnosis when the Hoyme guidelines were applied to the University of Washington FASD clinical population 2. To assess the specificity of the Hoyme FAS facial phenotype to prenatal alcohol exposure when the Hoyme diagnostic guidelines were applied to a study population with confirmed absence of prenatal alcohol exposure 3. To compare the prevalence of FAS (with and without confirmed prenatal alcohol exposure) between the Hoyme and 4-Digit Code criteria for FAS, when the 2 sets of criteria were applied to the University of Washington FASD clinical population or 2004 version of the 4-Digit Diagnostic Code 4. To compare, on a case-by-case basis, which patients did and did not receive a diagnosis of FAS when the Hoyme and 4-Digit Code FAS criteria were applied to

	the University of Washington FASD clinical population
Intervention/Instrument (Elliott et al., 2008)	
Instruments mentioned	4-Digit Diagnostic Code Hoyme FASD diagnostic guidelines Institute of Medicine (IOM) FASD guidelines Centres for Disease Control and Prevention (CDC) FAS guidelines Canadian FASD guidelines
Patient population (Elliott et al., 2008)	952 patients who had received an interdisciplinary fetal alcohol spectrum disorders, diagnostic evaluation at the University of Washington with the 4-Digit Diagnostic Code and 16 children with confirmed absence of alcohol exposure
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	The Hoyme facial feature criteria are less specific (68%) than the 4-digit diagnostic code (99.8%) across the entire study population Specificity was low (75%) for the facial phenotype in the control population (children with confirmed absence of exposure to alcohol in utero) ie. 25% of these children met the Hoyme facial feature criteria for FAS, even though they had confirmed absence of alcohol exposure in utero The Hoyme CNS criteria lack specificity (because they include OFC </= 10th centile) and sensitivity (because they do not take into account neurobehavioural deficit) The Hoyme criteria are not based on a representative sample (they were developed from a population of South Africans and Native Americans) Recommend that the 4-digit diagnostic code or the Canadian FASD guidelines be adopted Recommend caution in developing screening tools that are a relaxed version of diagnostic criteria because they may lead to false positives
Evaluation	
Notes	

Citation	**(First Nations and Inuit Health Committee, 2002)** **First Nations and Inuit Health Committee. Fetal alcohol syndrome. Paediatr Child Health. 2002;7(3):161-74.**
Publication status	Published Journal
Source	Internet
Country of origin/region	Canada
Instrument/s	They provide a list of age-related diagnostic criteria for FAS and/or atypical FAS. Infants: 6 criteria

	Preschool: 6 criteria Middle school: 10 criteria They list tests available to delineate neurodevelopmental problems which include tests for 1. measuring intelligence, 2. tests used to measure attention and hyperactivity, 3. tests of learning and memory, 4. tests of language, 5 tests of motor abilities, 6. tests of social skills and behaviour and 7. tests of visual-spatial difficulties.
Diagnostic Criteria	Position Statement: This statement addresses FAS prevention, diagnosis, early identification and management for health care professionals. They recommend the use of the four-digit diagnostic scale is recommended for making the diagnosis of FAS – simple, straightforward, can be carried out by a well-trained physician with a minimum of sophisticated tests. Testing by a psychologist may be useful in further defining disabilities and in planning intervention.
Notes	Although the paper makes reference to the high rates of FAS in Aboriginal Canadian population, except for identifying the at-risk women, which should be done in a cultural context that they briefly outline, this is not extended to assessment and diagnosis of FAS.

Citation	**(Alberta Partnership on Fetal Alcohol Syndrome, 2003)** **Alberta Partnership on Fetal Alcohol Syndrome. Guideline for the diagnosis of fetal alcohol syndrome (FAS). The Canadian Child and Adolescent Psychiatry Review. 2003;12(3):81-6.**
Publication status	Published Journal
Source	Internet
Country of origin/region	Canada
Instrument/s	Guidelines – no instrument used
Diagnostic Criteria	See below
Outcomes/ Recommendations (Elliott et al., 2008)	Diagnosis should use following indicators A history of maternal alcohol consumption during pregnancy Prenatal and/or postnatal growth retardation Neurodevelopment and behavioural characteristics Characteristic facial features Any person suspected of having FAS should be referred to appropriate specialist. After a diagnosis is made advice should be given re: contraception to reduce likelihood of further alcohol affected births Interventions should be delivered by multidisciplinary team Information and support should be given to care givers
Notes	No single diagnostic test can confirm FAS Comorbidites with FAS are common Maternal alcohol use should be investigated for children with ADHD

Citation	**(Goh et al., 2008)** **Goh YI, Chudley AE, Clarren SK, Koren G, Orrbine E, Rosales T, et al. Development of Canadian screening tools for fetal alcohol spectrum disorder. Canadian Journal of Clinical Pharmacology. 2008;15(2):e344-66.**
Publication status	Published Journal
Source	Key Informant
Country of origin/region	Canada
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	I = a systematic review of level II studies
Study type/design	Systematic review
Research question/aim	1. survey and critically evaluate FAS screening tools and methods 2. evaluate sensitivity, specificity and predictive value of these tools 3. develop practical guidelines
Intervention/Instrument (Elliott et al., 2008)	Multiple – see below
Instruments mentioned	**Neurocognitive tools** 1. Fetal Alcohol Behavior Scale – not able to discriminate between FASD and other clinical groups 2. CBCL – (modified using 7 sensitive and specific distinguishing items) ▪ Not replicated in a large population ▪ Potential for user bias ▪ Overlaps with other neurobehavioural disorders 3. Personality Inventory for Children – can only be administered by psychologists **Facial Dysmorphology** 1. Manual measurement 2. Digital Photo analysis software Both these methods are sensitive, specific and with high positive predictive value but only for FAS and pFAS. **Meconium Fatty Acid Ethyl Esters** ▪ cut off of 2nmol/g distinguishes between heavy and light alcohol exposure with good sensitivity and specificity ▪ Objective and non-invasive tool ▪ Sample must be collected within 72 hours of birth ▪ Does not capture exposure prior to 12-14 weeks' gestation **Growth Retardation** ▪ Confounded by variation between populations ▪ Only small proportion of children with FASD affected (10-30%) ▪ May be useful in combination with other screening tools **Youth Justice Population –** conclusion is there is no validated screening tool

	1. Manitoba screening project 12 -18 years and confirmed prenatal alcohol exposure - 60% positive predictive value 2. Saskatchewan project – 28 risk factor items ▪ 0.82 inter rater reliability ▪ 76% validity (not sure how this was determined) 3. Stony Mountain project – Brief Screen Checklist ▪ Items highly correlated with a FASD diagnosis 4. Asante screen questionnaire – 26.5% referred for assessment **Clinic tools** ▪ No tools have been validated ▪ Clinic intake criteria useful e.g. Complex Developmental Behavioural Condition Referral Form ▪ Also the Clinic for Alcohol and Drug Exposed Children (CADEC) intake criteria lead to a diagnostic rate of ~50%, specificity of 24.5% and claim sensitivity of 100% but unknown number of false negatives. **Community Tools** 1. Medicine Wheel tools for teacher and semi-structured parent interview developed for First Nations community ▪ 29/237 referred and 67% found to have a FASD diagnosis. ▪ Culturally specific ▪ Uses multiple sources of information and involves parents and teachers in the process. ▪ A SAMHSA FASD Center of Excellence screening tool and a FASD Functional Screening tool are mentioned but not described nor referenced.
Patient population (Elliott et al., 2008)	Individuals who may have FASD – varied between studies
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	No one tool will meet need for all populations Multiple tools needed Lack of validated tools
Evaluation	Discusses feasibility, accessibility of different tools Does not discuss antenatal screening for alcohol consumption
Notes	

Citation	**(Weiss et al., 2004)** **Weiss, M., C. Cronk, et al. (2004). "The Wisconsin Fetal Alcohol Syndrome Screening Project." Wisconsin Medical Journal 103(5): 53-60.**
Publication status	Published Journal
Source	Key Informant
Country of origin/region	USA
Level of evidence	IV = study of diagnostic yield (no reference standard)

(NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	
Study type/design	Diagnostic yield (no reference standard)
Research question/aim	Evaluate prevalence of FAS in Wisconsin
Intervention/Instrument (Elliott et al., 2008)	Multi-stage, multi-source prospective population-based screening methodology Wisconsin Fetal Alcohol Syndrome Surveillance Project Screen 1 – small for gestational age, defined as birth weight below the 10th percentile based on sex and gestational age-specific reference, lower birth weight values for African American infants were 200g less than those for other infants Screen 2 – neonatal medical records abstracted for birth head circumference, gestational age, maternal alcohol use Screen3 – assessment of facial features of FAS phenotype, measurements of growth and development.
Instruments mentioned	
Patient population (Elliott et al., 2008)	Birth cohort infants born in 1998 and 1999 in 22 birth hospitals to mothers resident in an 8 county region in southeast of Wisconsin. Included urban, suburban and rural households.
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	Children directly evaluated had fewer demographic, pregnancy and maternal substance use risk factors than lost to follow-up children. Using a combination of weight and head circumference below the 10th percentile at birth is a useful methodology for identifying children at substantial risk for growth and development delays from FAS or other unspecified etiologies.
Evaluation	
Notes	

Citation	**(May et al., 2007)** **May, P. A., J. Gossage, et al. (2007). "The epidemiology of fetal alcohol syndrome and partial FAS in a South African community." Drug and Alcohol Dependence 88.**
Publication status	Published Journal
Source	Key Informant
Country of origin/region	USA
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-3 = diagnostic case-control study
Study type/design	diagnostic case-control

Research question/aim	Prevalence characteristics of FAS and PFAS in primary school cohort in South Africa.
Intervention/Instrument (Elliott et al., 2008)	Two-tier screening system 1st tier – dysmorphology, growth (height, weight and head circumference) and developmental data 2nd tier – examination by two experienced dysmorphologists
Instruments mentioned	Institute of Medicine Diagnostic Criteria
Patient population (Elliott et al., 2008)	1013 first grade students. Characteristics of children with FAS and PFAS (n=818) contrasted with randomly selected control group (n=193)
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	The rate of FAS and PFAS continues to be the highest reported in any overall community and is much higher than rates elsewhere. In this cohort it is 68-89.2 per 1000. Various measures of maternal drinking are significantly correlated with negative outcomes of children in the areas of non-verbal intelligence (-0.26), verbal intelligence (-0.28), problem behaviour (0.31) and overall dysmorphology (0.59). Significantly more FAS and PFAS among children of rural residents (OR=3.79).
Evaluation	Revised IOM method is practical, consistent, reliable and produces highly specific diagnoses from examining all domains of variables (physical, psychological/developmental and alcohol exposure). Cataloguing physical and psychological trains of the children and maternal drinking practices provides the opportunity for comparing FAD across populations for examinations of relative risk.
Notes	

Citation	**(S. J. Astley et al., 2002)** **Astley, S. J., J. Stachowiak, et al. (2002). "Application of the fetal alcohol syndrome Facial Photographic Screening Tool in a foster care population." Journal of Pediatrics 141 (5): 712-717.**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-3 Diagnostic case-control study
Study type/design	1. Study of test accuracy, 2. Diagnostic yield.
Research question/aim	1. Screen a foster care population for the FAS facial phenotype, structural / functional brain abnormality (with documented antenatal alcohol exposure) and

	other syndromes identifiable with a facial photograph, 2. Provide diagnostic evaluations and management plans for all screen-positive children, 3. Determine the prevalence of FAS in a foster care population, 4. Evaluate the performance of the FAS Facial Photographic Screening Tool.
Intervention/Instrument (Elliott et al., 2008)	Screen: FAS facial photographic screening tool Occipito-frontal circumference Diagnosis: Attendance at a FAS DPN multidisciplinary clinic for assessment including FAS facial photography
Instruments mentioned	FAS facial photographic screening tool (University of Washington) FAS 4-digit diagnostic code Multidisciplinary neurodevelopmental assessment
Patient population (Elliott et al., 2008)	Children aged 0.6-13.3 in foster care in the King County (Washington State) Foster Care Passport Program
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	FAS photographic screening tool Positive predictive value is (6 / 7) 85.7% FAS photographic screening tool Negative predictive value is (590 / 590) 100% FAS photographic screening tool Sensitivity (6/6) 100% FAS photographic screening tool Specificity (590/591) 99.8% FAS photographic screening tool Accuracy (596/597) 99.8% Recommends use of the FAS photographic screening tool as cost-effective and accurate in a high risk population (those in foster care) Recommends nesting screening programs in existing health care / monitoring strategies
Evaluation	Impact: Allowed secondary prevention through awareness raising and counselling Supports the case for state-wide screening and diagnostic strategies
Notes	Statement made: “a screening tool to accurately identify persons at risk for Fetal Alcohol Effects (FAE) does not exist because the cognitive/behavioural dysfunction associated with prenatal alcohol exposure is not sufficiently specific…” The screening tool (FAS photographic screening tool) was the same as the tool used to diagnose FAS, therefore an assumption must be made that the diagnostic tool is valid in order to imply that the screening tool is valid.

Citation	**(Avner et al., 2006)** **Avner A, Henning P, Koren G, Nulman I. Validation of the facial photographic method in fetal alcohol spectrum disorder screening and diagnosis. JFAS Int. 2006;4(e20).**
Publication status	Published Journal

Source	Reference list
Country of origin/region	Canada
Level of evidence	III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence
Study type/design	study of test accuracy, comparison, diagnostic yield
Research question/aim	Validate computer assisted method of measuring the PFL and Philtrum smoothness using digital patient photographs
Intervention/Instrument (Elliott et al., 2008)	Digital facial photography compared with manual technique
Instruments mentioned	digital facial photography and manual measurements
Patient population (Elliott et al., 2008)	40 children referred for FASD assessment 21 under the age of 4 years
Comparator (Elliott et al., 2008)	Manual measurements of the palpebral fissure
Outcomes/ Recommendations (Elliott et al., 2008)	Digital facial Photographs 100% sensitive and 64% specificity with no false negatives Digital PFL measurements were significantly different from direct manual measurements in children under the age of four years; (digital PFL in children under four years of age tends to underestimate the PFL). Digital PFL measurements were not significantly different from direct technique manual measurement in children 4 years of age and older. Direct measurement scores for philtrum smoothness were different from the digital on frontal view alone but not different when considering three quarter view of the philtrum
Evaluation	Digital facial Photographs are 100% sensitive and therefore offer an efficient screening tool
Notes	

Citation	**(Fang et al., 2008)** **Fang S, McLaughlin J, Fang J, Huang J, Autti-Ramo I, Fagerlund A, et al. Automated diagnosis of fetal alcohol syndrome using 3D facial image analysis. Orthod Craniofac Res. 2008 Aug;11 (3):162-71.**
Publication status	Published Journal
Source	Online Database
Country of origin/region	USA
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic	III-3 = diagnostic case-control study

accuracy Studies)	
Study type/design	diagnostic case-control
Research question/aim	To develop a computational model that can automatically compute facial features from 3D scans and use data to identify FAS
Intervention/Instrument (Elliott et al., 2008)	Automated facial analysis
Instruments mentioned	3D facial laser scans and computer algorithm
Patient population (Elliott et al., 2008)	2 study populations – one form Finland, one from South Africa; Finnish Caucasian 36 kids with FAS, 31 controls, age range 2.8-21 years (mean 13 years); Cape Coloured pop 50 kids with FAS, 32 controls, mean age 5 years.
Comparator (Elliott et al., 2008)	Dysmorphology exam (Jones et al), diagnoses made using revised IOM criteria
Outcomes/ Recommendations (Elliott et al., 2008)	Finnish pop Sensitivity 88.2%, Specificity 100%; Cape Coloured pop Sensitivity 91.7%, Specificity 90%.
Evaluation	Good sensitivity, specificity within same ethnic group, lower test performance on missed populations; more expensive technology; less portable than a camera, further research needed to determine effects of age on facial features.
Notes	Alcohol exposure in utero assessed by questionnaire and stratified as non, minimal or greater than minimal.

Citation	**(Lang, 2006)** **Lang, J. (2006). "Ten brain domains: A proposal for functional central nervous system parameters for fetal alcohol spectrum disorder diagnosis and follow-up." Journal of FAS International 4(e12).**
Publication status	Published Journal
Source	Key Informant
Country of origin/region	USA
Research question/aim	To present specific brain domains of CNS involvement related to FASD and for use during FASD assessments and intervention recommendations
Instrument/s	
Diagnostic Criteria	4-Digit Diagnostic Code – magnitude of expression of the four key diagnostic features of FASD. Refinement of the 4-Digit Code clarified the functional CNS parameters as a way to: – Provide clear definitions of brain dysfunction for professionals and lay people to use – Specify empirical data needed for accurate diagnosis – Define intervention considerations that address the complex nature of the life-long disability with the intention to avoid common secondary disabilities.
Notes	Specific brain domains proposed: - achievement

	- adaptation - attention - cognition - executive functioning - language - memory - motor - sensory/soft neurological - social communication

Citation	**(Canada Northwest FASD Research Network, 2007a)** **Canada Northwest FASD Research Network. Psychometric Tools Used for Evaluating Individuals with FASD: Reaching Consensus – Phase 1 Meeting. Vancouver: Canada Northwest FASD Research Network 2007.**
Publication status	Published Report
Source	Internet
Country of origin/region	Canada
Instrument/s	Psychometric instruments used in the following areas of assessment were assessed: Cognition (16 different tools were used) Academic achievement (19 different tools were used) Memory (13 different tools were used) Executive function and abstract reasoning(26 different tools were used) Attention and hyperactivity (25 different tools were used) Adaptive behaviour, social skills and social communication (18 different tools were used) See attached JP Appendix 1 (Psychometric_Tools_Phase1) for the psychometric tools matrix (consensus achieved)
Diagnostic Criteria	Criteria about the population being assessed guided discussion about the tools. The following 5 criteria reflect a typical case scenario: • The person between 4 and 18 years of age • Has an IQ between 70 and 100 • Speaks English adequately • Has no sensory deficits • Has experience in life (it is valid to use tests for the general population)
Outcomes/ Recommendations (Elliott et al., 2008)	Psychometric tools matrix (consensus achieved)
Notes	In 2007 CanFASD Northwest (Phase 1) found that there was little consistency among clinics regarding psychometric tools that were used by multidisciplinary teams in diagnostic clinics who were conducting comprehensive assessments for individuals with FASD. They used a two phase process to bring together representatives of all disciplines working within diagnostic clinics who used a multidisciplinary approach. In this first phase a group of 30 psychologists (who are

	currently assessing individuals and representing 14 clinics) met for two days to achieve consensus on psychometric tools to be used in diagnosis. The group reviewed current approaches and tools and worked towards consensus on the most effective tools that would be used for assessment consistently across Canada Northwest's diagnostic clinics. A survey was distributed to all the clinics to collect information on tools currently in use – there was variability in the instruments used although these were valid and appropriate for each function. A decision about which measure to use needs to be made when considering: • Amount of time available • Patient literacy • Age of child • Approach – one on one interview or survey

Citation	**(Canada Northwest FASD Research Network, 2007b)** **Canada Northwest FASD Research Network. Psychometric Tools Used for Evaluating Individuals with FASD: Reaching Consensus – Phase 2 Meeting. Vancouver: Canada Northwest FASD Research Network 2007.**
Publication status	Published Report
Source	Internet
Country of origin/region	Canada
Instrument/s	Psychometric instruments used in the following areas of assessment were assessed: • Neurological signs (sensory motor) (28 different tools were used) • Communication (receptive and expressive) • Supplementary measures (emotional status) • Paediatrician administered measures
Diagnostic Criteria	Participants were instructed to work towards consensus and recommend tools that would be appropriate to a typically evaluated person, within the following parameters: • The person between 4 and 18 years of age • Has an IQ between 70 and 100 • Speaks English adequately • Has no sensory deficits • Has experience in life (it is valid to use tests for the general population)
Outcomes/ Recommendations (Elliott et al., 2008)	Sensitivity and specificity of FASD screening and FASD diagnosis
Notes	In 2007 CanFASD Northwest (Phase 2) found that there was little consistency among clinics regarding psychometric tools that were used by multidisciplinary teams in diagnostic clinics who were conducting comprehensive assessments for individuals with FASD. They used a two phase process to bring together

	representatives of all disciplines working within diagnostic clinics who used a multidisciplinary approach. In this second phase groups of 45 (in total) speech-language pathologists, occupational therapists, paediatricians (who were currently assessing individuals and representing 21 clinics) and a representative group (n=5) from the first phase met for two days to achieve consensus on psychometric tools to be used in diagnosis. The group reviewed current approaches and tools and worked towards consensus on the most effective tools that would be used for assessment consistently across Canada Northwest's diagnostic clinics. A survey was distributed to all the clinics to collect information on tools currently in use – there was variability in the instruments used although these were valid and appropriate for each function. The report describes processes and outcomes of this approach Tools for age categories 4-6, 6-16, 16-18 for each domain, by age group where stated to be attached as an appendix to the Phase 2 document. However, this appendix was not attached and could not be found when searching the web. A rationale is stated for most of the tests and also some supplementary tests for some of the domains. Participants also identified that additional tests may be used depending on how well the child performs and that these tests were based on clinical judgement. They also noted that clinical judgement was an important aspect of assessment and use of tools and choosing a battery of tests for every client was a challenge.

Citation	**(Hamilton, 2006)** **Hamilton, S. (2006). "Screening for developmental delay: Reliable, easy-to-use tools." The Journal of Family Practice 55(5): 415-422.**
Publication status	Published Journal
Source	Key Informant
Country of origin/region	USA
Research question/aim	Practice recommendations for screening for developmental delay
Instrument/s	PEDS – Parents' Evaluation of Developmental Status – consists of 2 open-ended questions and 8 yes/no questions. It is written at 5 grade reading level and takes approximately 5 minutes to administer of an interview is needed. It has been standardised and validated with 771 children. Sensitivity was 75% and specificity 74%. Validity was determined through comparison with a battery of tests including the Woodcock-Johnson Psychoeducational Battery: Tests of Acheivement, Standford-Binet Intelligence Scale and the Bayley Scales of Infant Development-II. The Ages and Stages Questionnaire – low cost and easily administered screening instrument relying on parental report. Items written at 4th to 6th grade reading level. Self-administered assessment can take 10-20 minutes and be scored in 1-5 minutes. Specificity ranging from 81% (16 months) to 92% (36 months), and 86% overall. Sensitivity averaged 72%. The instrument maintains its validity when screening high risk children, when used on premature children 90% sensitivity

	and 77% specificity. Brigance screens – 9 separate forms, each covering a 12-month age range. Requires 15 minutes to administer and score. The screens address speech-language, motor skills, readiness, and general knowledge at younger ages and reading and math at older ages. Standardised on 1156 children. Sensitivity 82% and 75% with a range of 72% to 100% across different years. Battelle developmental inventory – screen children 12-96 months old using direct assessment, observation and parental interview. 75% sensitivity and 73% specificity. Bayley infant neurodevelopmental screener – screening high risk infants aged 3-24 months. Standardised on 600 children 75% sensitivity and 86% specificity.
Diagnostic Criteria	
Notes	Screening tests can identify children with developmental delay with reasonable accuracy, and children may benefit from early intervention.

Citation	**(Spadoni et al., 2007)** **Spadoni AD, McGee CL, Fryer SL, Riley EP. Neuroimaging and fetal alcohol spectrum disorders. Neurosci Biobehav Rev. 2007;31(2):239-45.**
Publication status	Published **(Review)** Journal
Source	Online Database
Country of origin/region	USA
Instrument/s	MRI neuroimaging; Voxel-based morphometric analysis of tissue density; diffusion tensor imaging
Diagnostic Criteria	Descriptive of brain structure in FAS versus normal values
Notes	Promising technique to correlate structural brain abnormalities with neuropsych deficits

Citation	**(S. J. Astley et al., 2009b)** **Astley, S. J., T. Richards, et al. (2009). "Magnetic resonance spectroscopy outcomes from a comprehensive magnetic resonance study of children with fetal alcohol spectrum disorders." Magnetic Resonance Imaging 27(6): 760-78.**
Publication status	Published Journal
Source	Reference list
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	II = a study of test accuracy with: an independent, blinded comparison with a valid reference standard, among consecutive persons with a defined clinical presentation

Study type/design	Study of test accuracy
Research question/aim	Can MRS (Magnetic Resonance Spectroscopy) identify abnormalities of brain metabolism that are specific to FASD and able to differentiate between specific diagnoses within the spectrum?
Intervention/Instrument (Elliott et al., 2008)	Magnetic Resonance Spectroscopy. Neurobehavioural testing (in the areas of executive functioning, visual-spatial skills, Visual memory (Rey complex figure test), Verbal memory (California Verbal Learning Test), working memory (N-back task), academic achievement, speech/language ability and attention.
Instruments mentioned	MRI (including functional MRI, MR spectroscopy). Neurobehavioral testing battery (see above). 4-digit diagnostic code.
Patient population (Elliott et al., 2008)	65 children aged 8-15.9 years who had been diagnosed with either FAS/PFAS, Static encephalopathy-alcohol exposed, or neurobehavioural disorder-alcohol exposed at the Washington State FAS DPN clinic. 16 healthy controls with no antenatal alcohol exposure matched for age, gender and ethnicity.
Comparator (Elliott et al., 2008)	Non-human FAS MRS study (Astley et al. 1995)
Outcomes/ Recommendations (Elliott et al., 2008)	Compared with controls, children with FAS or PFAS had reduced concentrations of choline-containing compounds (reflecting cell membrane stability and myelination in frontal/parietal white matter regions lateral to the midsection of the corpus callosum. Increasing expression of the FAS facial features and neurobehavioural impairment correlated with decreased choline concentration. No significant decrease in choline concentration in the hippocampus. No significant decrease in NAA or Cre concentrations.
Evaluation	
Notes	Raises the usefulness of neuroimaging/neurometabolic studies in identifying specific brain regions affected by alcohol exposure, and their correlation with functional impairment.

Citation	**(Susan J. Astley et al., 2009)** **Astley, S. J., E. H. Aylward, et al. (2009). "Functional magnetic resonance imaging outcomes from a comprehensive magnetic resonance study of children with fetal alcohol spectrum disorders." Journal of Neurodevelopmental Disorders 1(1): 61-80.**
Publication status	Published Journal
Source	Database
Country of origin/region	USA (Washington State)
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic	III-2 = a comparison with reference standard that does not meet the criteria required for Level II and III-1 evidence

accuracy Studies)	
Study type/design	comparison
Research question/aim	To assess whether participants with FASD demonstrated impaired working memory as measured by: 1. Performance on the *N-back* assessment of working memory, and 2. Functional MRI scanning of targeted brain regions
Intervention/Instrument (Elliott et al., 2008)	N-back working memory task FMRI scanning
Instruments mentioned	4-digit FASD diagnostic code FMRI N-back test
Patient population (Elliott et al., 2008)	65 children aged 8-15.9 years who had been diagnosed with either FAS/PFAS, Static encephalopathy-alcohol exposed, or neurobehavioural disorder-alcohol exposed at the Washington State FAS DPN clinic. 16 healthy controls with no antenatal alcohol exposure matched for age, gender and ethnicity.
Comparator (Elliott et al., 2008)	
Outcomes/ Recommendations (Elliott et al., 2008)	Performance on the N-back task decreased linearly as patients progressed from the control to the FAS/PFAS groups Activation of targeted brain regions decreased on the 2-BACK task as patients progressed from the control to the FAS/PFAS groups Right sided brain regions showed higher activation on the more complex 2-BACK task Children across the entire FASD spectrum exhibit deficits in working memory
Evaluation	Provides compelling evidence that cognitive and behavioral deficits among individuals with FASD are, to an important extent, "brain-based" (they can't rather than won't think or behave as well as unaffected children). FMRI is developing as an important assessment tool to consider in FASD evaluation
Notes	Specific brain regions activated during the N-back task are discussed

Citation	**(Sowell et al., 2008)** **Sowell ER, Johnson A, Kan E, Lu LH, Van Horn JD, Toga AW, et al. Mapping White Matter Integrity and Neurobehavioral Correlates in Children with Fetal Alcohol Spectrum Disorders. J Neurosci. 2008 February 6, 2008;28(6):1313-9.**
Publication status	Published Journal
Source	Reference list
Country of origin/region	USA
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating	III-3 = diagnostic case-control study

Intervention and Diagnostic accuracy Studies)	
Study type/design	comparison, diagnostic case-control, diagnostic yield
Research question/aim	Evaluate white matter integrity in individuals with FASDs using a combination of diffusion tensor and T1-weighted magnetic resonance imaging.
Intervention/Instrument (Elliott et al., 2008)	Behavioural data MRI scan acquisition DTI T1-weighted series
Instruments mentioned	MRI and DTI comparison using voxel based morphometry (VBM) Neurocognitive deficits (implied – they were measured) (i) Wechsler(W ISC IV) (+FSIQ) (ii) Visuomotor integration (VMI-Beery) (iii) WRAT-RE
Patient population (Elliott et al., 2008)	- 17 children and adolescents with FASD, aged 7 to 15 years were compared with 19 typically developing age and gender matched controls aged 7 to 15 years - Cases: An experienced clinician examined alcohol-exposed children using the Diagnostic Guide for FAS and Related Conditions (Astley) - Controls: All subjects were screened for neurological impairments, psychiatric illness, history of learning disability or developmental delay.
Comparator (Elliott et al., 2008)	19 typically developing age and gender matched
Outcomes/ Recommendations (Elliott et al., 2008)	- Lower fractional anisotropy (FA) observed in individuals with FASDs relative to controls in Right temporal lobe and bilaterally in aspects of the splenium and corpus callosum. - Loss of white matter density in some but not all regions of reduced FA. - Significant correlations between performance on a test of visuomotor integration and FA in bilateral spenium but not temporal regions were observed within the FASD group. - Correlations between the visuomotor task and FA within the splenium were not significant within the control group and were not significant for measures of reading ability. - This suggests that this region of white matter is particularly susceptible to damage from prenatal alcohol exposure and that disruption of splenial fibers in this group in associated with poorer visumotor integration.
Evaluation	Good paper demonstrating associations between function and structure
Notes	

Citation	**(Gifford et al., 2010)** **Gifford AE, Farkas KJ, Jackson LW, Molteno CD, Jacobson JL, Jacobson SW, et al. Assessment of benefits of a universal screen for maternal alcohol use during pregnancy. Birth Defects Research Part A: Clinical and Molecular Teratology. 2010;88(10):838-46.**
Publication status	Published Journal

Source	Key informant
Country of origin/region	USA/South Africa
Study type/design	Economic analysis
Research question/aim	To estimate the benefits of universal meconium screening for maternal drinking during pregnancy.
Method	Literature search was conducted using online databases. Monetary values were recalculated from their value in US$ for 2006; consumer price index inflation estimates available through the Bureau of Labor Statistics division of the US Department of Labor.
Outcomes/ Recommendations (Elliott et al., 2008)	- Laboratory analysis of fatty acid ethyl esters (FAEEs) in meconium is a newly developed research procedure and its long term cost has not been definitively determined. - This research estimated the cost of screening to consist of administrative, materials, and professional analysis costs. - State regulated new-born screening programs already require lab tests and a physician-parent reporting process, so the administrative and reporting costs of adding a meconium screening are assumed to be 20% of the cost of the program for additional facilities, transport, training of personnel and specimen collection. - Data gaps that affect ability to accurately estimate costs and benefits of universal screening: cost of including meconium screening in established new-born screening system; number of women who would voluntarily participate in interventions; long-term effectiveness of each intervention; development of a second-stage screen that reduces false positives; impact of gestational age on the sensitivity/specificity of the meconium test; refinement of test to indicate level of drinking; ability to identify social drinkers; inclusion o FASD children into primary research; relation between binge drinking during pregnancy and alcohol dependence; strategy to reduce false negatives; cross-country generalizability, effect of multiple drug use on effectiveness of intervention. - A universal meconium analysis of new-borns and subsequent intervention for the identified mothers could be a cost-effective intervention strategy to reduce the incidence of FAS and FASD. - Conservatively estimated, savings could range from $97 to $6 per every dollar spent depending on the type of intervention strategy.
Evaluation	- Overall financial impact of FASD is difficult to estimate because of the range of effects that may be evident in the individual. - More attention is needed to incorporate individuals with other forms of FASD into on-going research - Sensitivity and specificity only used with binge drinking mothers, did not take into account social drinkers. - No proposed follow up for test negatives. - Implementation of universal screening is premature at this time.
Notes	- Meconium testing has been demonstrated to be a promising at-birth method for detection of drinking during pregnancy. - Other screening methods conducted during pregnancy may leave the patent feeling threatened or judged and may reduce the veracity of the responses. Under reporting may occur due to stigma, shame, fear of legal repercussions , fear of mandatory placement into detoxification programs and when the interviewer is not specifically trained to conduct such interviews.

Citation	**(Green et al., 2009)** **Green CR, Mihic AM, Nikkel SM, Stade BC, Rasmussen C, Munoz DP, et al. Executive function deficits in children with fetal alcohol spectrum disorders (FASD) measured using the Cambridge Neuropsychological Tests Automated Battery (CANTAB). J Child Psychol Psychiatry. 2009;50 (6):688-97.**
Publication status	Published Journal – Journal of Child Psychology and Psychiatry
Source	Online Database
Country of origin/region	Canada
Level of evidence (NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	III-3 = diagnostic case-control study
Study type/design	
Research question/aim	How does executive function compare in children with FASD and controls?
Intervention/Instrument (Elliott et al., 2008)	Cambridge Neuropsychological Tests Automated Battery (CANTAB)
Instruments mentioned	CANTAB is a standardised, computer-assisted battery of tests. Non-verbal. Touch screen response therefore easy to administer.
Patient population (Elliott et al., 2008)	89 children with pFAS, ARND
Comparator (Elliott et al., 2008)	92 age and sex-matched controls
Outcomes/ Recommendations (Elliott et al., 2008)	Children with FASD had: deficits in planning; deficits in spatial working memory); longer reaction and decision times, suggesting attention deficits; decreased problem solving ability; slower movement times; decreased memory (nearly 50% difference from controls in spatial working memory. Executive function was similar across FASD sub-categories
Evaluation	This was a well conducted study of executive functions using sensitive, non-verbal (language-independent) tests that could be delivered via computer thus making them easy to administer and standardised. They demonstrate that spatial working memory is most abnormal component of executive function in FASD versus controls.
Notes	

Citation	**(Greenbaum, 2000)** **Greenbaum R. Socioemotional functioning in children diagnosed with alcohol related neurodevelopmental disorder (ARND): Profile on the child behaviour checklist (CBCL). Toronto: University of Toronto; 2000.**
Publication status	Thesis (Masters)
Source	Internet
Country of origin/region	Canada
Level of evidence	III-3 = diagnostic case-control study

(NHMRC Interim Levels of Evidence for Evaluating Intervention and Diagnostic accuracy Studies)	
Study type/design	Diagnostic case-control study
Research question/aim	To test hypothesis that children with ARND would present a distinct clinical profile of disturbed social/emotional function compared to controls
Intervention/Instrument (Elliott et al., 2008)	Diagnostic checklist for ARND (clinic tool); Achenbach Child behaviour checklist; ARND diagnosis by IOM criteria.
Instruments mentioned	*See attachment* for Neuropsych battery; also dysmorphology exam; questionnaire for home/social background and education/treatment history
Patient population (Elliott et al., 2008)	Matched pair sample, 33 ARND, 33 control, mean age of ARND 8.36 (range 4-14.83 years)
Comparator (Elliott et al., 2008)	Diagnoses of ARND using IOM criteria, diagnoses based on dysmorphology exam, clinic checklist and neuropsychology test battery.
Outcomes/ Recommendations (Elliott et al., 2008)	CBCL is a sensitive instrument for identifying SE problems and differentiated ARND from controls, but may lack specificity
Evaluation	Gold standard neuropsych assessment for diagnosis of ARND; relatively small sample size; further research needed to test if CBCL could differentiate ARND from other clinical diagnoses e.g. ADHD without alcohol exposure; clinic checklist of assets and deficits is not validated.
Notes	Neuropsych test battery took 4.5 hours to administer to children 4-5 years and 6.5 hours for school age kids; god to include as potential confounders factors related to home/social background and education/treatment history

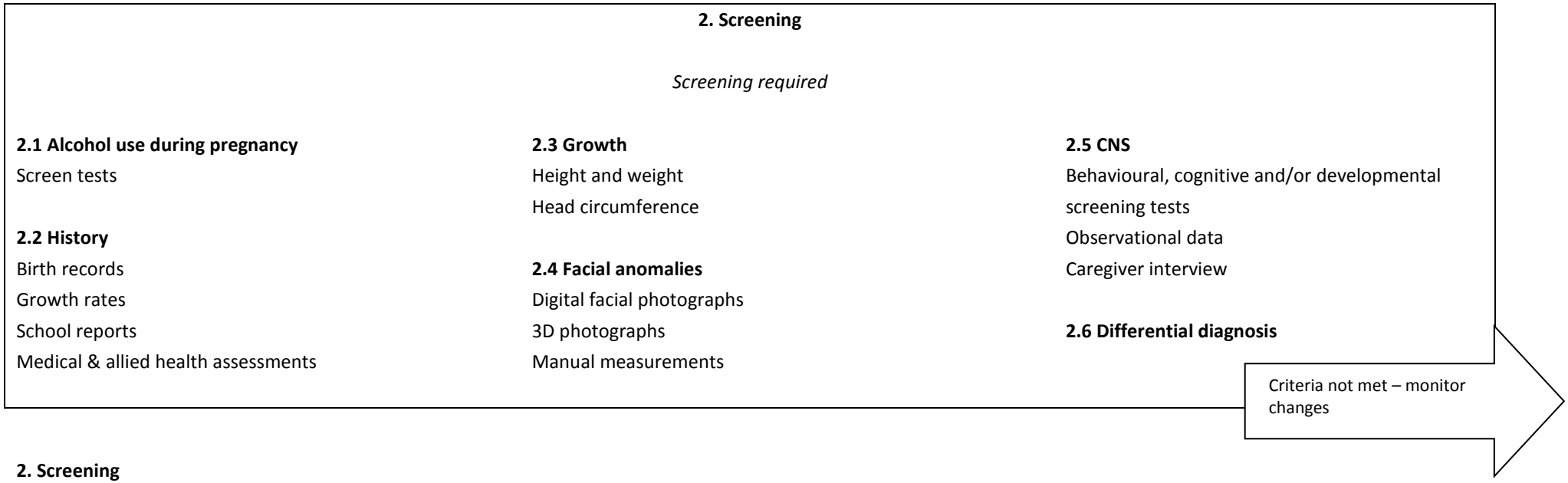
Appendix 5 Tabulated Data for Delphi Development (Full Literature Review)

FASD Project: Delphi Structure and Development

1. Identification		
Suspected alcohol-related effects *Identified from number of sources*		
Parents	School or day-care staff	Paediatricians
Family members	Community/Child Health Nurses	Obstetricians
Carers	Social workers	GPs
Juvenile justice officers	Foster care agencies	

1. Identification

	Statements	**Papers**
1a	Initial recognition that a child or older individual has a potential problem can come from many sources. Often, parents notice differences between a child and his or her siblings. School systems, including Head Start and day-care staff, interact with a large number of children and often recognize when someone is having difficulty. Social service professionals, such as WIC clinic staff, social workers, and foster care agencies frequently recognize children and individuals having difficulty and needing evaluation. And finally, healthcare providers (particularly paediatricians) often are the first to screen for and detect problems; or obstetricians, who might be aware of a maternal substance abuse problem, might refer a newborn. Recognition of many of the problems associated with FAS is exactly the type of condition the "well child" visits to the doctor's office are meant to identify. It is assumed that triggers, such as facial abnormalities, growth delay, developmental problems, or maternal alcohol use, will emerge from the contact. Recognition of a potential problem should lead the provider, regardless of specific profession, to facilitate getting the person and his or her family to the appropriate next step.	(National Center on Birth Defects and Developmental Disabilities, 2004)



2. Screening

	Statements	**Papers**
2a	Screening of the asymptomatic child may be used at a population level to identify children with fetal alcohol spectrum disorders from children known to have been exposed to alcohol.	(E. Elliott & Peadon, 2009)
2b	Include screening for FASD in child health nurse screening assessments of children in the care of child protection	(Department of Health Western Australia, 2010)
2c	There is an opportunity for screening for FASD in early and middle childhood in order to provide intervention and to prevent or minimise adverse outcomes.	(Department of Health Western Australia, 2010)
2d	Identification of one child affected by FASD allows for the opportunity for prevention of second and subsequent children being exposed to alcohol in pregnancy through maternal and family interventions.	(Department of Health Western Australia, 2010)
2e	Universal screening – time points at which screening activity could occur include new-borns and early childhood or at enrolment in full-time education (age 4-6 years).	(Department of Health Western Australia, 2010)
2f	Targeted screening – identifying sub-populations at high risk of the disorder: infants/children of mothers registered with the WA Newborn Drug and Alcohol Service, attending alcohol treatment services and those identified as using alcohol and/or other drugs; babies that are small for gestational age and/or microcephalic; infants/children referred to or in the care of the DCP; children referred to child	(Department of Health Western Australia, 2010)

	development services or Child and Adolescent Mental Health Services, particularly those referred for difficulties with attention, behaviour and social/emotional development; children registered with Disability Services Commission with a diagnosis of intellectual disability (ID) or vulnerable to ID, who do not have an established genetic etiology; children and adolescents referred to Child and Mental Health Services Complex Attention and Hyperactivity Disorders Service.	
2g	Consideration for screening – children referred to school psychology services for learning and behavioural difficulties; youth in juvenile justice settings; regional communities identified as having high levels of alcohol consumption.	(Department of Health Western Australia, 2010)

2.1 Alcohol use during pregnancy

	Feature/Criteria	Measure	Other/Notes	Papers
2.1a	Confirmed exposure to high levels of alcohol		BAC greater than 100mg/dL, weekly early in pregnancy	(S J Astley, 2004) 4-DDC
2.1b	Confirmed exposure to alcohol		Birth mother consumed alcohol during pregnancy but the quantity is unknown Birth mother consumed small amount early on but stopped when learning she was pregnant at 3 months	(S J Astley, 2004) 4-DDC
2.1c	Unknown exposure		Child is adopted and records are closed Birth mother is known to have a problem with drinking but there are no records or direct observation of her drinking during pregnancy	(S J Astley, 2004) 4-DDC
2.1d	Confirmed absence of exposure from conception to birth		Confirmed absence of drinking from conception to birth	(S J Astley, 2004) 4-DDC
2.1e	Alcohol screening questionnaire	T-ACE	5. Tolerance (T) – how many drinks does it take to make you feel high? 6. Annoyance (A) – have people annoyed you by criticising your drinking? 7. Cut down (C) – have you ever felt you ought to cut down on your drinking? 8. Eye-opener (E) – have you ever had a drink first thing in the morning to steady your nerves or get rid of a hangover?	(BMA Board of Science, 2007) Source: Sokol et al (1989)

			A single point is given for an affirmative answer to the A, C and E questions, and two points are given when a pregnant woman indicates a tolerance of more than two drinks to feel high. A total score of two or more on the test is suggestive of harmful drinking patterns during pregnancy.	
2.1f	Alcohol screening questionnaire	T-ACE	How much alcohol do you drink before you feel its effects (**Tolerance**)? Has anyone **Annoyed** you by saying you should cut down on your drinking? Have you ever though you should **Cut Down**? Have you ever had a drink to get going in the morning? **(Eye Opener)**	(First Nations and Inuit Health Committee, 2002)
2.1g	Alcohol screening questionnaire	TWEAK	TWEAK for populations with high levels of binge drinking: 1. Tolerance (T) – how many drinks does it take before the alcohol makes you fall asleep or pas out? Record number of drinks__(a positive score is six or more drinks) OR If you never drink until you pass out, what is the largest number of drinks that you have? Record number of drinks__(a positive score is six or more drinks) 2. Worried (W) – have your friends or relatives worried or complained about your drinking in the past year? 3. Eye opener (E) – do you sometimes take a drink in the morning when you first get up? 4. Amnesia (A) – are there times when you drink and you can't remember what you said or did? 5. Cut down (K) – do you sometimes feel the need to cut down on your drinking? TWEAK for populations with low levels of binge drinking: 1. Tolerance (T) – how many drinks does it take before you begin to feel the first effects of alcohol? Record number of drinks__(a positive score is three or more	(BMA Board of Science, 2007) Source: Chan et al (1993)

			drinks) 2. Worried (W) – have your friends or relatives worried or complained about your drinking in the past year? 3. Eye opener (E) – do you sometimes take a drink in the morning when you first get up? 4. Amnesia (A) – are there times when you drink and you can't remember what you said or did? 5. Cut down (K) – do you sometimes For each version, a positive response to question T or W yields two points each, and an affirmative reply to question E, A or K scores one point each. A total score of two or more points on the TWEAK test is suggestive of harmful drinking patterns during pregnancy.	
2.1h	Maternal alcohol consumption	Retrospective - just before pregnancy and during pregnancy	a) average and maximum number of drinks per drinking occasion b) average number of drinking days per week c) type of alcohol consumed (beer, wine, liquor) d) trimester(s) during which drinking occurred	(S. J. Astley et al., 2009b)
2.1i	Fatty acid ethyl esters (FAEE)	Biomarker screening – maternal hair Meconium analysis	Detects alcohol use in the 6 months before collection ▪ Not validated ▪ Forms from 20 weeks' gestation ▪ Can be collected up to 3 months (>2 nmol/g) detects heavy fetal alcohol exposure. Must be collected within 72 hours of birth. Does not capture first trimester of alcohol consumption ▪ Non-invasive and objective ▪ Limited window of collection and requires specific handling ▪ Only detects alcohol exposure after 12-14 weeks	(E. Elliott & Peadon, 2009) (Goh & Rosenbaum, n.d.) Elliott and Peadon 2009) (Goh & Rosenbaum, n.d.)

2.1j	Fatty acid ethyl esters (FAEE)	Meconium	- State regulated new-born screening programs already require lab tests and a physician-parent reporting process, so the administrative and reporting costs of adding a meconium screening are assumed to be 20% of the cost of the program for additional facilities, transport, training of personnel and specimen collection. - Data gaps that affect ability to accurately estimate costs and benefits of universal screening: cost of including meconium screening in established new-born screening system; number of women who would voluntarily participate in interventions; long-term effectiveness of each intervention; development of a second-stage screen that reduces false positives; impact of gestational age on the sensitivity/specificity of the meconium test; refinement of test to indicate level of drinking; ability to identify social drinkers; inclusion o FASD children into primary research; relation between binge drinking during pregnancy and alcohol dependence; strategy to reduce false negatives; cross-country generalizability, effect of multiple drug use on effectiveness of intervention. - A universal meconium analysis of new-borns and subsequent intervention for the identified mothers could be a cost-effective intervention strategy to reduce the incidence of FAS and FASD. - Conservatively estimated, savings could range from $97 to $6 per every dollar spent depending on the type of intervention strategy. - Meconium testing has been demonstrated to be a promising at-birth method for detection of drinking during pregnancy. - Other screening methods conducted during pregnancy may leave the patent feeling threatened or judged and may reduce the veracity of the responses. Under reporting may occur due to stigma, shame, fear of legal repercussions , fear of mandatory placement into detoxification programs and when the interviewer is not specifically trained to conduct such interviews.	Gifford (2010)

			- Further research needed	
2.1k	Fatty acid ethyl esters (FAEE)	Meconium	- Ethyl oleate was the FAEE that correlated most strongly with maternal self-reported drinking, especially with the average ounces of absolute alcohol ingest per drinking day. - Ethyl oleate was most strongly related to drinking in the second an third trimesters. - Using a cut-off value of 32ng/g, sensitivity was 84.2% and specificity was 83.3%	Bearer (

	Statement	**Papers**
2.1l	When prenatal alcohol exposure is known, a child should be referred for full FAS evaluation when substantial prenatal alcohol use has been confirmed.	(Bertrand et al., 2005)
2.1m	When information regarding prenatal alcohol exposure is unknown, a child should be referred for full FAS evaluation for any one of tho following: - report of concern by parent or caregiver that a child might have FAS - presence of all three facial features - presence of one or more of these facial features with growth deficits in height, weight, or both - presence of one or more facial features with one or more CNS abnormalities, or - presence of one or more facial features, with growth deficits and one or more CNS abnormalities	(Bertrand, et al., 2005)
2.1n	Alcohol exposure should be considered for persons experiencing or have experienced one or more of the following: - premature maternal death related to alcohol use (either disease or trauma) - living with an alcoholic parent - current or previous abuse or neglect - current or previous involvement with child protective services agencies - a history of transient care giving situations, or - foster or adoptive placements (including kinship care)	(Bertrand, et al., 2005)
2.1o	Studies investigating effects of prenatal alcohol consumption should include a clear a-priori definition of what constitutes low, moderate and high levels of maternal alcohol intake.	(Muggli et al., 2010)
2.1p	For accurate reporting of alcohol use: collection of information should be accompanied by a comprehensive and locally relevant pictorial	(Muggli, et al., 2010)

	drinks guide; graphics should represent the number of standard drinks in range of common alcoholic beverages and the guide should be structured without requiring women to estimate their alcohol consumption in standard drinks, but should include conversion factors for the researchers.	
2.1q	Alcohol screening instruments in clinical practice focus on identifying at risk drinkers and are not designed to quantify actual amounts of alcohol consumed. There are currently no clear guidelines for general practitioners and maternity care providers as to how pregnant women should be asked about their alcohol consumption. Recommendation that women should be screened for alcohol intake with a validated clinical instrument that includes assessment of consumption patterns, clear instructions for the practitioner on how to interpret and discuss the information and hand-outs of educational material for the woman.	(Muggli, et al., 2010)
2.1r	To maintain best practice there needs to be systematic prospective data collection of information about alcohol use for every pregnancy; and systematic retrospective data collection of information about alcohol use in pregnancy for every child identified with a developmental disability.	(Department of Health Western Australia, 2010)

2.2 History

	Feature/Criteria	**Measure**	**Other/Notes**	**Papers**
2.2a	Potential genetic conditions, exposures or prenatal conditions		Associated with physical or neurodevelopmental problems Poor prenatal care, patients whose parents have mild mental retardation, attention deficit disorders, significant learning disabilities or learning problems, prenatal exposure to drugs during pregnancy Some risk	(S J Astley, 2004) 4-DDC
2.2b	Circumstances that have significant effect on development		Clear physical and sexual abuse, multiple disrupted placements with clear impact on the child, neglect resulting in failure to thrive, serious head injury, or medical conditions which lead to brain damage High risk	(S J Astley, 2004) 4-DDC
2.2c	Other risk factors		When historical information is missing Unknown risk	(S J Astley, 2004) 4-DDC

	Statement	Papers
2.2d	Detailed alcohol histories are frequently unavailable on patients presenting to a FASD diagnostic clinic.	(S. J. Astley, et al., 2009b)
2.2e	The pattern and severity of diagnostic outcome is dependent on the timing, frequency, and quantity of alcohol exposure	(S J Astley, 2004) 4-DDC
2.2f	The diagnostic outcome is frequently confounded by other adverse prenatal and postnatal exposures and events	(S J Astley, 2004) 4-DDC
2.2g	Maternal history should include alcohol consumption (amount and frequency during pregnancy), drug and teratogen exposure during pregnancy (including illicit and prescribed drugs to rule out differential diagnoses) and any medical conditions. A family history may identify genetic disease, familial birth defects, or patterns of malformation. Information on gestation, intrauterine growth retardation, birth weight, birth order, and birth defects should be ascertained.	(E. Elliott & Peadon, 2009)
2.2h	When details of the family background and gestation are unknown Unknown risk	(S J Astley, 2004) 4-DDC
2.2i	Information such as birth records, growth records, history of out-of-home care if relevant, school reports, and medical and allied health assessment reports should be obtained if available	(Department of Health Western Australia, 2010)

2.3 Growth

	Feature/Criteria	Measure	Other/Notes	Papers
2.3a	Height	Ruler	Should be age and gender adjusted Adjustment for mid-parent stature when both parents' heights are known	(S J Astley, 2004) 4-DDC
2.3b	Weight	Scales	Should be age and gender adjusted	(S J Astley, 2004) 4-DDC
2.3c	Height, weight or both		Should be <10th percentile, adjusted for age, sex, gestational age, and race or ethnicity	(Bertrand, et al., 2005)

2.3d	Head circumference			(E. Elliott & Peadon, 2009)
2.53e	Growth retardation		▪ Confounded by variation between populations ▪ Only small proportion of children with FASD affected (10-30%) ▪ May be useful in combination with other screening tools	(Goh et al., 2008)

	Statement	**Papers**
2.3f	Growth records should be separated into prenatal growth and postnatal growth; and the growth record with the greatest deficiency in the height percentile should be selected.	(S J Astley, 2004) 4-DDC
2.3g	The prevalence and severity of growth deficiency generally increases from controls to FAS/PFAS	(S. J. Astley, et al., 2009b)

2.4 Facial anomalies

	Feature/Criteria	**Measure**	**Other/Notes**	**Papers**
2.4a	Small palpebral fissure lengths	Direct measurement - clear plastic ruler Digital facial photograph	2 or more standard deviations below the mean <10^{th} percentile	(S J Astley, 2004) 4-DDC (Bertrand, et al., 2005)
2.4b	Smooth Philtrum	Lip-Philtrum Guide (Caucasian or African American) Digital photograph	Rank 4 or 5 on the Lip-Philtrum Guide	(S J Astley, 2004) 4-DDC
2.4c	Thin upper lip	Lip-Philtrum Guide (Caucasian or African American) Digital photograph	Rank 4 or 5 on the Lip-Philtrum Guide	(S J Astley, 2004) 4-DDC
2.4d	Ear		Underdeveloped upper part of the ear parallel to the ear crease	(BMA Board of

			below. “Railroad track” appearance Large ears with “railroad-track” ear abnormality	Science, 2007) (E. Elliott & Peadon, 2009)
2.4e	Other features		Flat midface; epicanthic folds; hypertelorism (wide-spaced eyes); ptosis (droopiness of the eyelids); micrognathia (undersized jaw); microphthalmia (small eyes); and cleft lip and/or palate	(E. Elliott & Peadon, 2009)
2.4f	Facial phenotype		Sensitivity estimated at 99-100%, specificity 64-99%, Positive predictive value 85.7%, Negative predictive value 100% Possible underestimation of palpebral fissure length, especially under age 4 years	(Goh & Rosenbaum, n.d.)
2.4g	Facial phenotype	Facial Photographic Screening Tool	Screen: FAS facial photographic screening tool Occipitof-rontal circumference FAS photographic screening tool Positive predictive value is (6 / 7) 85.7% FAS photographic screening tool Negative predictive value is (590 / 590) 100% FAS photographic screening tool Sensitivity (6/6) 100% FAS photographic screening tool Specificity (590/591) 99.8% FAS photographic screening tool Accuracy (596/597) 99.8% Recommends use of the FAS photographic screening tool as cost-effective and accurate in a high risk population (those in foster care) Recommends nesting screening programs in existing health care / monitoring strategies	(S. J. Astley et al., 2002)
2.4h	Facial phenotype	Digital facial photography and manual measurements	Digital facial Photographs 100% sensitive and 64% specificity with no false negatives Digital PFL measurements were significantly different from direct	(Avner et al., 2006)

			manual measurements in children under the age of four years; (digital PFL in children under four years of age tends to underestimate the PFL). Digital PFL measurements were not significantly different from direct technique manual measurement in children 4 years of age and older. Direct measurement scores for philtrum smoothness were different from the digital on frontal view alone but not different when considering three quarter view of the philtrum	
2.4i	Facial features	Gestalt approach to diagnosis	Patients were classified into **1** of **4** categories defined as follows: FAS: Reported in utero alcohol exposure, CNS dysfunction, distinct presentation of the FAS facial phenotype, with or without documented growth deficiency. 1. AFAS (atypical fetal alcohol syndrome): Reported in utero alcohol exposure, CNS dysfunction, mild presentation of the FAS facial phenotype, 2. with or without documented growth deficiency. 3. PFAE: Reported in utero alcohol exposure, CNS dysfunction, absence of the FAS facial phenotype, with or without documented growth deficiency. - Other: In utero alcohol exposure reported or suspected, but no diagnosis of FAS, AFAS, or PFAE was made because of the absence of both FAS-like facial anomalies and CNS dysfunction Assessment of diagnostic inter-rater agreement between trained dysmorphologists and testing in other clinic populations will be needed to assess the tool's external validity. Useful screening tool for facial dysmorphology – with further research required.	(S. J. Astley & Clarren, 1995)

	Statement	Papers
2.4j	The FAS phenotype is not simply present of absent.	(S. J. Astley, et al., 2009b)
2.4k	The magnitude of expression of the FAS facial phenotype is highest among the FAS/PFAS group; significantly lower in the SE/AE and ND/AE groups but significantly higher than the control group.	(S. J. Astley, et al., 2009b)

2.5 CNS

	Feature/Criteria	Measure	Other/Notes	Papers
2.5a	Behavioural, cognitive and/or developmental problems	Standardized psychometric tests, observational data, and/or caregiver interview	Delay and/or dysfunction that suggests the possibility of CNS damage.	(S J Astley, 2004) 4-DDC
2.5b	Head circumference	Measure tape	Occipital frontal circumference 2 or more standard deviations below the mean	(S J Astley, 2004) 4-DDC

	Statement	Papers
2.5c	Many individuals with prenatal alcohol exposure exhibit cognitive difficulties and significant maladaptation that prevent them from leading productive, independent lives.	(Stratton et al., 1996)
2.5d	Neuropsychological/behavioural problems stem from the prenatal brain damage.	(S. J. Astley, et al., 2009b)
2.5e	Psychiatric disorders are prevalent across the FASD groups and significantly more prevalent than among controls.	(S. J. Astley, et al., 2009b)
2.5f	Prevalence of impairment – 20-50% of children with FAS/PFAS performs significantly below the population mean in any single domain of function. A comparable prevalence of impairment was observed among the children in the SE/AE group. Prevalence was markedly less in the ND/AE group and absent in the control group.	(S. J. Astley, et al., 2009b)

2.6 Differential Diagnosis

	Facial features	Syndromes	Papers
2.6a	Smooth philtrum	Cornelia de Lange syndrome Floating-Harbor syndrome	(National Center on Birth Defects and

		Geleophysic dysplasia Opitz syndrome Toluene embryopathy	Developmental Disabilities, 2004)
2.6b	Thin Vermillion border	Miller-Dieker (Lissencephaly) syndrome Fetal Valproate syndrome Geleophysic dysplasia Cornelia de Lange syndrome Toluene embryopathy	(National Center on Birth Defects and Developmental Disabilities, 2004)
2.6c	Small palpebral fissures	Campomelic dysplasia DiGeorge sequence Dubowitz syndrome Duplication 10q sequence Duplication 15q sequence FG syndrome Maternal phenylketonuria (PKU) fetal effects Oculodentodigital syndrome Opitz syndrome Trisomy 18 syndrome Williams syndrome Velocardiofacial syndrome Toluene embryopathy	(National Center on Birth Defects and Developmental Disabilities, 2004)

3. Referral

3. Referral

3a	The referral process is initiated at the point a clinician starts to have suspicions of an alcohol-related disorder for a child. This process is facilitated by thorough knowledge of the physical and neurodevelopmental domains affected in individuals with FAS, as well as characteristics that could trigger a referral. Examples of triggers are presented later, in the Referral section of these guidelines. In making a referral for a complete diagnostic evaluation for FAS, it is helpful for the referring provider to gather and document specific data related to the FAS criteria. These data will assist the provider in making the decision to diagnose the child or to refer the child to a multidisciplinary evaluation team for a confirmed diagnosis. In addition, these data could be forwarded to the multidisciplinary evaluation team to guide the diagnostic process. A complete review of systems, noting features consistent with FAS, would be most productive.	(National Center on Birth Defects and Developmental Disabilities, 2004)
3b	Referral – this process is initiated at the point a clinician starts to suspect an alcohol-related disorder for a child.	(National Center on Birth Defects and Developmental Disabilities, n.d.)
3c	Refer children with suspected FASD to appropriate assessment and intervention services	(Department of Health Western Australia, 2010)
3d	If FASD referral criteria are not met, continue to monitor changes in child's health over time	(National Center on Birth Defects and Developmental Disabilities, 2004)

4. Diagnosis

Suspected alcohol-related disorder

Diagnosis required by health professional

4.1 Facial anomalies

Analysis of photographs

4.2 Screening results

4.3 CNS

Neurological assessments

Functional assessments

4.4 CNS

Structural assessments

4.5 Other

Cardiac anomalies

Musculoskeletal anomalies

Renal anomalies

Ocular anomalies

4.6 Differential diagnosis

4. Diagnosis

	Statements	**Papers**
4a	At this stage, the child would be presented to a multidisciplinary team who would engage in a more thorough assessment of the child using FAS diagnostic procedures to evaluate dysmorphia and growth parameters, as well as obtain appropriate neurodevelopmental evaluation data. Once a diagnosis is made, an intervention plan would be developed using a multidisciplinary team approach. A variety of specialists could contribute to the multidisciplinary team, including dysmorphologists, developmental pediatricians, psychiatrists, psychologists, social workers, and educational specialists. Other clinicians, such as pediatricians and family practitioners, also might make the FAS diagnosis, with appropriate training in use of these guidelines. In many rural and less populated regions, these clinicians must make the diagnosis for many types of birth defects and developmental disabilities. Many of these evaluation services are available within the community setting, for example school systems could provide neurocognitive assessments.	(National Center on Birth Defects and Developmental Disabilities, 2004)
4b	Professionals from multiple disciplines are needed to accurately assess and interpret the road array of outcomes that define the diagnoses	(S J Astley, 2004) 4-DDC
4c	Interrater reliability refers to the ability of two clinicians to look at the same phenomena and reach similar diagnostic conclusions. At least two clinicians should be involved in evaluating clinical history, facial features, examine other ancillary data and reach precisely the same diagnosis	(Stratton, et al., 1996) IOM
4d	Diagnosis – multidisciplinary team thoroughly assess the child using FAS diagnostic procedures to devalue dysmorphia (abnormality of shape or form) and growth parameter and to obtain appropriate neurodevelopmental evaluation data.	(National Center on Birth Defects and Developmental

		Disabilities, n.d.)
4e	Information from multiple sources (school records, hospital record, social services, previous assessments) should be obtained; this might involve meeting with relevant professionals who know the patients (teachers, physicians, social workers, psychologists). Other relevant documentation would include birth and pregnancy records, medial and hospital records, adoption records, academic records, achievement tests, developmental assessments, psychological and psychometric assessments, legal reports and documentation of the family history.	(Chudley et al., 2005)
4f	A full diagnostic evaluation should only be performed by a trained specialist, and often requires a multi-disciplinary team.	(L. Elliott et al., 2008)

4.1 Facial anomalies

	Feature/Criteria	**Measure**	**Other/Notes**	**Papers**
4.1a	Small palpebral fissure lengths	Digital facial photograph – computerised analysis	2 or more standard deviations below the mean <10th percentile	(S J Astley, 2004) 4-DDC (Bertrand, et al., 2005)
4.2b	Smooth Philtrum	Digital photograph – FAS Facial Photographic Analysis Software		(S J Astley, 2004) 4-DDC
4.2c	Thin upper lip	Digital photograph – FAS Facial Photographic Analysis Software		(S J Astley, 2004) 4-DDC
4.2d	Facial dysmorphology		3. Manual measurement 4. Digital Photo analysis software Both these methods are sensitive, specific and with high positive predictive value but only for FAS and pFAS.	(Goh, et al., 2008)
4.2e	Facial phenotype	Facial Photographic Screening Tool	Diagnosis: Attendance at a FAS DPN multidisciplinary clinic for assessment including FAS facial photography Image analysis software was used (Astley) to measure FAS facial phenotype from digital images.	(S. J. Astley, et al., 2002)
4.2f	Facial phenotype	3D facial image analysis	- 2 study populations – one form Finland, one from South Africa; Finnish Caucasian 36 kids with FAS, 31 controls, age range 2.8-21	(Fang et al., 2008)

			years (mean 13 years); Cape Coloured pop 50 kids with FAS, 32 controls, mean age 5 years. - Finnish pop Sensitivity 88.2%, Specificity 100%; Cape Coloured pop Sensitivity 91.7%, Specificity 90%. - Good sensitivity, specificity within same ethnic group, lower test performance on missed populations; more expensive technology; less portable than a camera, further research needed to determine effects of age on facial features.	

	Statement	**Papers**
4.2g	Astley and Clarren (2000) and Hoyme et al (2005) have confirmed, using two large clinical datasets, that the majority of individuals diagnosed with FAS by a gestalt approach lose that diagnostic classification when more rigorous diagnostic guidelines are applied.	(S. J. Astley, et al., 2009b) Source: (S. J. Astley & Clarren, 2000) (Hoyme et al., 2005)
4.2h	A dysmorphologist, using a *gestalt* approach, correlates the computerized data with clinical diagnosis to calculate sensitivity and specificity of the screening tool. This method has been found to have a specificity of 100% and sensitivity of 100% in clinical settings.	(Burd et al., 2000) Source: (S. J. Astley & Clarren, 1995)
4.2i	In a child with abnormal facial features, a digital photograph can be used in conjunction with facial diagnostic software to aid confirmation of diagnosis.	(E. Elliott & Peadon, 2009)
4.2j	Phenotype varies with age and makes it more difficult to identify appropriate features for inclusion in diagnostic criteria that are not age specific.	(Stratton, et al., 1996) IOM

4.2 Screening Results

	Statement	**Papers**
4.2a	Birth records, growth records, history of out-of-home care if relevant, school reports, and medical and allied health assessment reports should be used in diagnostic assessment. This will allow the multi-disciplinary team can more effectively plan the assessment process and avoid duplication	(Department of Health Western Australia, 2010)

4.3 CNS - Functional/Neurological

	Feature/Criteria	Measure	Other/Notes	Papers
4.3a	Seizures	EEG	Not due to a postnatal insult or other postnatal process Definite CNS damage = at least one significant finding that is 2 or more standard deviations below the norm if measured on a standardised scale or when assessed by clinical radiologist or neurologist.	(S J Astley, 2004) 4-DDC (E. Elliott & Peadon, 2009)
4.3b	Executive function, memory, cognition, social/adaptive skills, academic achievement, language, motor, attention or activity level	Standardized, validated psychometric assessments: WISC-III, WAIT-II, TOLD, PLS3, D-KEFS, VMI-II	Probable CNS damage = significant impairment across three or more domains. Administered directly to the affected individual or obtained from reliable informants and interpreted by qualified professionals (psychologists, psychiatrists, occupational therapists, speech-language pathologists)	(S J Astley, 2004) 4-DDC
4.3c	Intellectual functioning	Wechsler Intelligence Scale for Children (WISC-III) Wechsler Preschool and Primary Scale of Intelligence (WPPSI-R) Wechsler Adult Intelligence Scale (WAIS-III) McCarthy Scales of Children's Abilities	School age Pre-school age Adult Pre-school age	(Greenbaum, 2000)
4.3d	Memory	Wide Range Assessment of Memory and Learning (WRAML) Denman Neuropsychological Memory Test	School age Adult	(Greenbaum, 2000)
4.3e	Language	Peabody Picture Vocabulary Test (PPVT-IIIR) Expressive One Word Picture Vocabulary Test (EOWPVT-R) Preschool Language Scale (PLS-3) Token Test for Children	All ages Preschool/school age Preschool age Preschool/school age	(Greenbaum, 2000)

		Test for the Reception of Grammar (TROG) Test of Language Development-2 (TOLD2) Test of Language Competence (TLC)	Preschool/school age School age School age/adult	
4.3f	Visual-Motor Coordination	Beery-Buktenica Developmental Test of Visual-Motor Integration 4th Edition (VMI) Grooved Pegboard	All ages School age/adult	(Greenbaum, 2000)
4.3g	Attention/Executive functioning	Connor Continuous Performance Test (CPT) TRAILS A&B Wisconsin Card Sorting Test Verbal Fluency Test	All ages School age/adult School age/adult School age/adult School age/adult	(Greenbaum, 2000)
4.3h	Academic Functioning	Wide Range Achievement Test (WRAT-3) EINSTEIN Woodcock Reading and Mastery Tests Keymath: Time and Money	School age/adult Preschool age School age/adult School age/adult	(Greenbaum, 2000)
4.3i	Behaviour and Socioemotional functioning	Conners' Parent Rating Scale (CRS-R) Carey Temperament Scales Werry Weis Peters Activity Scale Child Behaviour Checklist (CBCL)	All ages Preschool/school age Preschool/school age All ages	(Greenbaum, 2000)
4.3j	Soft Neurological Signs	Quick Neurological Screening Test II (QNST-II)		(S. J. Astley, et al., 2009b)
4.3k	General Intellectual Function	Wechsler Intelligence Scale for Children (WISC-III)		(S. J. Astley, et al., 2009b)
4.3l	Academic Achievement	Wechsler Individual Achievement Test (WAIT) Basic Reading subset KeyMath Revised/NU: A Diagnostic Inventory of Essential Mathematics		(S. J. Astley, et al., 2009b)
4.3m	Visuospatial Skills, Visual	Beery-Buktenica Developmental Test of Visual-		(S. J. Astley, et al.,

	Memory and Organisation	Motor Integration (VMI) Rey Complex Figure Test (RCFT)		2009b)
4.3n	Executive Function	Delis-Kaplan Executive Function System (D-KEFS) Wisconsin Card Sorting Test: Computer Version 3 (WCST) Research Edition	Trail Making Test Tower Test Color-Word Interference Test Verbal Fluency Test: Standard Form	(S. J. Astley, et al., 2009b)
4.3o	Verbal Memory	California Verbal Learning Test-Children's Version (CVLT-C)		(S. J. Astley, et al., 2009b)
4.3p	Attention	Integrated Visual and Auditory Continuous Performance Test (IVA CPT)		(S. J. Astley, et al., 2009b)
4.3q	Receptive and Expressive Language	Test of Language Development-Intermediate: Third Edition (TOLD-I:3) Test of Language Competence-Expanded Edition (TLC-1-Expanded) Level 1 Test of Language Competence-Expanded Edition (TLC-2-Expanded) Level 2 Test of Word Knowledge (TOWK)	Sentence Combining subtest (subjects 8-10 years) Oral Expression: Recreating Speech Arts subtest (subjects 8-9 years) Oral Expression: Recreating Sentences subtest (subjects 10-15.9 years) Conjunctions and Transition Words subtest (subjects 11-15.9 years)	(S. J. Astley, et al., 2009b)
4.3r	Adaptive Behaviour	Vineland Adaptive Behavior Scales (VABS) Interview Edition, Survey Form		(S. J. Astley, et al., 2009b)
4.3s	Behaviour Problems and Social Competence	Child Behavior Checklist (CBCL/6-18)	For Ages 6-18	(S. J. Astley, et al., 2009b)
4.3t	Caregiver Report of Behaviours Related to Executive Function	Behavior Rating Inventory of Executive Function (BRIEF)		(S. J. Astley, et al., 2009b)
4.3u	Psychiatric Conditions	Computerized Diagnostic Interview Schedule for Children: Parent Form (C-DISC)		(S. J. Astley, et al., 2009b)
4.3v	CNS Dysfunction	Standardised neuropsychological tests administered by professionals	Severe dysfunction is defined by the presence of three or more domains (e.g. cognition, executive function, language, memory, attention etc) of brain function, tow or more standard	(S. J. Astley, et al., 2009b)

			deviations below the norm	
4.3w	Neurologic		Neurologic problems (motor problems or seizures) not resulting from a postnatal insult or fever, or other soft neurologic signs outside normal limits	(Bertrand, et al., 2005)
4.3x	Functional		Test performance substantially below that expected for a person's age, schooling, or circumstances, as evidenced by either: 1) Global cognitive or intellectual deficits representing multiple domains of deficit (or substantial developmental delay in younger children) with performance below the third percentile (i.e. two standard deviations below the mean of standardised testing), OR 2) Functional deficits <16th percentile (i.e. one standard deviation below the mean for standardised testing) in at least three of the following domains: - cognitive or developmental deficits or discrepancies - executive functioning deficits - motor functioning delays - problems with attention or hyperactivity - social skills, or - other (e.g. sensory problems, pragmatic language problems, or memory deficits).	(Bertrand, et al., 2005)
4.3y	Cognition	WPPSI-III: Wechsler Preschool and Primary Scale of Intelligence Alternate: DAS: Differential Ability Scales WISC-IV: Wechsler Intelligence Scale for Children WAI-III: Wechsler Adult Intelligence Scale	For age groups 4-6 years 6-16 years >16 years	(Canada Northwest FASD Research Network, 2007a)
4.3z	Academic Achievement	BBCS-R: Bracken Basic Concept Scale-Revised School Readiness Composite	4-6 years	(Canada Northwest FASD Research Network, 2007a)

		Alternate: DAS: Differential Ability Scales Math: WAIT-II: Wechsler Individual Achievement Test Reading: WIAT-II: Wechsler Individual Achievement Test Spelling: WIAT-II: Wechsler Individual Achievement Test Written Expression (story only): TOWL-3: Test of Written Language Alternate: WJ-R to WJ III: Woodcock-Johnson Test of Achievement WRAT-4: Wide Range Achievement Test Alternate: WJ-R to WJ III: Woodcock-Johnson Test of Achievement	6-16 years >16 years	
4.3aa	Memory	NEPSY Learning and Memory WRAML2: Wide Range Assessment of Memory and Learning WRAML2: Wide Range Assessment of Memory and Learning Supp: CAVLT: Children's Auditory Verbal Learning Test, OR CVLT-C: California Verbal Learning Test-Children's Version WARML 1-2: Wide Range Assessment of Memory and learning Supp: RAVLT: Rey Auditory Verbal Learning Test, OR CVLT-II: California Verbal Learning Test 2nd Ed	4-6 years 6-16 years >16 years	(Canada Northwest FASD Research Network, 2007a)
4.3ab	Executive Functioning &	BRIEF-P: Behaviour Inventory of Executive Function,	4-6 years	(Canada Northwest

	Abstract Reasoning	Preschool Version NEPSY: Attention and Executive Functioning NEPSY II: Second edition for <6 BRIEF: Behaviour Rating Inventory of Executive Function RCFT: Rey Complex Figure Test WISC-IV Digit Span Backwards and Letter-Number Sequencing D-KEFS: Delis-Kaplan Executive Function System (>8). Subsets: verbal, fluency, design fluency, color-word interference, sorting. Children's Color Trials Test WRAML-2 Verbal and Symbolic Working Memory BRIEF: Behavior Rating Inventory of Executive Function RCFT: Rey Complex Figure Test WISC-IV Digit Span Backwards and Letter-Number Sequencing DKEFS: Delis-Kaplan Executive Function System (>8). Subsets: verbal, fluency, design fluency, color-word interference, sorting. Color Trails Test WRAML-2 Verbal and Symbolic Working Memory	6-16 years >16 years	FASD Research Network, 2007a)
4.3ac	Attention & Hyperactivity	BASC-2: Behavior Assessment System for Children	4-6 years, 6-16 years, >16 years	(Canada Northwest FASD Research Network, 2007a)
4.3ad	Adaptive Behaviour	ABAS-II Adaptive Behavior Assessment System VABS-II Vineland Adaptive Behavior Scale Choice of measure depends on situation:	4-6 years	(Canada Northwest FASD Research Network, 2007a)

		- limitation of time - parent literacy - age of child - need for an interview rather than a questionnaire ABAS Adaptive Behavior Assessment System VABS Vineland Adaptive Behavior Scale Choice of measure depends on situation: - limitation of time - parent literacy - age of child - need for an interview rather than a questionnaire	6-16 years, >16 years	
4.3ae	Sensory Motor Motor: Sensory:	The Peabody Developmental Motor Scales–Second Edition (PDMS-2) The Bruininks-Oseretsky Test of Motor Proficiency–Second Edition (BOT-2) (full form) BOT-2 (short form) Short Sensory Profile (SSP) Sensory Profile Caregiver Questionnaire could be used for supplementary information Short Sensory Profile Adult Adolescent Self-Questionnaire, with the	4-11 years Selected as the most age appropriate motor-based assessment providing information on visual motor integration, manual dexterity and gross motor skills. 5-18 years Most appropriate overall motor assessment providing information on both fine and gross motor skills Also considered an acceptable option for those situations where time constraints or the client's attention span might limit the amount of time allotted to complete the assessment. 4 years 5-10 years 11/12 – 18 years	(Canada Northwest FASD Research Network, 2007b)2

		option of having a caregiver assist the adolescent when filling out the questionnaire		
4.3af	Communication	Core Language: CELF-P2 Narrative Language (Bus Story) Expressive language: PLAI-II Receptive language: CELFP-2 Pragmatics checklist CELF-4, TNL TOPS-2Elementary Pragmatics Profile CELF-4 for core language, receptive and expressive TOPS – Adolescent or TOPS Elementary, Pragmatics Profile of CELF-4 and Word Definitions of CELF-4 for 12 year olds CASL for inferred and non-literal language	4-6 years 6-11 years 12+ years	(Canada Northwest FASD Research Network, 2007a) 2
4.3ag	Neurocognitive tools		4. Fetal Alcohol Behavior Scale – not able to discriminate between FASD and other clinical groups 5. CBCL – (modified using 7 sensitive and specific distinguishing items) ▪ Not replicated in a large population ▪ Potential for user bias ▪ Overlaps with other neurobehavioural disorders 6. Personality Inventory for Children – can only be administered by psychologists	(Goh, et al., 2008)
4.3ah	Executive function deficits	Cambridge Neuropsychological Tests Automated Battery (CANTAB)	Standardised computer-assisted battery of tests. Non-verbal, touch screen response and easy to administer. Children with FASD had: deficits in planning; deficits in spatial working memory); longer reaction and decision times, suggesting attention deficits; decreased problem solving ability;	(Green et al., 2009)

			slower movement times; decreased memory (nearly 50% difference from controls in spatial working memory. Executive function was similar across FASD sub-categories	

	Statement	Papers
4.3ai	MRI, MRS and fMRI offer non-invasive methods for in vivo assessment of neuroabnormalities.	(S. J. Astley, et al., 2009b)
4.3aj	Neuropsychological performance among the FAS/PFAS and SE/AE groups was comparably impaired – but significantly more impaired than the ND/AE and control groups. The ND/AE group was almost always significantly less impaired than the FAS/PFAS and SE/AE groups, and significantly more impaired than the control group.	(S. J. Astley, et al., 2009b)
4.3ak	The ND/AE group did not show significant differences from the control group on direct testing measures of executive function	(S. J. Astley, et al., 2009b)
4.3al	Children are not distinguishable solely by their neuropsychological profiles. While children within a group (FAS/PFAS; ND/AE; SE/AE) share the same magnitude of neuropsychological impairment, no two children necessarily shared the same pattern of impairment.	(S. J. Astley, et al., 2009b)
4.3am	Simple IQ tests are inadequate to differentiate children with ARND from those with developmental disabilities resulting from other causes. There is an emerging consensus that children with ARND are markedly impaired in executive functioning, however, these children perform in the normal range with relatively simple tests.	(Hoyme, et al., 2005)
4.3an	Childhood Behavior Checklist: CBCL: Modification proposed as a screening tool with 2 step approach, not been replicated in a large sample size nor across different populations.	(Goh & Rosenbaum, n.d.)

4.4 CNS - Structural

	Feature/Criteria	Measure	Other/Notes	Papers
4.4a	Brain abnormalities	Imaging techniques	Hydrocephaly, heterotopias, change in shape and/or size of brain regions Definite CNS damage = at least one significant finding that is 2 or more standard deviations below the norm if measured on a standardised scale or when assessed by clinical radiologist or neurologist.	(S J Astley, 2004) 4-DDC
4.4b	Size of total brain, frontal lobe, caudate, hippocampus, putamen, corpus callosum and	MRI, MRS and fMRI Using General Electric 1.5 Tesla scanner	MRI (MRI, functional MRI and MR spectroscopy (MRS) – not validated	(S. J. Astley, et al., 2009b) (Goh & Rosenbaum,

	cerebellar vermis			n.d.)
4.4c	Structural brain differences	MRI	to FAS / PFAS, the mean absolute size of the total brain, frontal lobe, caudate, putamen, hippocampus, cerebellar vermis, and corpus callosum length decreased incrementally and significantly. The FAS / PFAS group (the only group with the 4-Digit FAS facial phenotype) had disproportionately smaller frontal lobes relative to all other groups. Magnetic resonance imaging provided further validation that ND / AE, SE / AE, and FAS / PFAS as defined by the FASD 4-Digit Code are 3 clinically distinct and increasingly more affected diagnostic sub classifications under the umbrella of FASD.	(S. J. Astley et al., 2009a)
4.4d	Concentration of neuroabnormalities	MRS	1. Choline, a marker of cell membrane stability and myelination 2. N-acetyl aspartate, a neuronal or axonal marker 3. Creatine, a marker of metabolic activity	(S. J. Astley, et al., 2009b)
4.4e	Abnormalities in brain metabolism	Magnetic Resonance Spectroscopy. Neurobehavioural testing (in the areas of executive functioning, visual-spatial skills, Visual memory (Rey complex figure test), Verbal memory (California Verbal Learning Test), working memory (N-back task), academic achievement, speech/language ability and attention.	– Compared with controls, children with FAS or PFAS had reduced concentrations of choline-containing compounds (reflecting cell membrane stability and myelination in frontal/parietal white matter regions lateral to the midsection of the corpus callosum. – Increasing expression of the FAS facial features and neurobehavioural impairment correlated with decreased choline concentration. – No significant decrease in choline concentration in the hippocampus. – No significant decrease in NAA or Cre concentrations. – Raises the usefulness of neuroimaging/neurometabolic studies in identifying specific brain regions affected by alcohol exposure, and their correlation with functional impairment.	(S. J. Astley et al., 2009c)

4.4f	Neuroactivation in brain regions	fMRI	(anterior cingulated; anterior and posterior parietal lobe; and the dorsolateral prefrontal, inferior frontal, middle frontal, and precentral regions of the frontal lobe) during performance of N-back working memory tasks	(S. J. Astley, et al., 2009b)
4.4g	Structural abnormalities		Head circumference <10th percentile, adjusted for age and sex Clinically meaningful brain abnormalities observable through imaging (e.g. reduction in size or change in shape of the corpus callosum, cerebellum, or basal ganglia)	(Bertrand, et al., 2005)
4.4h	Brain abnormalities	MRI neuroimaging; Voxel-based morphometric analysis of tissue density; diffusion tensor imaging	Promising technique to correlate structural brain abnormalities with neuropsych deficits	(Spadoni et al., 2007)
4.4i	White matter integrity & neurobehavioural correlates	Behavioural data MRI scan acquisition DTI T1-weighted series	- Lower fractional anisotropy (FA) observed in individuals with FASDs relative to controls in Right temporal lobe and bilaterally in aspects of the splenium and corpus callosum. - Loss of white matter density in some but not all regions of reduced FA. - Significant correlations between performance on a test of visuomotor integration and FA in bilateral spenium but not temporal regions were observed within the FASD group. - Correlations between the visuomotor task and FA within the splenium were not significant within the control group and were not significant for measures of reading ability. - This suggests that this region of white matter is particularly susceptible to damage from prenatal alcohol exposure and that disruption of splenial fibers in this group in associated with poorer visumotor integration. - (paper demonstrates associations between function and structure)	(Sowell et al., 2008)

	Statement	Papers
4.4j	MRI, MRS and fMRI offer non-invasive methods for in vivo assessment of neuroabnormalities.	(S. J. Astley, et al., 2009b)
4.4k	Neuropsychological performance among the FAS/PFAS and SE/AE groups was comparably impaired – but significantly more impaired than the ND/AE and control groups. The ND/AE group was almost always significantly less impaired than the FAS/PFAS and SE/AE groups, and significantly more impaired than the control group.	(S. J. Astley, et al., 2009b)
4.4l	The ND/AE group did not show significant differences from the control group on direct testing measures of executive function	(S. J. Astley, et al., 2009b)

4.5 Other Risk Factors

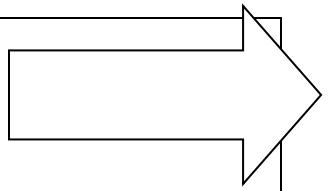
	Feature/Criteria	Measure	Other/Notes	Papers
4.5a	Cardiac anomalies	ECG Echocardiogram	Atrial septal defects; ventricular septal defects; aberrant great vessels; tetralogy of Fallot; conotruncal defects EEG not validated	(E. Elliott & Peadon, 2009) (Goh & Rosenbaum, n.d.)
4.5b	Musculoskeletal anomalies	x-ray	Hypoplastic nails; shortened fifth fingers; radioulnar synostosis; flexion contractures; camptodactyly; clinodactyly of the fifth finger; pectus excavatum or carinatum; Klippel-Feil syndrome; hemivertebrae; scoliosis; hockey-stick palmar creases	(E. Elliott & Peadon, 2009)
4.5c	Renal anomalies	Renal ultrasonography	Aplastic, dysplastic or hypoplastic kidneys; ureteral duplication; hydronephrosis; horseshoe kidneys	(E. Elliott & Peadon, 2009)
4.5d	Ocular anomalies		Strabismus; retinal vascular anomalies; refractive problems Ocular motor testing (assesses executive function) ▪ Saccadic reaction times – children with FASD had prolonged reaction times, excessive direction error and no express saccades compared to controls ▪ Needs validation	(E. Elliott & Peadon, 2009) (Goh & Rosenbaum, n.d.)
4.5e	Alternate genetic conditions		Fragile X, velocardiofacial syndrome, down syndrome	(S J Astley, 2004)

			High risk	4-DDC
4.5f	Exposure to known teratogens		Dilantin, valproic acid High risk	(S J Astley, 2004) 4-DDC

4.6 Differential Diagnosis

	Disease/condition	Differentiating signs/symptoms	Differentiating tests	Papers
4.6a	Fetal hydantoin syndrome	Maternal history of phenytoin use during pregnancy Depressed nasal bridge and short nose with bowed upper lip	None – diagnosis based on history and exam	(E. Elliott & Peadon, 2009)
4.6b	Fetal valproate syndrome	Maternal history of valproate use during pregnancy High forehead, infraorbital crease or groove, small mouth and narrow bifrontal diameter	None – diagnosis based on history and exam	(E. Elliott & Peadon, 2009)
4.6c	Toluene embryopathy	Maternal history of toluene exposure during pregnancy Large anterior fontanelle, downturned corners of the mouth, hair patterning abnormalities, and ear abnormalities	None – diagnosis based on history and exam	(E. Elliott & Peadon, 2009)
4.6d	Wiliam syndrome	Wide mouth with full lips, stellate pattern of the iris, a loquacious (talkative) personality and musculoskeletal and cardiac problems	Deletion of one copy of the elastin gene in the 7q11.23 region of chromosome 7 seen on fluorescent in-situ hybridization Elevated serum calcium levels Supravalvular aortic stenosis, pulmonary stenosis or peripheral pulmonary stenosis on echocardiogram	(E. Elliott & Peadon, 2009)
4.6e	Brachmann-de Lange syndrome	A single bushy eyebrow, long eye lashes, downturned mouth, high arched palate and short limbs	Genetic analysis may show mutations in the NIPBL and SMC1A genes	(E. Elliott & Peadon, 2009)
4.6f	Maternal phenylketonuria	Small upturned nose, round face and a prominent	Elevated maternal phenylalanine levels	(E. Elliott & Peadon, 2009)

		glabella	Genetic analysis detects one of numerous mutations that have been found in the gene that encodes the phenylalanine hydroxylase on chromosome 12 in the region q22-24.1	
	Syndrome	**Differentiating features**	**Overlapping features**	**Papers**
4.6g	Aarskog syndrome	Rounded face, down-slant to palpebral fissures, widow's peak, crease below lower lip, incomplete out folding of upper helices, and dental eruption problems	Small nose with anteverted nares, broad philtrum, maxillary hypoplasia, wide spaced eyes	(National Center on Birth Defects and Developmental Disabilities, 2004)
	Williams syndrome	Wide mouth with full lips, stellate pattern of the iris, periorbital fullness, connective tissue disorders	Short palpebral fissures, anteverted nares, long philtrum, depressed nasal bridge, epicanthal folds	
	Noonan's syndrome	Down-slant to palpebral fissures, keratoconus, wide mouth, protruding upper lip	Low nasal bridge, wide-spaced eyes, epicanthal folds	
	Dubowitz syndrome	Shallow suraorbital ridge with nasal bridge near the level of the forehead, broad nasal tip	Short palpebral fissures, wide spaced eyes, epicanthal folds	
	Brachmann-DeLange syndrome	Single, bushy eyebrow extending across forehead, long eyelashes, downturned mouth, high arched palate, short limbs	Long philtrum, thin vermillion border, anteverted nares, depressed nasal bridge	
	Toluene embryopathy	Micrognathia, large anterior fontanel, down-turned mouth corners hair patterning abnormalities, bifrontal narrowing, ear abnormalities	Short palpebral fissures, midface hypoplasia, smooth philtrum, thin vermillion border	
		Short nose with bowed upper lip	Wide-spaced eyes and depressed nasal bridge	
	Fetal hydantoin syndrome (Fetal	High forehead, infraorbital crease or groove, small mouth	Epicanthal folds, anteverted nares, long philtrum	

	dilantin syndrome) Fetal valproate syndrome Maternal PKU fetal effects	 Small upturned nose, round facies, prominent glabella	with thin vermillion border, wide spaced eyes Epicanthal folds, short palpebral fissures, long underdeveloped philtrum, thin vermillion border	

FAS/PFAS	**ARND**	**ARBD**	**Not FASD**	Support and

5. Diagnostic Criteria/Guidelines

	Statement	**Papers**
5a	There are several problems with the IOM criteria for the diagnosis of FAS and alcohol-related effects. They are vague, with no specific parameters being set forth for diagnosis in each category. Neither the degree of growth deficiency nor the exact facial dysmorphic features required for each category are defined. In addition, the specific behavioural/cognitive phenotype is not characterised, and no guidelines for assessment of the complex pattern of behavioural or cognitive difficulties are suggested. Assessment of the family and genetic history of each affected child is not addressed adequately. Finally, ARBD and ARND are not practically defined in a clinical sense.	(Hoyme, et al., 2005)
5b	The 4-Digit Diagnostic Code appears to be extremely accurate in placing each child into a specific diagnostic category within the spectrum of alcohol-associated abnormalities, but the myriad of diagnostic categories is confusing and the system is impracticable for routine use in clinical practice. The Washington criteria also suffer from the same ambiguities as the IOM criteria. The family and genetic background of the child is not adequately integrated into the criteria. There is potential for over diagnosis of alcohol-related disabilities. However, this method does attempt to define objectively the facial phenotype of FAS.	(Hoyme, et al., 2005)
5c	An ideal classification system for FASD would allow accurate diagnoses of affected individuals by minimizing the false-positive and false-negative rates, precisely defining diagnostic categories, taking genetic and family histories it account, using a multidisciplinary approach, and creating straightforward, understandable, practical terms that could be applied easily in local clinical settings.	(Hoyme, et al., 2005)

Category	Statements	Papers
5.1 4-Digit Diagnostic Code These criteria were developed to ensure objectivity and reproducibility in the diagnosis of FAS through specifying cutoff points (e.g., for growth parameters and palpebral fissure length). The concept of the 4-digit diagnostic code was introduced to give greater diagnostic scope for describing children adversely affected by alcohol but who did not fulfill the diagnostic criteria for FAS. This system introduces the use of a number of other terms to describe clinical patterns, including the terms "static encephalopathy - alcohol exposed," and "neurobehavioral disorder - alcohol exposed." (E. Elliott & Peadon, 2009)		
FAS	Fetal Alcohol Syndrome is defined by evidence of growth deficiency, a specific set of subtle facial anomalies, and evidence of significant central nervous system damage/dysfunction that occur in individuals exposed to alcohol during gestation. FAS (diagnosis requires all 4 criteria): 1. Confirmed or unconfirmed maternal alcohol exposure. 2. Facial features - all 3 of: Philtrum rank 4 or 5 Upper lip rank 4 or 5 Palpebral fissure length <3rd percentile. 3. Growth retardation: prenatal or postnatal height or weight ≤10th percentile. 4. CNS - at least one of: Structural evidence of CNS damage (e.g., head circumference <3rd percentile, significant brain abnormalities on neuroimaging) Neurological evidence of CNS damage Significant impairment across 3 or more domains of brain function (generally ≤2 standard deviations). Domains include executive function, memory, cognition, social/adaptive skills, academic achievement, language, motor, attention, and activity level.	(S J Astley, 2004) 4-DDC (E. Elliott & Peadon, 2009)
pFAS	When a patient's characteristic features are very close to the classic features of FAS and the alcohol history strongly suggests that alcohol exposure during gestation was at high risk and likely to have played a role in the syndrome. Patients with Partial FAS either have the full set of facial anomalies found with FAS and evidence of CNS damage/dysfunction, but do not have growth deficiency; or they have growth deficiency and evidence of CNS damage/dysfunction, and most, but not all of the FAS facial features. Partial FAS (diagnosis requires 1 and 2 and 3):	(S J Astley, 2004) 4-DDC (E. Elliott & Peadon, 2009)

	1. Confirmed maternal alcohol exposure. 2. Facial features - at least 2 of: Philtrum rank 4 or 5 Upper lip rank 4 or 5 Palpebral fissure length <3rd percentile. 3. CNS - at least one of: Structural evidence of CNS damage (e.g., head circumference <3rd percentile, significant brain abnormalities on neuroimaging) Neurologic evidence of CNS damage Significant impairment across 3 or more domains of brain function (generally ≤2 standard deviations). Domains include executive function, memory, cognition, social/adaptive skills, academic achievement, language, motor, attention, and activity level.	
ARND or ARBD	ARND or ARBD: The 4-digit code uses different categories and terminology to describe children with neurodevelopmental problems, some of which may be comparable to alcohol-related neurodevelopmental disorder.	(E. Elliott & Peadon, 2009)
	The four digits of the diagnostic code reflect the magnitude of expression of the four key diagnostic features of FASD, in the following order: (1) growth deficiency, (2) FAS facial phenotype, (3) CNS abnormalities, and (4) prenatal alcohol exposure. There are 256 possible 4-digit diagnostic codes, ranging from 1111 to 4444. Each of the 4-digit diagnostic codes falls into one of 22 unique clinical diagnostic categories. Eight of the 22 diagnostic categories fall broadly under the designation of FASD.	(S J Astley, 2004) Cited: (BMA Board of Science, 2007)
5.2 IoM Diagnostic Criteria for FASD Developed by a panel of experts, based on review of a large number of children with clinical abnormalities who were born following confirmed alcohol exposure in utero. These criteria provided the first systematic approach to delineating diagnostic categories for children adversely affected by alcohol exposure in utero. The categories are as follows: FAS, partial FAS, alcohol-related neurodevelopmental disorder (ARND), and alcohol-related birth defects (ARBD). (E. Elliott & Peadon, 2009)		
FAS with confirmed maternal alcohol exposure	E. Confirmed maternal alcohol exposure F. Evidence of a characteristic pattern of facial anomalies that in clues features such as short palpebral fissures and abnormalities in the premaxillary zone (flat upper lip, flattened philtrum and flat midface) G. Evidence of growth retardation, as in at least one of the following: - low birth weight for gestational age - decelerating weight over time not due to nutrition - disproportional low weight to height	(Stratton, et al., 1996) IOM Cited: (BMA Board of Science, 2007)

	H. Evidence of CNS neurodevelopmental abnormalities, as in at least one of the following: - decreased cranial size at birth - structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia) - neurological hard or soft signs (as age appropriate), such as impaired fin motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination	
FAS without confirmed maternal alcohol exposure	B. Evidence of a characteristic pattern of facial anomalies that in clues features such as short palpebral fissures and abnormalities in the premaxillary zone. (flat upper lip, flattened philtrum and flat midface) C. Evidence of growth retardation, as in at least one of the following: - low birth weight for gestational age - decelerating weight over time not due to nutrition - disproportional low weight to height D. Evidence of CNS neurodevelopmental abnormalities, as in at least one of the following: - decreased cranial size at birth - structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia) - neurological hard or soft signs (as age appropriate), such as impaired fin motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination	(Stratton, et al., 1996) IOM Cited: (BMA Board of Science, 2007)
Partial FAS with confirmed maternal alcohol exposure	F. Confirmed maternal alcohol exposure G. Evidence of some components of the pattern of characteristic facial anomalies Either C or D or E C. Evidence of growth retardation, as in at least one of the following: - low birth weight for gestational age - decelerating weight over time not due to nutrition - disproportional low weight to height D. Evidence of CNS neurodevelopmental abnormalities, as in at least one of the following: - decreased cranial size at birth - structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia) - neurological hard or soft signs (as age appropriate), such as impaired fin motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination	(Stratton, et al., 1996) IOM Cited: (BMA Board of Science, 2007)

	E. Evidence of a complex pattern of behaviour or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone, such as learning difficulties; deficits in school performance; poor impulse control; problems in social perception; deficits in higher level receptive and expressive language; poor capacity for abstraction or metacognition; specific deficits in mathematical skills; or problems in memory, attention, or judgement	
Alcohol-related Birth Defects	History of maternal alcohol exposures and where clinical o animal research has linked maternal alcohol ingestion to an observed outcome. Cardiac: atrial septal defects; ventricular septal defects; aberrant great vessels; tetralogy of Fallot Skeletal: hypoplastic nails; shortened fifth digits; redioulnar synostosis; flexion contractures; camptodactyly; clinodactyly; pectus excavatum and carinatum; Klippel-Feil syndrome; hemivertebrae; scoliosis Renal: aplastic, dysplastic, hypoplastic kidneys, horseshoe kidneys, ureteral duplications, hydronephrosis Ocular: strabismus, refractive problems secondary to small globes, renal vascular anomalies Auditory: conductive hearing loss, neurosensory hearing loss Other: virtually every malformation has been described in some patient with FAS. The etiologic specificity of most of these anomalies to alcohol teratogensis remains uncertain	(Stratton, et al., 1996) IOM Cited: (BMA Board of Science, 2007)
Alcohol-related Neurodevelopmental Disorder	A. Evidence of CNS neurodevelopmental abnormalities, as in any one of the following: - decreased cranial size at birth - structural brain abnormalities (microcephaly, partial or complete agenesis of the corpus callosum, cerebellar hypoplasia) - neurological hard or soft signs (as age appropriate), such as impaired fin motor skills, neurosensory hearing loss, poor tandem gait, poor eye-hand coordination AND/OR B. Evidence of a complex pattern of behaviour or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone, such as learning difficulties; deficits in school performance; poor impulse control; problems in social perception; deficits in higher level receptive and expressive language; poor capacity for abstraction or metacognition; specific deficits in mathematical skills; or problems in memory, attention, or judgement	(Stratton, et al., 1996) IOM Cited: (BMA Board of Science, 2007)
5.3 Revised IoM Criteria for FASD Diagnosis The authors sought to clarify the criteria for diagnostic categories described in these guidelines to make them both more specific and more useful for clinicians. In particular, these criteria included specified cut-off points for measurements such as growth and palpebral fissure length. The criteria for ARBD were made stricter, requiring 2 or more of the facial features of FAS in addition to specified birth defects. (E. Elliott & Peadon, 2009)		
FAS with confirmed	I. FAS With Confirmed Maternal Alcohol Exposure (requires all features of A–D)	(Hoyme, et al., 2005)

maternal alcohol exposure	(A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide) (3) Smooth philtrum (score 4 or 5 with the lip/philtrum guide) (C) Evidence of prenatal and/or postnatal growth retardation (1) Height and/or weight p10%, corrected for racial norms, if possible (D) Evidence of deficient brain growth and/or abnormal morphogenesis, including 1 or more of the following: (1) Structural brain abnormalities (2) Head circumference p10%	Cited: (Manning & Eugene Hoyme, 2007) (BMA Board of Science, 2007) (E. Elliott & Peadon, 2009)
FAS without confirmed MAE	II. FAS Without Confirmed Maternal Alcohol Exposure	
PFAS with confirmed MAE	IB, IC, and ID as above III. Partial FAS With Confirmed Maternal Alcohol Exposure (requires all features, A-C) (A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide) (3) Smooth philtrum (score 4 or 5 with the lip/philtrum guide) (C) One of the following other characteristics: (1) Evidence of prenatal and/or postnatal growth retardation (a) Height and/or weight p10% corrected for racial norms, if possible (2) Evidence of deficient brain growth or abnormal morphogenesis, including 1 or more of the following: (a) Structural brain abnormalities (b) Head circumference p10% (3) Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background, or environment alone	

PFAS without confirmed MAE ARBD ARND	(a) This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behavior (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction) IV. Partial FAS Without confirmed Maternal Alcohol Exposure IIIB and IIIC, as above V. ARBD (requires all features, A-C) (A) Confirmed maternal alcohol exposure (B) Evidence of a characteristic pattern of minor facial anomalies, including 2 or more of the following: (1) Short palpebral fissures (p 10%) (2) Thin vermilion border of the upper lip (score 4 or 5 with the lip/philtrum guide) (3) Smooth philtrum (score 4 or 5 with the lip/ philtrum guide) (C) Congenital structural defects in 1 or more of the following categories, including malformation and dysplasias (if the patient displays minor anomalies only, X 2 must be present): cardiac: atrial septal defects, aberrant great vessels, ventricular septal defects, conotruncal heart defects; skeletal: radioulnar synostosis, vertebral segmentation defects, large joint contractures, scoliosis; renal: aplastic/hypoplastic/dysplastic kidneys, ‘‘horseshoe’’ kidneys/ureteral duplications; eyes: strabismus, ptosis, retinal vascular anomalies, optic nerve hypoplasia; ears: conductive hearing loss, neurosensory hearing loss; minor anomalies: hypoplastic nails, short fifth digits, clinodactyly of fifth fingers, pectus carinatum/excavatum, camptodactyly, ‘‘hockey stick’’ palmar creases, refractive errors, ‘‘railroad track’’ ears VI. ARND (requires both A and B) (A) Confirmed maternal alcohol exposure (B) At least 1 of the following: (1) Evidence of deficient brain growth or abnormal morphogenesis, including 1 or more of the following: (a) Structural brain abnormalities (b) Head circumference p10% (2) Evidence of a complex pattern of behavioral or cognitive abnormalities inconsistent with developmental level that cannot be explained by	

	genetic predisposition, family background, or environment alone (a) This pattern includes marked impairment in the performance of complex tasks (complex problem solving, planning, judgment, abstraction, metacognition, and arithmetic tasks); higher-level receptive and expressive language deficits; and disordered behaviour (difficulties in personal manner, emotional lability, motor dysfunction, poor academic performance, and deficient social interaction)	
	The authors proposed revision and clarification of the 1996 IOM criteria for diagnosis of FASD. Data from this large multiracial cohort of children prenatally exposed to alcohol indicate that this method can be applied easily in clinical practice, thus improving care for affected children and leading to improved precision of clinical and population-based research in FASD.	(Hoyme, et al., 2005)
	The Revised IOM Diagnostic Classification System (Hoyme et al., 2005) has an advantage over the Canadian system in that it has been tested in a large multiracial international cohort of children and found to be straightforward to use with reproducible results. In addition to stressing a multidisciplinary approach to evaluating alcohol exposed children and adults, this system also emphasizes the importance of considering the full differential diagnosis of genetic and teratogenic causes of developmental disabilities before a designation within the FASD spectrum is made.	(Manning & Eugene Hoyme, 2007)
5.4 CDC Diagnostic Criteria for FAS A committee of experts, mandated by US federal law, was convened by the CDC to update and refine the diagnostic criteria for FAS. Criteria were only developed for FAS because there was deemed to be lack of evidence to support the development of reliable diagnostic criteria for the rest of the spectrum. (E. Elliott & Peadon, 2009)		
FAS	Facial dysmorphia – based on racial norms, individual exhibits all three characteristic facial features - Smooth philtrum – measured as 4 or 5 on Lip-Philtrum Guide - Thin vermillion border – measured as 4 or 5 on Lip-Philtrum Guide - Small palpebral fissures – measured as <10th percentile Growth problems – confirmed prenatal or postnatal height or weight, or both, at or below the 10th percentile, documented at any one point in time (adjusted for age, sex, gestational age, and race or ethnicity). CNS abnormalities - Structural 5) Head circumference (OFC) at or below the 10th percentile adjusted fro age and sex. 6) Clinically significant brain abnormalities observable through imaging. - Neurological Neurological problems not due to a postnatal insult or fever, or other soft neurological signs outside normal limits. - Functional Performance substantially below that expected for an individual's age, schooling, or circumstances, as evidenced by:	(National Center on Birth Defects and Developmental Disabilities, 2004) Cited: (BMA Board of Science, 2007) (E. Elliott & Peadon, 2009)

	5) Global cognitive or intellectual deficits representing multiple domains of deficit (or significant developmental delay in younger children) with performance below the 3rd percentile (2 standard deviations below the mean for standardised testing). OR, 6) Functional deficits below the 16th percentile (1 standard deviation below the mean for standardised testing) in at least three of the following domains: a) cognitive or developmental deficits or discrepancies b) executive functioning deficits c) motor functioning delays d) problems with attention hyperactivity e) social skills f) other, such as sensory problems, pragmatic language problems, memory deficits, etc. Maternal Alcohol Exposure 3) Confirmed prenatal alcohol exposure 4) Unknown prenatal alcohol exposure Criteria for diagnosis requires all three of the following: - Documentation of all three facial abnormalities (smooth philtrum, thin vermillion border, small palpebral fissures) - Documentation of growth deficits - Documentation of CNS abnormality	
5.5 Canadian FASD Guidelines The Canadian guidelines include elements of both the IOM criteria and the 4-digit diagnostic code and provide specific cutoff values for growth parameters. The criteria for CNS involvement are more stringent that other classifications, requiring evidence of involvement of 3 or more CNS domains. (E. Elliott & Peadon, 2009)		
FAS	FAS (diagnosis requires all 4 criteria): 1. Confirmed or unconfirmed maternal alcohol exposure 2. Facial features - all 3 of: Philtrum rank 4 or 5 Upper lip rank 4 or 5 Palpebral fissure length ≤3rd percentile. 3. Growth retardation - at least one of: Birth weight or birth length ≤10th percentile for gestational age	(Chudley, et al., 2005) Cited: (BMA Board of Science, 2007) (E. Elliott & Peadon, 2009)

	Height or weight ≤10th percentile Disproportionately low weight-to-height ratio (≤10th percentile). 4. CNS - evidence of impairment in 3 or more of the following CNS domains: Hard or soft neurologic signs Brain structure Cognition Communication Academic achievement Memory Executive functioning and abstract reasoning Attention deficit/hyperactivity Adaptive behavior Social skills Social communication.	
PFAS	Partial FAS (diagnosis requires all 3 criteria): 1. Confirmed maternal fetal alcohol exposure. 2. Facial features - 2 or more of: Philtrum rank 4 or 5 Upper lip rank 4 or 5 Palpebral fissure length <3rd percentile. 3. CNS - evidence of impairment in 3 or more of the following CNS domains: Hard or soft neurologic signs Brain structure Cognition Communication Academic achievement Memory Executive functioning and abstract reasoning Attention deficit/hyperactivity	

ARND	Adaptive behavior Social skills Social communication. ARND (diagnosis requires 1 and 2): 1. Confirmed maternal alcohol exposure. 2. CNS - evidence of impairment in 3 or more of the following CNS domains: Hard or soft neurologic signs Brain structure Cognition Communication Academic achievement Memory Executive functioning and abstract reasoning Attention deficit/hyperactivity Adaptive behavior Social skills Social communication.	
5.6 Age-related diagnostic criteria for FAS and/or atypical FAS		
Infants	History of prenatal alcohol exposure Facial abnormalities Growth retardation – height, weight, head circumference Hypotonia, increased irritability Jitteriness, tremulousness, weak such Difficulty 'habituating', getting used to stimulation	(First Nations and Inuit Health Committee, 2002)
Preschool	History of alcohol exposure, growth retardation, facial abnormalities Friendly, talkative and alert Temper tantrums and difficulty making transitions Hyperactive; may be over sensitive to touch or over-stimulation Apparent skill levels may appear to be higher than their tested levels of ability	(First Nations and Inuit Health Committee, 2002)

	Attention deficits, developmental delays – speech, fine motor difficulties	
Middle childhood	History of alcohol exposure, growth retardation, facial abnormalities Hyperactivity, attention deficit, impulsiveness Poor abstract thinking Inability to foresee consequences of actions Lack of organisational skills Inappropriate behaviour: overly affectionate – does not discriminate between family and strangers; lack of inhibitions; communication problems – lack of social skills to make and keep friends, unresponsive to social clues, uses behaviour as communication; Difficulty making transitions Academic problems – reading and mathematics Behaviour problems – 'stretched toddler'	(First Nations and Inuit Health Committee, 2002)
Adolescent and adult	History of alcohol exposure, growth retardation, facial abnormalities Intelligence Quotient – average to mildly retarded with wide range; continued school difficulties Difficulty with adaptive and living skills Attention deficits, poor judgement, impulsivity lead to problems with employment, stable living and the law Serious life adjustment problems – depression, alcoholism, crime, pregnancy and suicide	(First Nations and Inuit Health Committee, 2002)
5.7 FAS SCREEN		
	- Designed for use in community settings (public schools, preschool programs) and clinical settings by both paraprofessional and medically trained personnel. - Screening for 4-18 year olds - Rapid test – 15 minutes or less Sensitivity (How good is this test at picking up people who have the condition?) = 100% Specificity (How good is this test at correctly excluding people without the condition?) = 94.1% Positive predictive value (If a person tests positive, what is the probability they have the condition?) = 9.1% Negative predictive value (If a person tests negative, what is the probability that they do not have the condition?) = 100% The tool is rapid; has adequate performance characteristics, and the test is cost effective. DOB, age, gender, race Height, weight, head circumference Ears stick out (Protruding auricles)- 4	(Burd et al., 1999)

Growth	Skin folds near inner eye (Epicanthal folds)-5	
Head& face	Drooping of eyelids (Ptosis)-4	
	Cross-eyes, one or both eyes (Strabismus)-3	
	Flat midface/cheeks (Hypoplastic maxilla)-7	
	Flat/low nose between eyes (Low nasal bridge)-2	
	Upturned nose-5	
	Groove between lip & nose absent or shallow (Flat philtrum)-5	
	Thin upper lip-4	
	Cleft lip or cleft of roof of mouth (present or repaired)-4	
	Short, broad neck-4	
	Curvature of the spine (Scoliosis)-1	
Neck & back	Spina bifida (history of neural tube defect)-4	
	Fingers, elbows (limited joint mobility)-4	
	Permanently curved, small fingers, especially pinkies (Clinomicrodactyly)-1	
Arms & hands	Deep or accentuated palmar creases-4	
	Small nails/nail beds (Hypoplastic nails)-1	
	Tremulous, poor finger agility (fine motor dsyfunction)-1	
	Sunken chest (Pectus Excavatum)-3	
	Chest sticks out (Pectus Carinatum) optional-1	
Chest	History of heart murmur or any heart defect-4	
	Raised red birthmarks (Capillary Hemangiomas)-4	
	Greater than normal body hair, hair also on forehead an back (Hirsutism)-1	
Skin	Mild to moderate mental retardation (IQ < 70)-10	
	Speech and language delays-2	
Development	Hearing problems-1	
	Vision problems-1	
	Attention concentration problems-2	
	Hyperactivity-5	
	(referral if score is 20 or above)	
	Reported sensitivity 100%, specificity 94-95%, Positive predictive value 92%, accuracy (?) 94-95%	(Goh & Rosenbaum,

		n.d.)
	FAS Screen: a 32 item screening test, a rapid screening tool for community-based screening of FAS. The goal is to screen out low-risk children and identify a high risk population. The FAS Screen in a community setting typically screens out as low risk about 94–96% of children The sensitivity in the norming sample was 100%, the specificity was 94%, the positive predictive value was 92%, and the accuracy was 94%.	(Poitra et al., 2003)
5.8 FAS		
History & Exam	Key Factors: presence of risk factors; gestation <37 weeks; height, weight, head circumference <10th percentile; characteristic facial dysmorphology; presence of birth defects; developmental delay and behavioural problems; mental health problems; and sibling with similar symptoms. Other Factors: hearing or vision impairment; poor feeding; and irritability	(E. Elliott & Peadon, 2009)
Diagnostic Tests	1st tests to order: facial photographic assessment Other tests to consider: ECG; echocardiogram; EEG; MRI/CT head; renal ultrasonography; skeletal x-ray Emerging tests: functional MRI; magnetic resonance spectroscopy	(E. Elliott & Peadon, 2009)
Age	Developmental history should be elicited if child is being examined later in life: Infants: poor feeding, growth retardation, irritability, or developmental delay, including delayed motor milestones or delayed speech and language development. Children: growth retardation, or problems with language, speech, hearing, vision, learning, or behaviour Adolescents: drug and alcohol abuse, poor educational performance, poor social skills, or contact with the law or incarceration.	(E. Elliott & Peadon, 2009)
5.9 Diagnostic Criteria Checklist Used with ARND Sample		
ARND (deficits)	Decreased intelligence; poor math; poor reading comprehension; chattiness; anomia; poor comprehension; problems with word meanings; difficulty with sentence structure; problem with pragmatics; preservative; poor gross and fine motor; poor time management/planning; poor organisation/planning; poor memory; poor associative learning; concrete thinkers; poor social skills; behaviour problems; poor attention/ADHD; high activity; poor adaptive skills	(Greenbaum, 2000)
ARND (assets)	Relatively good visuospatial skills; good face recognition; air of competence/self-confidence; good rote memory; good verbal fluency; good immediate object memory	(Greenbaum, 2000)
5.10 Youth Probation Officers' Guide to FASD Screening and Referral The FASD Screening Tool and Referral Form for Youth Probation Officers was developed to be used as part of a referral process for an FASD diagnostic assessment in the Youth Justice FASD Program at the Asante Centre. The rating scores are not on a continuous scale with cut-off points representing a greater or lesser probability of the youth Having		

FASD. It is a screening and referral from for a more formal assessment. (Conry & Asante, 2010)		
	A) Social Factors are those that may identify a youth at-risk for FASD. That is, these factors may increase the probability that the youth could have FASD: - Youth is adopted - Youth currently, or previously, was in foster care or involved with child protection services - Youth has a sibling with a documented diagnosis of FASD - There is documentation that the youth is suspected of having FASD - Youth's mother has known history of alcoholism or prenatal alcohol use B) Personal Factors are those that have been associated with (but not necessarily unique to) FASD. - Developmental delay in early childhood (speech/language therapy, occupational therapy, infant development or child development services prior to school entry) - Learning difficulties (learning assistance, modified program or experienced school failure or drop-out) - Growth deficiency (appears short compared to peers, or of a low weight for age) - Diagnosis of ADHD - Mental health diagnosis (anxiety, depression, Oppositional Defiant Disorder, Conduct Disorder) The youth should be referred for assessment if he/she had - 1 social factor PLUS at least 2 personal factors, OR - No social factors PLUS at least 3 personal factors - Where there is a probability that a client's problems may be related to prenatal alcohol exposure, the officer should endeavour to gather information from the client's past medical records and other sources (birth mother, physician's/midwife's prenatal and birth records, maternal grandparents/aunts, social workers' records, father's or mother's partners). - Personal factors can be obtained from family members, social workers, previous reports and school records. Behaviour Checklist – to characterise the youth: - attention seeking, demanding, loud - misuse of alcohol and other drugs - easily manipulated and led by others - has a high need for acceptance - poor understanding of personal boundaries - chronically misses appointments - disinhibited about sharing personal information	(Conry & Asante, 2010)

	- has trouble following rules or requirements - poor decision maker, poor problem solver, lacks insight - does not understand effects of his/her actions on others - requires supervision and management of time and money - impulsive - anger control problem - socially inept/immature - concrete and literal thinker	

6. Other Statements

	Statements	**Papers**
6a	Funding for development, training and maintenance of multidisciplinary diagnostic teams is necessary so that major centres will have the expertise and capacity to serve their communities.	(Chudley, et al., 2005)
6b	All healthcare professionals as part of routine clinical care should provide ongoing advice and support to expectant mothers at every stage of pregnancy and this should include the risks of maternal alcohol consumption	(BMA Board of Science, 2007) Guide for health professionals
6c	All healthcare professionals involved in the provision of antenatal care should ensure that alcohol use among pregnant women is monitored and recorded appropriately	(BMA Board of Science, 2007) Guide for health professionals
6d	Six paediatricians conferred and detailed the physician role, goals for the medical examination and specific tasks and tools recommended for the achievement of goals. They outlined the tasks and tools as: - History analysis - Current function of the child and how this has changed over time is obtained by past and present documentation from school and caregivers. The tool could be the Caregiver Interview from the DPN Manual - Health determinants that impact development and function - Physical exam - Mental status - Formulation of diagnosis - Development of intervention strategies and support systems after diagnosis. - Longitudinal follow up	(Canada Northwest FASD Research Network, 2007b)2

Appendix 6 Publications Excluded from Systematic Review (Full Literature review)

Alcohol Healthwatch (2010). *Towards multidisciplinary diagnostic services for fetal alcohol spectrum disorder*. Auckland: Alcohol Healthwatch. Reason for exclusion: small sample size

Aragon, A. S., Kalberg, W. O., Buckley, D., Barela-Scott, L. M., Tabachnick, B. G., & May, P. A. (2008). Neuropsychological study of FASD in a sample of American Indian children: Processing simple versus complex information. *Alcoholism: Clinical and Experimental Research, 32 (12)*, 2136-2148. Reason for exclusion: small sample size, further validation required

Astley, S. J. (2010). Profile of the first 1,400 patients receiving diagnostic evaluations for fetal alcohol spectrum disorder at the Washington State Fetal Alcohol Syndrome Diagnostic & Prevention Network. *Canadian Journal of Clinical Pharmacology, 17*(1), e132-164. Reason for exclusion: Not postnatal screening or diagnostic instrument or method

Astley, S. J., Aylward, E. H., Olson, H. C., Kerns, K., Brooks, A., Coggins, T. E., et al. (2009). Magnetic resonance imaging outcomes from a comprehensive magnetic resonance study of children with fetal alcohol spectrum disorders. *Alcoholism: Clinical & Experimental Research, 33*(10), 1671-1689. Reason for exclusion: further validation required

Autti-Ramo, I., Autti, T., Korkman, M., Kettunen, S., Salonen, O., & Valanne, L. (2002). MRI findings in children with school problems who had been exposed prenatally to alcohol. *Developmental Medicine and Child Neurology, 44 (2)*, 98-106. Reason for exclusion: limited methodology, further validation required

Autti-Ramo, I., Fagerlund, A., Ervalahti, N., Loimu, L., Korkman, M., & Hoyme, H. E. (2006). Fetal alcohol spectrum disorders in Finland: Clinical delineation of 77 older children and adolescents. *American Journal of Medical Genetics, 140 A (2)*, 137-143. Reason for exclusion: Not postnatal screening or diagnostic instrument or method

Bearer, C. F., Jacobson, J. L., Jacobson, S. W., Barr, D., Croxford, J., Molteno, C. D., et al. (2003). Validation of a new biomarker of fetal exposure to alcohol. *The Journal of Pediatrics, 143*(4), 463-469. Reason for exclusion: small sample size, further validation require

Benoit, T., Bowes, C., Bowman, N., Cantin, D., Chudley, A., Crolly, D., et al. (2002). Telemedicine diagnosis for fetal alcohol syndrome - The Manitoba experience. *Paediatrics and Child Health, 7 (3)*, 147-151. Reason for exclusion: Not postnatal screening or diagnostic instrument or method

Bookstein, F. L., Sampson, P. D., Connor, P. D., & Streissguth, A. P. (2002). Midline corpus callosum is a neuroanatomical focus of fetal alcohol damage. *Anatomical Record, 269*(3), 162-174. Reason for exclusion: only for use in adult population (>26 years), further validation required

Burd, L., Klug, M. G., Martsolf, J. T., & Kerbeshian, J. (2003). Fetal alcohol syndrome: Neuropsychiatric phenomics. *Neurotoxicology and Teratology, 25*(6), 697-705. Reason for exclusion: observational, comparative and descriptive, further validation required

Burd, L., Martsolf, J. T., Klug, M. G., & Kerbeshian, J. (2003). Diagnosis of FAS: A comparison of the Fetal Alcohol Syndrome Diagnostic Checklist and the Institute of Medicine Criteria for fetal alcohol syndrome. *Neurotoxicology and Teratology, 25 (6)*, 719-724. Reason for exclusion: Poor agreement between diagnostic codes employed

Cappiello, M. M., & Gahagan, S. (2009). Early child development and developmental delay in indigenous communities. *Pediatric Clinics of North America, 56*(6), 1501-1517. Reason for exclusion: Not postnatal screening or diagnostic instrument or method

Caprara, D. L., Nash, K., Greenbaum, R., Rovet, J., & Koren, G. (2007). Novel approaches to the diagnosis of fetal alcohol spectrum disorder. *Neuroscience and Biobehavioral Reviews, 31*(2), 254-260. Reason for exclusion: Further validation required

Carr, J. L., Agnihotri, S., & Keightley, M. (2010). Sensory processing and adaptive behavior deficits of children across the fetal alcohol spectrum disorder continuum. *Alcoholism: Clinical and Experimental Research, 34 (6)*, 1022-1032. Reason for exclusion: Not postnatal screening or diagnostic instrument or method

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APPENDIX E: COMMUNITY CONVERSATIONS REPORT (FULL VERSION)

ACKNOWLEDGEMENTS

The Australian Collaboration would like to thank the women who participated in the alcohol and pregnancy community conversations in Perth and Cairns. The willingness of these women to provide realistic, truthful and insightful responses has made a significant contribution to the Fetal Alcohol Spectrum Disorders (FASD) Project and future planning for prevention, education and advocacy in the fetal alcohol spectrum disorders field.

Thanks are also extended to the presenters and table facilitators who were an integral part of the community conversations.

CONSUMER AND COMMUNITY PARTICIPATION

Since 2005, the National Health and Medical Research Council (NHMRC) has required applicants for research grants to give details of their plans to involve consumers and community members. The Telethon Institute for Child Health Research (the Institute) and many other academic institutions also recognise the involvement and active engagement of consumers and community in health and medical research. In 2002 the Institute implemented a consumer and community participation engagement program to increase and enhance opportunities for greater participation in all research projects.

BENEFITS OF COMMUNITY AND CONSUMER PARTICIPATION IN RESEARCH PROJECTS

The establishment of effective partnerships between consumers, the community and researchers will:

- add value to evidence based research;
- facilitate and enhance understanding of consumer and community priorities, perspectives and issues;
- enhance the planning, conduct and analysis of research programs;
- enhance the dissemination of research findings and increase translation into policy and practice; and

- provide increased opportunities to obtain funding for research which addresses the needs and priorities of the community.

COMMUNITY PARTICIPATION PROCESS FOR THE FASD PROJECT

The Australian Collaboration includes four consumer and community representatives:

- Ms Anne McKenzie, Consumer Advocate at the Telethon Institute for Child Health Research and the University of Western Australia
- Ms Sue Miers AM, Spokesperson for The National Organisation for Fetal Alcohol Syndrome and Related Disorders Inc (NOFASARD)
- Ms Elizabeth Russell, The National Organisation for Fetal Alcohol Syndrome and Related Disorders Inc (NOFASARD) and Russell Family Fetal Alcohol Disorders Association (rffada)
- Ms Maureen Carter, Chief Executive Officer Nindilingarri Cultural Health services

The FASD Project will use a Delphi process to reach a consensus on what should be included in the screening and diagnostic instrument. The Delphi process requires a systematic review of literature in the field of FASD followed by the development of a series of statements and questions regarding the screening and diagnosis of FASD. Using a Likert scale (ratings of 1 – 5 from strongly agree to strongly disagree), participants are asked to indicate their agreement with the statements via an on-line questionnaire (Delphi instrument). While the project plan stated that it would include health professionals and consumers, the statements were in reality targeted at health professionals. The consumer members of the Steering Group were of the view that the process would exclude adequate consumer and community engagement and gave consideration to how consumers and the community could be actively engaged in this project outside the formal Delphi process. It was proposed that consumer and community participation follow a format used in four successful 'community conversations' (world café process) held at the Institute in 2009 and 2010 and facilitated by Ms Anne McKenzie.

WORLD CAFÉ PROCESS

BRIEF DESCRIPTION

The world café is a method which makes use of an informal café style for participants to explore an issue by holding discussions in small table groups. These are held in multiple rounds of 20-30 minutes and conclude with a summary of the discussions.

Participants discuss the issue at hand around their table and at regular intervals they move to a new table. The table facilitator remains and summarises the previous conversation to the newly arrived participants. By moving participants around the room the conversations at each table are cross-fertilised with ideas from other tables. At the end of the process the main ideas are summarised and follow-up possibilities are discussed.

The world café has been used in many different settings. It is good at generating ideas, sharing knowledge, stimulating innovative thinking, and exploring action in real life situations. The informal but deep conversations that the world café encourages can lead to improved relationships between participants and between wider groups.

COMMUNITY CONVERSATION PURPOSE

The purpose of the alcohol and pregnancy community conversation was to provide input to the Steering Group for use when developing the screening and diagnostic instrument. The community voice aimed to engender information from consumers and the community who are not as well informed about FASD as the Steering Group consumer representatives and in a manner that would elicit the most truthful information from women. It was particularly important when the Steering Group was considering questions that maybe asked of women as part of the screening tool. The community conversation was an important foundation for meaningful and inclusive consumer and community participation.

COMMUNITY CONVERSATION FUNDING

Funding for the community conversations was allocated from the FASD Project budget. An honorarium of $30.00 was paid to participants to cover parking, transport and meet any other out of pocket expenses. The logistics costs (venue hire for Cairns, catering, printing etc) were also met though the FASD Project budget.

COMMUNITY CONVERSATION LOCATIONS

In making the decision on potential locations for the community conversations, members were cognisant that they should offer the opportunity to women in both rural and urban communities and from a range of backgrounds. Cairns was selected as the more remote location as several members of the Steering Group were located in this region. Perth was selected as the urban location as the

Project Team is based at the Institute and there is an established network of consumer and community organisations.

COMMUNITY CONVERSATION INVITATIONS

Invitations to attend the Perth (7 December 2010) and Cairns (18 February 2011) community conversations were placed on the FASD Project website and circulated via numerous email distribution lists that were provided by members of the Steering Group. Personal contact was made with key people in consumer organisations to discuss the seminars and to seek their support in forwarding the invitations through their networks. A copy of the invitations can be found in Appendix 1.

The process worked well in Perth where there is an established network of consumer organisations and reference groups. In Cairns it was more difficult to reach the target audience. Although it was distributed broadly it was challenging to garner wide spread interest. This was also exacerbated by the cyclone which hit north Queensland in the weeks before the community conversation was held.

COMMUNITY CONVERSATIONS PERTH AND CAIRNS

FACILITATOR

Since 2004, **Anne McKenzie** has worked as the Consumer Advocate at the University of Western Australia's School of Population Health and the Telethon Institute for Child Health Research. The key task of this position is to increase consumer and community participation in health and medical research.

Anne is also involved in the following community areas as:

- Chairperson of the Health Consumers' Council WA and senior consumer representative on several state health committees;
- a senior consumer representative for the Consumers' Health Forum of Australia on the National Prescribing Service New Drugs Working Group and Medicines Australia Code of Conduct Committee;
- a member of the Cochrane Consumer Network, the consumer arm of the Cochrane Collaboration; and
- the lay member on the Silver Chain Nursing Association Human Research Ethics Committee.

TABLE FACILITATORS

Perth

Ms Laura Bond – Research Officer, FASD Project

Ms Lyn Colvin – Research Officer, Telethon Institute for Child Health Research (author published papers on FASD)

Ms Kathryn France – NHMRC Postgraduate Scholar & Project Manager, Centre for Applied Social Marketing Research Faculty of B&L, Edith Cowan University (author published papers on FASD)

Ms Jan Payne – Senior Research Officer, Telethon Institute for Child Health Research (author published papers on FASD)

Cairns

Ms Louise Orbons – has a number of businesses in Cairns and is currently doing a degree in Social Work

Mrs Sue Turner – National Quality and Compliance Manager, ITEC Employment

The role of the table facilitators was to:

- encourage people to put forward their ideas/comments
- guide – encourage and affirm each person
- stay neutral and don't express your views
- read the background information, ask the question
- keep discussion focussed, keep discussion moving and ensure that everyone has a chance to participate
- not let one person dominate the conversation

Table facilitators were advised that during the conversation they should not suggest that women may have particular feelings (eg guilt, anxiety). If these feelings were expressed by a participant/s it was the facilitator's role to engage with the group and not to isolate one person or make them feel that they are the only person to have those feelings.

The table facilitators stayed at the same table and the groups moved around the room discussing a different question at each table. At the conclusion of the small group discussion sessions the table facilitators were responsible for collating the participant statements and preparing a brief report back to the whole group.

PRESENTERS

Clinical Professor Carol Bower is one of the Institute's founding senior researchers. Professor Bower has been a driving force behind its epidemiological research program, in particular in the establishment of the WA Birth Defects Registry. In the 1990's, Professor Bower was part of the international team that showed the link between folate intake during pregnancy and the reduction in neural tube defects and in 2007 was awarded a Leadership Award from the Flour Fortification Initiative for her folate advocacy role. In addition to folate, Professor Bower is also leading research projects into other factors that can influence health outcomes of newborn babies including alcohol consumption, prescription medication and in-vitro fertility treatment.

Heather Jones is the FASD Project Manager based at the Institute.

These presenters provided an overview of recent and current alcohol and pregnancy research and information on fetal alcohol spectrum disorders.

Sue Miers AM is the spokesperson for the National Organisation for Fetal Alcohol Syndrome and Related Disorders (NOFASARD). Sue is a long term foster parent (over 20 years) to a 25 year old daughter with a partial fetal alcohol spectrum disorder. Sue has lobbied extensively on both a national and state level to raise awareness of FASD. She believes all people should have access to correct information, advice and guidance to make informed choices about the effects of alcohol during pregnancy, understanding the detrimental impact on children and adults. She is a recognised parent authority in this area and has presented at numerous workshops and conferences.

Elizabeth Russell represents both NOFASARD and the Russell Family Fetal Alcohol Disorders Association (rffada). Elizabeth is a recovering alcoholic who in 2001 found that her addiction had physically harmed her two sons. Her eldest son Mick, who is 29 years old, was diagnosed with Neuro-developmental Disorder-Alcohol Exposed and her youngest son Seth who is 25 years of age has full Fetal Alcohol Syndrome. She is now in recovery and with her recovery comes the determination to find a way, through love and understanding to assist her sons; but even more to awaken Australians to the dangers of alcohol and pregnancy presently unknown to the vast majority of people in Australia. Elizabeth believes that those who have the ability to take action have the responsibility to take action and she feels that the birth mother's perspective is one which is least heard when it comes to this condition. To this end she has written from this unique perspective three books on Fetal Alcohol Syndrome: *'Alcohol and Pregnancy – No Shame, No Blame'* , *'Alcohol*

and Pregnancy A Mother's Responsible Disturbance' and the first book in the world on employment strategies for people with FASD called *'Strategies for Employment Services Specialists'*

These presentations based on personal insight and experience provided compelling stories of raising children with a fetal alcohol spectrum disorder.

AGENDA

The agenda was designed to allocate the majority of time to the small group discussions and whole group discussion, keeping presentations to a minimum. A copy of the Perth and Cairns community conversation agendas can be found in Appendix 2.

QUESTIONS USED IN THE WORLD CAFÉ PROCESS

The questions for the Perth community conversation were developed in consultation with the facilitator, two of the table facilitators, presenters and two health professional members of the FASD Collaboration. The questions were designed to:

- elicit responses on what information women expected their health professional to provide with respect to alcohol use in pregnancy;
- discuss the manner in which the health professional interacts with the woman; and
- elicit responses on the level of information a woman would provide to the health professional about their alcohol use in pregnancy.

Following a review of the evaluation forms from the Perth community conversation the questions were revised for the Cairns community conversation as some questions appeared to be repetitive. The Perth and Cairns community conversation questions can be found in Appendix 3.

STATEMENTS FROM COMMUNITY CONVERSATION PARTICIPANTS

Using the world café process participants were asked to write individual statements on sticky notes and place them onto the group sheet. 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
- general

- health professionals
- health professional training
- how to ask
- information to the public
- information to women
- language
- resources
- schools
- timing

Overwhelmingly women wanted health professionals to provide them with evidence based information that no alcohol during pregnancy is the safest option. Women wanted to know how alcohol could affect the fetus. Women were in agreement that any questions asked by a health professional with respect to alcohol consumption during pregnancy should form part of a series of questions asked of all women, that is, questions about general health, smoking, diet, lifestyle etc.

There was also consensus for information to be provided to partners and families to help them understand the issues and provide support for the women.

There was unanimous support for a national campaign to raise awareness of the risks of drinking alcohol during pregnancy. Ideally the campaign would extend across all media – television, radio and social media such as YouTube and Twitter. It was important that the campaign include coasters and posters in hotels, bars, restaurants and liquor outlets, including placement behind toilet doors. There was also strong support for the inclusion of information on alcohol and pregnancy in high school health curricula.

Table 1: Summary of the statements from participants sorted by themes (Perth and Cairns)

Theme	Summary of Participant Statements
Counselling Support	• Support options – help lines, counselling, put a plan in place – where to from here, who's to follow up , culturally appropriate environment • Health professionals need to know where to refer and how to refer • Health professionals should ask the questions but at the same time offer solutions. Hard to offer up information if you are feeling there's nothing being given in return ie this information will help people in the future but it won't necessarily help your situation • Need support not guilt – need to be mindful of mental health issues • Word of mouth – referral from community (cultural) group and someone has to be responsible for asking the hard questions • 2 scenarios – walk in self-referred is an open conversation and health professional diagnosis
Family Community	• Information about alcohol use in pregnancy available to the whole community, not just the mother. Family members need to understand the issues (including men) • Family and community support important. It is hard when communities/friends are all into it – bored, nothing to do and if you don't drink, not part of the group, not fun • Provide with information to share with partner and families (support no drinking) on FASD to help support other women/men who might be thinking about having a baby. Ask questions in private – not in front of partners or family • This is about the child & their difficulties, not about their culture • History and relationships develop in small communities
Feelings	• Defensive • Sensitive • Ashamed and possibly lie about my alcohol consumption • Denial – can depend if parent has accepted diagnosis • A feeling of guilt or doing something wrong • Anxious • Concerned • Scared • Hostility • Felt offended, didn't even drink coffee, let alone drugs/alcohol • Wondering why they want to know • Confronted • Felt stereotyped by race/ethnicity • Fearful of staff • Happy and appreciative they pre-empted the issue • Is there something wrong with my baby?
General	• FASD is not curable – it's for life • About child not culture • Rename FASD as just points the finger at the mother and labels the child. Should represent the symptoms not the cause • We should all want the same thing – to give birth to a healthy baby not burdened by a preventable disease • Do not make class assumptions about alcohol use

	• Mandatory reporting will impact on what information is provided – scared that it would be reported to Department of Child Protection or police • Intergenerational trauma, mother may have FASD, refugees, stolen generation • Informed consent – parents/guardians should be asked if they want to proceed with screening • Instrument must be appropriate for all Australian children and at different ages • If tool developed need to provide a guide to referral pathways – ensure seen by appropriate health professionals • Website register of resources, networking opportunities and hear what is being done and what works well in Australia • Australian conference – networking opportunities and hear what is being done and what works well in Australia • FASD on agenda at community events and medical conferences • Should establish a register of children with a FASD
Health professionals	• Perception is doctors know best – they don't need anything to justify and often information is more of a lecture. Body language is easy to read. • Important for health professionals to build relationships. Women preferred the information coming from a child health nurse, midwife or from a female doctor. They take more time and seem more caring. Building trust – shame factors associated with how much you drink. • Research and women's feedback is that health professionals are not providing information to women or they are giving mixed messages about alcohol use in pregnancy • GPs are used to seeing one person with one complaint not a range of issues related to women • Health professionals should be able to give a woman information when planning a pregnancy – hope they have the skill/knowledge to help you • Women might see many different health professionals – no-one twice therefore difficult to build up any rapport. • Health professionals should ask a woman what she knows about alcohol & pregnancy – go from there and ask if she would like to talk about this or would like to take some information away to read. Explain why these questions are being asked and that you are not being singled out – asked of all pregnant women • If you can't give up, cut down – provide options for changing behaviour. Women know that many health professionals drink • Prefer to hear about a child's learning difficulties from a health professional than from a teacher, childcare, playgroup
Health professional training	• Health professionals need to be trained to ask, need to be comfortable asking the questions and need to have follow up options • Training should commence at university – workforce development re standard drinks/other issues such as the risk factors and what disability would look like • Health professionals to have cultural sensitivity training – indigenous, African, Indian, Malay etc • Most health professionals do not have the skills to break it down for indigenous people • Education and training not just for paediatricians – rural areas will not see one, it will probably be the aboriginal health worker or child health nurse • Others trained to assist and knowledge to refer – school nurses, child care workers, teachers

How to ask	• Not a lecture • From the woman's perspective what are the benefits in answering these questions in detail and honestly – be friendly and explain why the questions are being asked in the context of diet and lifestyle. This will vary depending on whether asked as a general question during pregnancy or at birth, or if screening for learning difficulties or developmental delays. How will this help my child? Explain that delayed development is not only alcohol related and there could be a number of other causes • Make the question about alcohol use part of a standard set of questions that are asked of all pregnant women • Questions should be simple, clear and easy to understand for all races/classes within society and not like an interrogation • Cultural awareness – nodding head does not always mean 'yes', I agree • Just ask the question, there's no single way of asking that will please everyone – are you currently drinking any alcohol. If yes how much on any day or week • Put equal emphasis on alcohol as other substances such as tobacco or drugs • Explain how alcohol actually affects the baby – how alcohol crosses the placenta, everything the mother drinks reaches the baby and the baby will be drunk with her • Describe the consequences of alcohol to the baby – what can happen to their brain and development • Ask if the woman has any questions or would like to know more – provide information or where she can access it • Don't use forms for a woman to complete – can be difficult for everyone to understand what is required, easier to lie, no follow up • Health professional should be aware of , and be prepared to deal with feelings of defensiveness, fear, guilt, shame, panic, what irreversible decision have I made, I am not a drunk, I am a bad mother • Don't be judgemental – have follow-up information to hand over straight away, so if yes you did drink during pregnancy then here's the name of a good counsellor, children's hospital department and a recommended website • Focus on the future, not on the past • Explain what are you going to do with this information and how it will benefit the child if there are any developmental problems with the child
Information to public	• Priority is prevention, need national campaign – TV, posters, coasters, fridge magnets, social media – pubs, clubs, bars, behind toilet doors, Centre Link, Medicare, doctors and clinic waiting rooms, buses • Public campaign will help women who don't go to the doctor and also make it easier for a health professional to raise • Don't put too much emphasis on doctors to educate – put it out there in the media • Put warning labels on alcohol eg USA • Alcohol companies should put money back into education
Information to women	• Explain how alcohol actually affects the baby – how alcohol crosses the placenta, everything the mother drinks reaches the baby and the baby will be drunk with her (You drink, your baby drinks) • Describe the consequences of alcohol to the baby – what can happen to their brain and development • Recommend "no alcohol" zero tolerance – be honest , there is no known safe limit • Awareness that even though not curable correct diagnosis can help with strategies • Visual tools

	• Tailor the education to the community you are delivering to • Tell women no level of alcohol is safe at any time – misconception that only impacts up to 12 weeks • The truth • Consistent and accurate messages • Can people tell looking at my baby if I've had grog when pregnant?
Language	• Simplify the terminology – break down big words so people understand and at a level people can relate to • Consider language barriers – non English speaking, where English is not the first language, linguistic cultural issues • Language and how information set out –pictorial rather than lots of words • User friendly
Resources	• Use visual aids to help explain how alcohol gets to the baby and how it can affect the baby – FAS dolls/FAS brain models, egg curdling by addition of alcohol • Need pictures/photos of children with FASD, must be real and culturally appropriate • Pamphlets and brochures that can be taken away • Resources should be available in urban, regional and remote communities
Schools	• Should be part of school drug and alcohol health education for 12 – 16 year olds. Not specifically alcohol use on pregnancy as a stand-alone subject, could use school health nurses • Educate young people about the effects of drinking on babies. Focus on the positives of how to have a healthy baby • Teachers need more information on FASD and referral pathways • Not OK for a teacher to ask about alcohol use
Timing	• Information to women and community on alcohol use in pregnancy so women better informed before they get pregnant • Health professionals to talk about alcohol use before women become pregnant and at regular visits to GP by young women and women who may be contemplating becoming pregnant • Information in antenatal classes • Asking about alcohol before or early in pregnancy means a woman would be more comfortable about being asked at birth – if you drank and were asked after pregnancy there is judgment and feeling that you have damaged your child • Routine questions on alcohol use to commence from first pregnancy visit – contradictory view was that a health professional should not ask a woman about her alcohol use at the first visit as a level of rapport needs to be built up before asking deep and personal questions such as alcohol use • Part of a routine set of questions asked by the midwife of all women at birth – should not be in an admission pack questionnaire • Consistent reminder to everyone all the time

KEY ISSUES FROM THE COMMUNITY CONVERSATIONS

Key issues were identified from the community conversations held in Perth and Cairns.

- Prevention – national awareness campaign
- Messages in campaign and from health professionals must be consistent
- FASD is not curable – it's for life, but correct diagnosis can help with strategies to assist the child and family
- Instrument needs to provide a guide and referral pathways and must be appropriate for all Australian children and for different ages and throughout Australia (remote, regional, urban)
- Informed consent – parents/guardians/kinship carers should be asked if they want to proceed with screening
- Standard set of questions (diet, smoking, alcohol, lifestyle etc) that are asked by health professionals (GP, obstetrician, midwife, child health nurse) of all women, no individual singled out and reduce the opportunity for judgemental questions
- Education and training for health professionals – all health professionals (not just paediatricians) need education and training on FASD otherwise the instrument will not be used and children will not be screened or diagnosed
- Education for young people in schools

EVALUATION

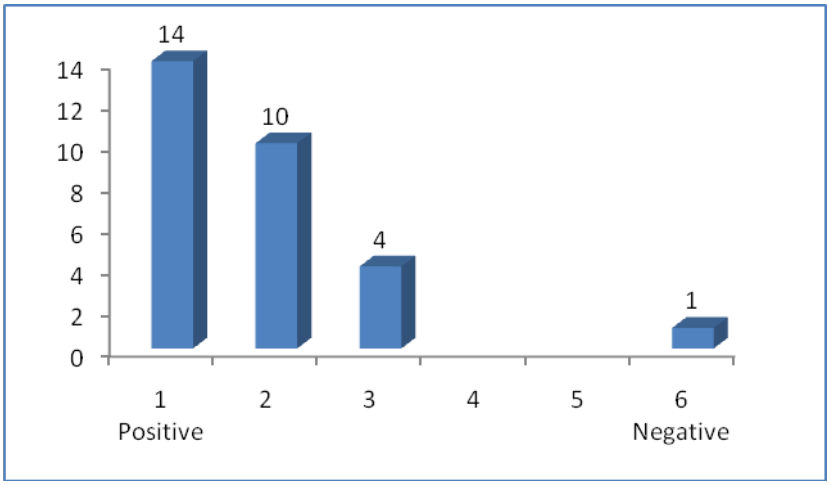
In line with good practice all participants were encouraged to complete an evaluation form. Women were asked to rank their opinion of their experience (one = positive through to 6 = negative)

Evaluation

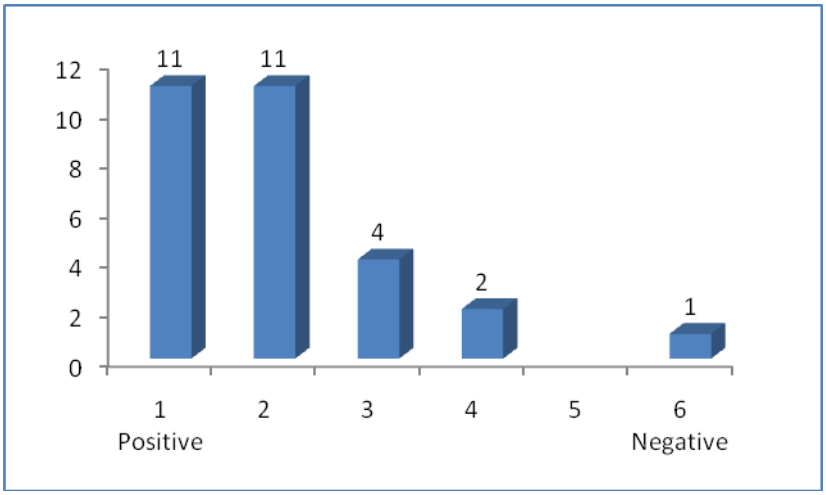
32 people attended the two alcohol & pregnancy community conversations

30 evaluation forms were returned but not all attendees completed all sections

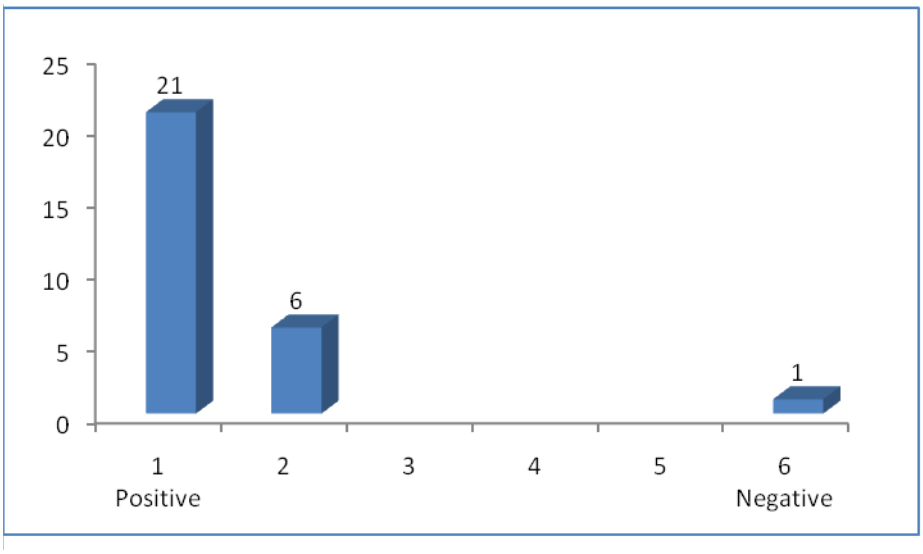
1.1 The 'community conversation' was informative: (N = 29)



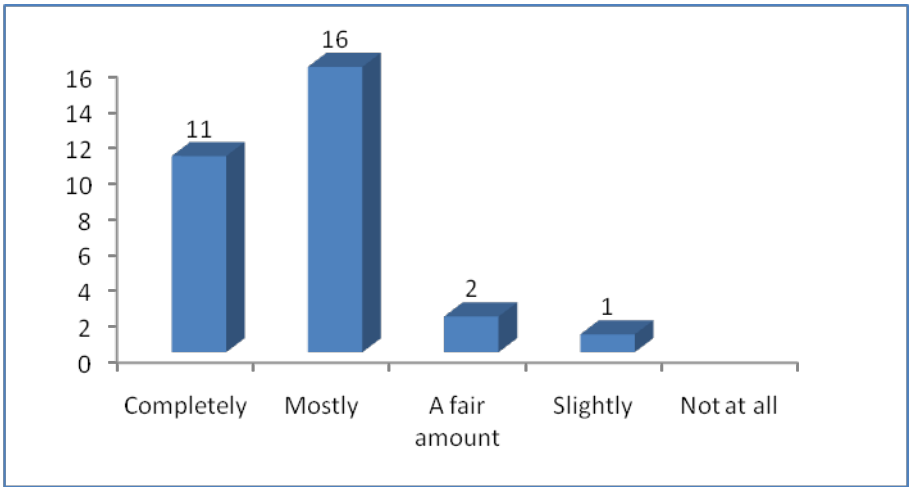
1.2 The 'community conversation' was useful: (N = 29)



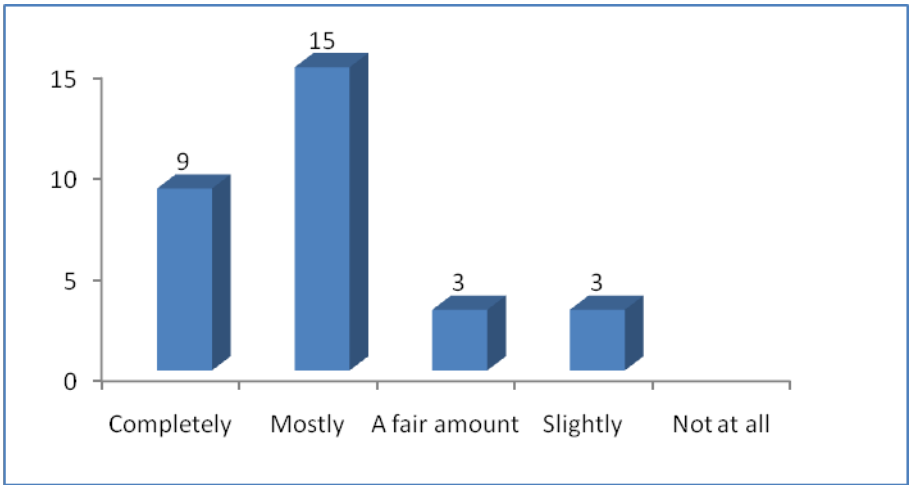
1.3 The 'community conversation' was participative: (N = 28)



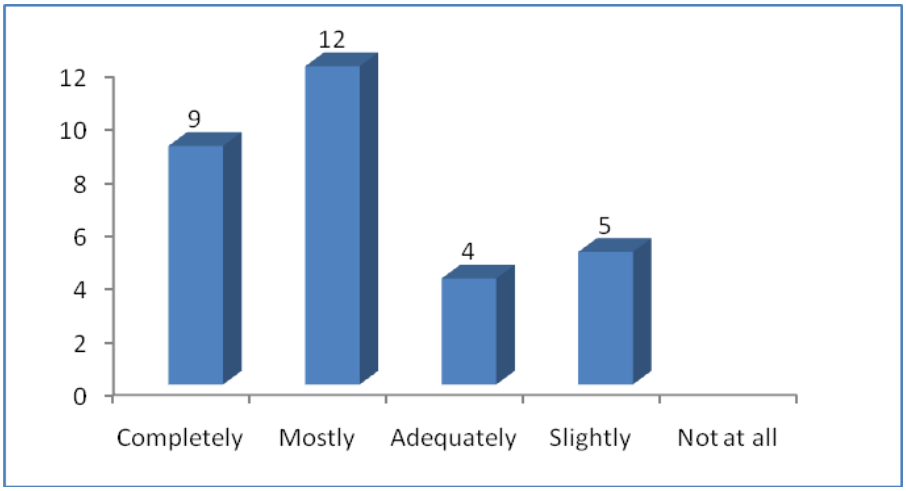
2. Did the 'community conversation' meet your expectations? (N = 30)



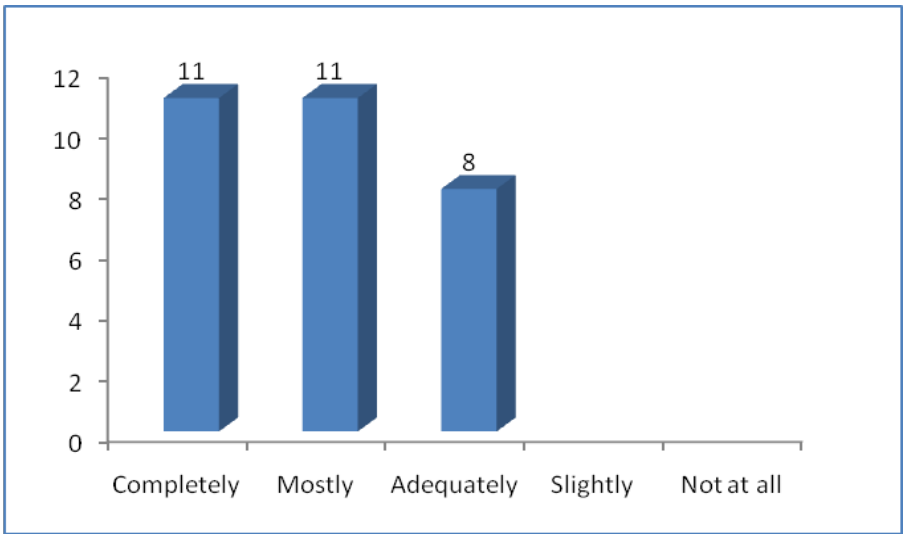
3. Did the 'community conversation' cover most areas that were important to you? (N = 30)



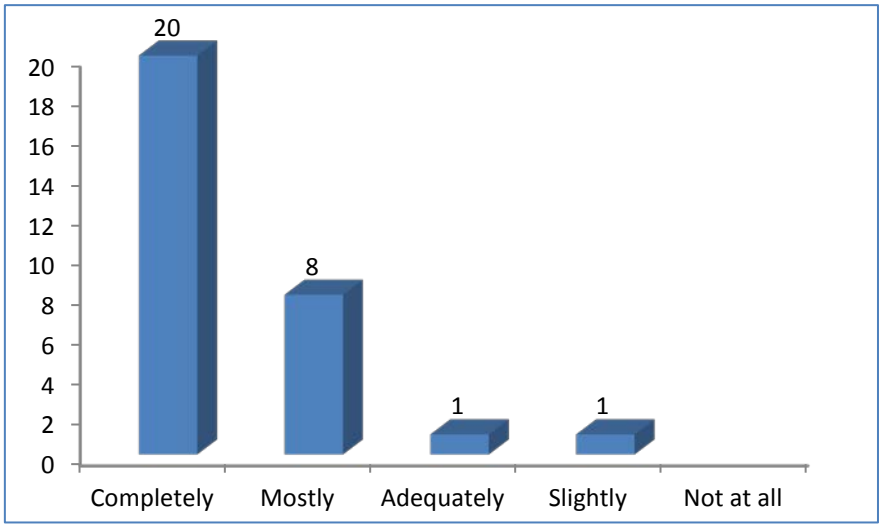
4. Did the presentation on current research projects provide enough information? (N =30)



5. How well were your questions answered? (N = 30)



6. Did you have an opportunity to put forward your ideas/priorities for research? (N = 30)



7. Is there anything else you would like to add?

- Because we can say/ask anything not on the specific global café process questions during lunch
- Thank you for the opportunity
- Felt a more age unified conversation group would have allowed everyone to express their different opinions
- Doctors need to give correct information 'no alcohol is safe' when pregnant
- Focus on developing screening when issue is bigger
- Get into schools
- Too many technical terms & too much information on slides
- As a health professional I would like to hear more about current research and recommendations
- Would have been beneficial to have more information about FASD & issues surrounding diagnosis
- More information about signs/symptoms of FAS as well as behaviours
- Health professionals have a different perspective from community members
- Questions repetitive about health professionals
- Internet networking of FASD would be great to access up to date sharing of information and resources
- A big thank you to Elizabeth Russell – an inspiring story

8. The <u>best</u> thing about the 'community conversation' was:

- Good opportunity to put forward views and was very informative
- Meeting people about FASD research and information sharing
- Opportunity to meet & interact with variety of different people, contribute to research & learn about the problem
- Meeting people who are concerned
- Different views
- Gaining information and opinions on a topic with which I was not familiar
- Learning other ideas
- Interactions with others
- Brainstorming
- Everyone getting their chance to have a say
- Having people interested in what we say

- Being in a group that gets to bounce ideas off each other
- Hearing different opinions
- Interaction, discussion and networking
- Information sharing
- I enjoyed the whole process
- All of it
- Our voices and points of view were heard
- The concept of the 'world café process'. It gave all participants the ability to put their ideas forward

9. The <u>worst</u> thing about the 'community conversation' was:

- Questions repetitive
- More distinct topics or different parts of the question @ different tables
- Too rushed
- Need more groups & more diverse questions
- Not enough people representative of 'pregnant people' or the ones who would have to answer the questions and young people who do drink
- Constantly moving tables
- No time for whole room discussion
- Not enough aboriginal groups or organisations participating
- Long time away from breast-fed baby. Maybe informal crèche – give honorarium towards cost of crèche
- Not interested in feedback summary
- Questions not deep enough
- I didn't see what the problem was – it wouldn't be a problem for me
- Not enough women from Cape York communities were present
- More people could have attended who could have benefited
- Nil
- There was no worst thing
- The lack of other community and health members
- Nothing – all good

10. Do you have any suggestions about how we might improve future 'community conversations'?

- Longer session x 3
- More time on the questions x 2
- More pre-information on the questions x 5
- Different process to the 'world café' 6
- Other (please specify):
  - ✓ Facilitators should move
  - ✓ Email answers so process not so long
  - ✓ Talk over questions before answering and specify what sort of response is wanted
  - ✓ More facilitators & mixed groups
  - ✓ Change nothing
  - ✓ Time for more questions to 'speakers'
  - ✓ Local statistics and concerns
  - ✓ Involvement of other organisations working in FASD people/issues
  - ✓ More advertising
  - ✓ Perhaps more specific groups. I realise a lot were targeted and didn't come
  - ✓ Can't think of a way to improve
  - ✓ Visit Cape communities during the 'off pay' week, in the evening

11. Would you be interested in attending future 'community conversations' on other research areas at the Institute?

- *Yes x 26*
- *No x 1*
- *Maybe x 2*

A copy of the evaluation form can be found in Appendix 4.

CONCLUSION

As set out in the NHMRC *Statement on Consumer and Community Participation in Health and Medical Research*[1] the collaboration of consumers and researchers to draw upon each other's knowledge will build on and strengthened the quality of health and medical research in Australia. As end users of health and medical research, consumers can provide valuable input to decisions about medical research and practices.

The outcomes from the two alcohol and pregnancy community conversations, together with current Australian research and Delphi process outcomes, will be used to inform the Australian Collaboration when developing the screening and diagnostic instrument and associated guidance notes for fetal alcohol spectrum disorders in Australia. This valuable community information supports and extends findings of Peadon and France.

Peadon et al[2,3] found that women expected their health professional to ask and advise them about alcohol and pregnancy; however Payne et al[4] concluded that only 45% of health professionals routinely ask about alcohol use in pregnancy. The France et al[5] survey indicated that some health professionals were making an assumption that women knew to minimise alcohol consumption during pregnancy. These findings are supported by the statements from the women participating in the two community conversations. (See the summary of statements by participants and the key issues listed on pages 11 and 16 respectively)

As discussed on page 4, the FASD Project will use a Delphi process to reach a consensus on what should be included in the screening and diagnostic instrument. To develop the statements and questions a systematic review of literature was conducted and a report provided to the Steering Group. The report of outcomes from the community conversations was also provided to the subgroup responsible for developing the Delphi instrument. An example of a statement in the Delphi instrument which arose from the community conversations and the ranking scale is listed below.

Statement from Prenatal Alcohol Exposure Section

4.24 Alcohol exposure should be assessed alongside other lifestyle factors including diet, physical exercise and smoking

Strongly agree	Agree	Neither	Disagree	Strongly disagree

The Steering Group will hold a two-day workshop to finalise the screening and diagnostic instrument and the Community Conversation Report will be considered alongside the published evidence and outcomes from the Delphi process.

Issues arising from the community conversation specific to the instrument:

- Standard set of questions (diet, smoking, drugs, lifestyle etc) that are asked by health professionals (GP, obstetrician, midwife, maternal/child health nurse) of all 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 – don't use medical terminology
- Informed consent – parents/guardians/kinship carers should be asked if they want to proceed with screening for FASD
- Instrument needs to provide a guide and referral pathways
- Appropriate for all Australian children (urban, rural and remote) and for different ages

Not all statements from the community conversations were specific to the development of the screening and diagnostic instrument. However these statements are relevant to the topic of alcohol and pregnancy and FASD.

Issues arising from the community conversations that are related to the introduction of the screening and diagnostic instrument 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 not just for paediatricians – children in rural and remote areas will not have ready access to a paediatrician. Education and training important for GPs, 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 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.

Issues arising from the community conversations that are related to alcohol and pregnancy and FASD that are important but outside the scope of the FASD Project:

- Prevention is the key – national awareness campaign with information in a range of media and venues – TV, radio, social media (YouTube, Twitter etc), buses, clubs, bars, restaurants, nightclubs, Centre Link, Medicare, doctors and clinic waiting rooms
- Education in schools (12 – 16 year olds)

The Steering Group will ensure that the Delphi statements reflect the community conversation information and, at the Steering Group Workshop, they will be considered as the screening and diagnostic instrument is finalised. Some may also be incorporated into recommendations arising from the FASD Project.

The final FASD Project Report will include a section on the community conversations and how the outcomes were incorporated into the final instrument and identified in the recommendations.

APPENDIX 1(FULL COMMUNITY CONVERSATION REPORT)

COMMUNITY CONVERSATION INVITATION

We would like to invite you to a *'Community Conversation'* seminar to be held in Perth/Cairns on alcohol and pregnancy. The seminar will give an overview of the current research projects on alcohol use in pregnancy. There will also be an opportunity for you to put forward your ideas on how you would like your health professionals to talk to you about alcohol and pregnancy and the possible risks of drinking alcohol while pregnant.

The seminar is part of the Fetal Alcohol Spectrum Disorders Project. Fetal Alcohol Spectrum Disorders is not a diagnosis but an umbrella term used to describe a range of potential effects resulting from fetal alcohol exposure. These effects include physical defects, facial abnormalities and problems with growth, development and learning.

The seminar is partly funded through the project and also sponsored by the Institute's Consumer and Community Advisory Council and the Collaboration for Applied Research and Evaluation.

Attendees at the seminar will receive a small honorarium to cover parking and any out of pocket expenses. As places are very limited (maximum 30 at each location) we would appreciate your early response.

We are really excited about the potential for this *'Community Conversation'* and do hope that you will join us.

Details for the seminar in Perth on page 2/Details for the seminar in Cairns on page 2

Kind regards
Julie Ireland
Chair, Consumer & Community Advisory Council
and
Carol Bower
Senior Principal Research Fellow
Telethon Institute for Child Health Research

PERTH

Date	7 December 2010
Time	10.00am – 2.30pm
Venue	Telethon Institute for Child Health Research, Roberts Road Subiaco
RSVP	30 November 2010
	Phone: 08 9489 7724
	Email: hjones@ichr.uwa.edu.au

How to get there

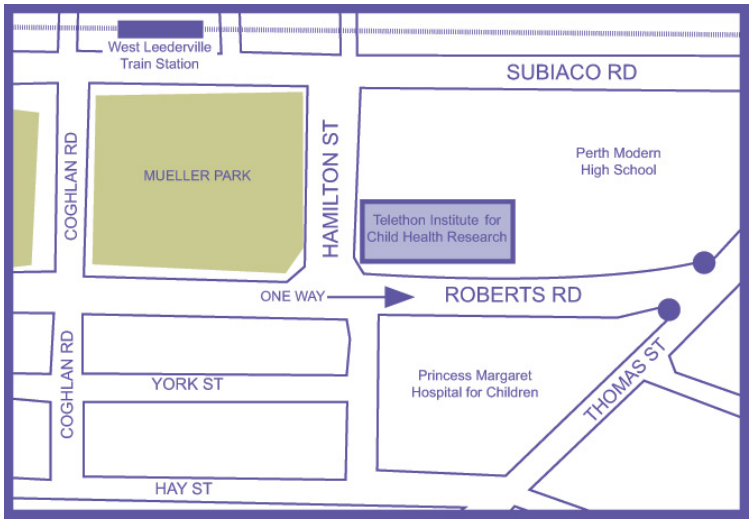
Please note that parking is very limited so we suggest that people allow plenty of time to find a parking spot.

Paid parking is available:

- on Hamilton Street between Roberts Road and Subiaco Road
- on Roberts Road
- at Subiaco Football Oval
- in the York Street car park between Hamilton Street and Coghlan Road.

West Leederville Station is only a few minutes walk from the Institute and Subiaco Station is approximately 15 minutes walk from the Institute.

There is a free Cat Bus that comes from the city to Princess Margaret Hospital (The institute is adjacent to the hospital)



CAIRNS

Date: Friday 18 February 2011

Time: 9.00am – 1.30pm

Venue: Pacific International Hotel, 43 The Esplanade Cairns

RSVP: 4 February 2011

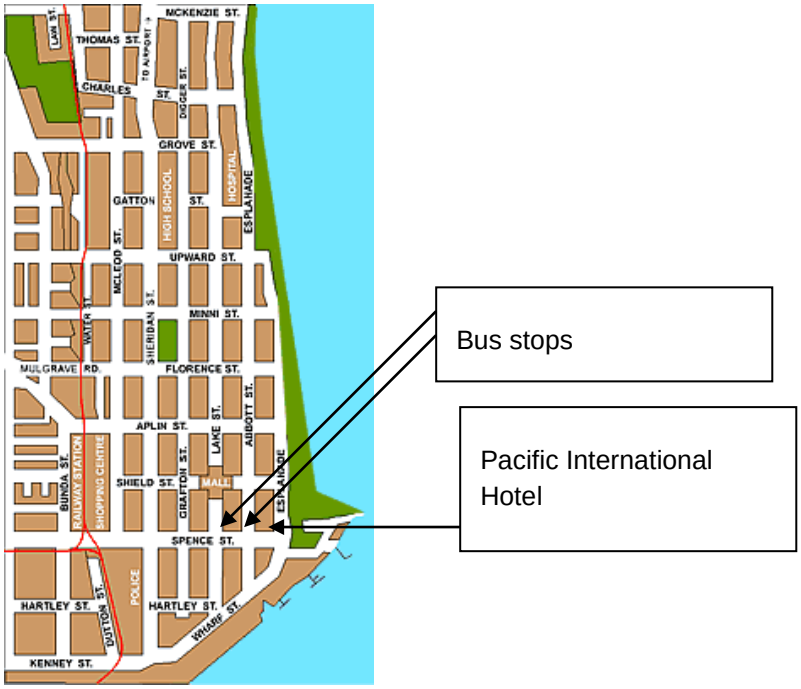
Phone: 08 9489 7724 (please call and we will phone you back)

Email: hjones@ichr.uwa.edu.au

How to get there

There is no parking at the Pacific International Hotel. Paid parking is available across the road from the hotel.

If you are coming via the local bus service, the nearest bus stop would be either in Abbott Street (1 street away) or Lake Street (2 streets away).



The Telethon Institute for Child Health Research

Consumer and Community Advisory Council

and the Fetal Alcohol Spectrum Disorders Project

'Community Conversation'

Alcohol & Pregnancy

Telethon Institute for Child Health Research

Agenda

10.00am	Registration & morning tea
10.30am	Welcome & Introductions
10.35am	Outline & Aims for the day
10.40am	Alcohol & Pregnancy Presentation
11.10am	World Café Process
11.15am	Question 1
11.45am	Question 2
12.10pm	*Lunch Break*
12.40pm	Question 3
1.10pm	Question 4
1.35pm	*Short Break*
1.50pm	Reports from each table & questions
2.20pm	Evaluation and thanks

The Telethon Institute for Child Health Research

Consumer and Community Advisory Council

and the Fetal Alcohol Spectrum Disorders Project

'Community Conversation'

Alcohol & Pregnancy

Pacific International Hotel Cairns

Agenda

9.00am	Registration
9.30am	Welcome & Introductions
9.35am	Outline & Aims
9.40am	Fetal Alcohol Spectrum Disorders
10.10am	World Café Process
10.15am	*Morning Tea Break*
10.35am	Question 1
11.05am	Question 2
11.35am	Question 3
12.00pm	*Lunch Break*
12.45 pm	Question 4 – whole group discussion
1.10 pm	Summary from Questions 1 -3
1.30pm	Questions
1.40pm	Evaluation and thanks

APPENDIX 3 (FULL COMMUNITY CONVERSATION REPORT)

PERTH COMMUNITY CONVERSATION QUESTIONS

Question 1

a) If you were pregnant, what would you want your health professional to say to you to about alcohol?
b) How would you want the health professional to raise it with you? Are there ways of asking that might work better for particular groups of women or that account for cultural sensitivities?
c) What information would you want a health professional to give you?

Question 2

HOW WOULD YOU FEEL IF YOU WERE PREGNANT AND ASKED TO PROVIDE MORE DETAIL ABOUT YOUR ALCOHOL USE? THIS INFORMATION COULD include:

a) When during the nine months of your pregnancy did you drink alcohol (months 1-3, months 4-6, months 7-9)?
b) How much alcohol did you drink at each occasion (for example 3 full strength beers, 1 glass of wine)?
c) How frequent were those occasions when you drank alcohol (for example three times a day, daily, weekly etc)?
d) What do you think is the best way for a health professional to ask these questions? Are there ways of asking that might work better for particular groups of women or that account for cultural sensitivities?

Question 3

If you had just given birth, would you agree to answer a question (or questions) about your alcohol use during pregnancy?

a) What do you think is the best way for a health professional to ask this question? Are there ways of asking that might work better for particular groups of women or that account for cultural sensitivities?

Question 4

If you had a child with *DELAYED DEVELOPMENT, LOW IQ OR LEARNING DIFFICULTIES*, would you agree to answer questions about your alcohol use during pregnancy?

a) What do you think is the best way for a health professional to ask these questions? Are there ways of asking that might work better for particular groups of women or that account for cultural sensitivities?

Do you have any further suggestions that you would like to make regarding this issue?

CAIRNS COMMUNITY CONVERSATION QUESTIONS

Question 1

If you were pregnant, what would you want your health professional to say or provide to you about alcohol use and its potential harm?

Question 2

HOW WOULD YOU FEEL ANSWERING QUESTIONS ABOUT YOUR ALCOHOL USE EITHER DURING PREGNANCY OR STRAIGHT AFTER GIVING BIRTH? THESE QUESTIONS MIGHT INCLUDE:

a) When during the nine months of your pregnancy did you drink alcohol (months 1-3, months 4-6, months 7-9)?
b) How much alcohol did you drink at each occasion (for example 3 full strength beers, 1 glass of wine)?
c) How frequent were those occasions when you drank alcohol (for example three times a day, daily, weekly etc)?

Question 3

If you had a child with *DELAYED DEVELOPMENT OR LEARNING DIFFICULTIES*, would you agree to answer questions about your alcohol use during pregnancy?

Question 4 (Whole group)

APPENDIX 4 (FULL COMMUNITY CONVERSATION REPORT)

EVALUATION FORM FOR PERTH AND CAIRNS

Community Conversation Evaluation Form

Please tick or circle the responses which best match your view:

1. The Community Conversation was:

POSITIVE							Negative
a. Informative	1	2	3	4	5	6	Very poor
b. Useful	1	2	3	4	5	6	Not useful
c. Participative	1	2	3	4	5	6	One of two people talked too much

2. Did the Community Conversation meet your expectations?

☐ Completely ☐ Mostly ☐ A fair amount ☐ Slightly ☐ Not at all

3. Did the Community Conversation cover areas that were important to you?

☐ Completely ☐ Mostly ☐ A fair amount ☐ Slightly ☐ Not at all

If “not at all” please specify what additional information could have been included?

4.Did the presentation on current research projects provide enough information?

☐ Completely ☐ Mostly ☐ Adequately ☐ Slightly ☐ Not at all

5. How well were your questions answered?

☐ Completely ☐ Mostly ☐ Adequately ☐ Slightly ☐ Not at all

6.Did you have an opportunity to put forward your ideas/priorities for research?

☐ Completely ☐ Mostly ☐ Adequately ☐ Slightly ☐ Not at all

7. Is there anything else you would like to add?

8. The best thing about the Community Conversation was?

9. The worst thing about the Community Conversation was?

10. Do you have any suggestions about how we might improve future Community Conversation?

- ☐ Longer session
- ☐ More time on questions
- ☐ More pre-information on the questions
- ☐ Different process to the 'world café'
- ☐ Other (please specify)

11. Would you be interested in attending future Community Conversations on other research areas at the Institute?

☐ Yes ☐ No ☐ Maybe

If yes please provide contact details:

Thank you for attending and your valuable feedback

REFERENCES (FULL COMMUNITY CONVERSATION REPORT)

1. *Statement on Consumer and Community Participation in Health and Medical Research (the Statement on Participation)* National Health and Medical Research Council 2000-2001 and updated in 2004. Accessed on-line at http://www.nhmrc.gov.au/publications/synopses/r22syn.htm

2. *Women's knowledge and attitudes regarding alcohol consumption in pregnancy: a national survey*, by Peadon E, Payne J, Henley N, D'Antoine H, Bartu A, O'Leary C, Bower C, Elliott EJ in BMC Public Health, August 2010. *(Telephone interviews with 1103 Australian women 18 – 45 years and not pregnant)*

3. *The role of the General Practitioner* by Peadon E, O'Leary C, Bower C, Elliott E. Impacts of alcohol use in pregnancy.in Aust Fam Physician. 2007; 36(11):935-939.

4. *Health professional's knowledge, practice and opinions about Fetal Alcohol Syndrome and alcohol consumption in pregnancy* by Payne J, Elliott E, D'Antoine H, O'Leary C, Mahony A, Haan E, et al.in Aust N Z J Public Health. 2005; 29(6):558-564.

5. *Health Professionals Addressing Alcohol Use with Pregnant Women in Western Australia: Barriers and Strategies for Communication* by Kathryn France, Nadine Henley, Jan Payne, Heather D'Antoine, Anne Bartu, Colleen O'Leary, Elizabeth Elliott and Carol Bower in Substance Use and Misuse, 45: 1474 – 1490

APPENDIX F: DELPHI QUESTIONNAIRE

APPENDIX F1: ROUND 1 QUESTIONNAIRE

Welcome to the Fetal Alcohol Spectrum Disorders (FASD) Screening and Diagnosis Expert Questionnaire

We are extremely grateful for your valuable contribution to the development of a screening and diagnostic instrument for FASD in Australia.

Instructions

- after you login you will be taken to the questionnaire contents page
  - each section of the questionnaire can be accessed from the contents page
- at the end of each section there are *'save'* and *'submit'* buttons:
  - clicking **'save'** indicates that you need to return to the section and review your response at a later time,
  - clicking **'submit' finalises** your response and the section is considered complete
- this questionnaire will take around 45 minutes to complete, and does not need to be completed in a single sitting
  - **links at the top of each page** allow you to return to the contents page or log out at any time
  - a **progress bar** at the bottom of the contents page shows how close you are to completion of the questionnaire
  - some questions may appear repetitive, as requirements for both screening and diagnosis are evaluated in detail
- we do not expect all participants to be able to provide expert input in all areas of the questionnaire
  - the **'no comment'** option can be used to indicate that the question is outside your area of expertise
- also, the questions may not always reflect your beliefs about how FASD screening and diagnosis should be conducted in Australia
  - if so, **please let us know in the comment boxes provided**
- if you encounter any difficulties, or forget your password, please contact the project manager Heather

Jones

Confidentiality statement

This project has been approved by the Human Research Ethics Committee of The University of Western Australia.
Study participants' identifying information is stored securely and separately from questionnaire responses to protect individual confidentiality.
See the full Telethon Institute for Child Health Research privacy statement here.

Login

Please login using your personal username and password supplied in your invitation email:

Username:

Password:

Login Reset

FASD Questionnaire Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Demographic Information	To be completed
2. Screening Programs	To be completed
3. Targeted Screening	To be completed
4. Screening Providers	To be completed
5. Screening Methods Part 1: Prenatal Alcohol Exposure	To be completed
6. Screening Methods Part 2: Growth Deficit, Facial Anomalies and Birth Defects	To be completed
7. Screening Methods Part 3: Central Nervous System Abnormalities	To be completed
8. Definition of Abnormal Screening Findings	To be completed
9. Criteria for Conducting a Full Diagnostic Evaluation for FASD	To be completed
10. Diagnostic Systems and Guidelines	To be completed
11. Diagnostic Processes	To be completed
12. Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)	To be completed
13. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders (FASD)	To be completed
14. Acknowledgement and Feedback	To be completed

Progress: 0% of sections completed

FASD Questionnaire Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Demographic Information	Completed
2. Screening Programs	Completed
3. Targeted Screening	Completed
4. Screening Providers	Completed
5. Screening Methods Part 1: Prenatal Alcohol Exposure	Completed
6. Screening Methods Part 2: Growth Deficit, Facial Anomalies and Birth Defects	Completed
7. Screening Methods Part 3: Central Nervous System Abnormalities	Completed
8. Definition of Abnormal Screening Findings	Completed
9. Criteria for Conducting a Full Diagnostic Evaluation for FASD	Completed
10. Diagnostic Systems and Guidelines	Completed
11. Diagnostic Processes	Review
12. Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)	To be completed
13. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders (FASD)	To be completed
14. Acknowledgement and Feedback	To be completed

Progress: 71% of sections completed

FASD Questionnaire Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Demographic Information	Completed
2. Screening Programs	Completed
3. Targeted Screening	Completed
4. Screening Providers	Completed
5. Screening Methods Part 1: Prenatal Alcohol Exposure	Completed
6. Screening Methods Part 2: Growth Deficit, Facial Anomalies and Birth Defects	Completed
7. Screening Methods Part 3: Central Nervous System Abnormalities	Completed
8. Definition of Abnormal Screening Findings	Completed
9. Criteria for Conducting a Full Diagnostic Evaluation for FASD	Completed
10. Diagnostic Systems and Guidelines	Completed
11. Diagnostic Processes	To be completed
12. Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)	To be completed
13. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders (FASD)	To be completed
14. Acknowledgement and Feedback	To be completed

Progress: 71% of sections completed

Section 1: Demographic Information

This section will help us to provide a general description of study participants

For each question please select a response from the list, or type your response in the box provided.

Q1. In which country do you reside?

(•) Australia ( ) Other

Q1.1 Please specify in which Australian state or territory: select one...

Q2. Please specify your sex:

( ) female ( ) male

Q3. What is your main occupation? select one... **or**

Q4. How long have you been working in that occupation? select one... years

Q5. In which locations do you work? (select all that apply)

- [ ] metropolitan
- [ ] regional
- [ ] rural
- [ ] remote

Q6. Have you ever been involved in **screening** for FASD?

○ no ◉ yes

Q6.1 How many years experience do you have in screening for FASD? select one... years

Q7. Have you ever been involved in **diagnosing** FASD? (making the ***full diagnosis***)

○ no ◉ yes

Q7.1 How many years experience do you have in diagnosing FASD? select one... years

Q7.2 Approximately how many cases have you diagnosed? select one... cases

Q8. Have you ever **contributed to the diagnosis** of FASD? (e.g. assessed a child, but ***not made the full diagnosis***)

○ no ◉ yes

Q8.1 How many years experience do you have in contributing to the diagnosis of FASD? select one... years

Q8.2 Approximately how many cases have you contributed to the diagnosis of? select one... cases

Q9. Have you ever completed specific training on screening for FASD?

○ no ○ yes

Q10. Have you ever completed specific training on diagnosis of the FASD?

○ no ○ yes

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 2: Screening Programs

*This section examines the **timing and methods** of screening for Fetal Alcohol Spectrum Disorders in Australia.*

Screening Coverage

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. Screening for FASD at birth should be universal	○	○	○	○	○	○
Q2. Screening for FASD at birth should be targeted	○	○	○	○	○	○
Q3. Screening for FASD in childhood should be universal	○	○	○	○	○	○
Q4. Screening for FASD in childhood should be targeted	○	○	○	○	○	○

Please qualify your responses to the above questions about screening coverage:

Screening at Birth

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Screening for FASD at or around birth should assess and record:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q5. prenatal alcohol exposure	○	○	○	○	○	○
Q6. birth weight, length and head circumference	○	○	○	○	○	○

Q7. fatty acid esters (FAEE) in meconium collected within 72 hours of birth	○	○	○	○	○	○
Q8. characteristic FAS facial anomalies	○	○	○	○	○	○
Q9. birth defects	○	○	○	○	○	○
Q10. evidence of withdrawal from alcohol or other drugs	○	○	○	○	○	○

Q11. Do any other characteristics need to be assessed during screening for FASD at birth? ○ **no** ◉ **yes** ○ **no comment**

Q11.1 Please specify the other characteristics that need to be assessed during screening for FASD at birth:

Enter any comments about screening for FASD at birth:

Screening in Childhood

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Screening for FASD in childhood should assess and record:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q12. prenatal alcohol exposure	○	○	○	○	○	○
Q13. growth (height and weight)	○	○	○	○	○	○
Q14. head circumference	○	○	○	○	○	○
Q15. developmental delay	○	○	○	○	○	○
Q16. neurological signs	○	○	○	○	○	○
Q17. functional central nervous system abnormalities (e.g. cognition, behaviour disorders)	○	○	○	○	○	○
Q18. hearing and vision	○	○	○	○	○	○
Q19. characteristic FAS facial anomalies	○	○	○	○	○	○
Q20. birth defects	○	○	○	○	○	○

Q21. Do any other characteristics need to be assessed during screening for FASD in childhood? ○ **no** ◉ **yes** ○ **no comment**

Q21.1 Please specify the other characteristics that need to be assessed during screening for FASD in childhood:

Enter any comments about screening for FASD in childhood:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 3: Targeted Screening

*This section examines the most appropriate **indications** for targeted screening for Fetal Alcohol Spectrum Disorders in Australia.*

Indications for Targeted Screening

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Targeted screening for FASD should be conducted for all children who present with:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. an alcohol-related event, illness or dependency in the birth mother	○	○	○	○	○	○
Q2. a parent or foster parent who is concerned that their child might have a FASD	○	○	○	○	○	○
Q3. prenatal alcohol exposure	○	○	○	○	○	○

In the absence of other known causes, targeted screening for FASD should be conducted for all children who present with:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q4. developmental delay	○	○	○	○	○	○
Q5. growth retardation or failure to thrive	○	○	○	○	○	○
Q6. structural central nervous system abnormalities	○	○	○	○	○	○
Q7. neurological signs	○	○	○	○	○	○
Q8. functional central nervous system abnormalities	○	○	○	○	○	○
Q9. characteristic FAS facial anomalies	○	○	○	○	○	○
Q10. birth defects	○	○	○	○	○	○
Q11. reported or observed problems with behaviour	○	○	○	○	○	○

Q12. Should any other presentations prompt targeted screening for FASD? ○ **no** ◉ **yes** ○ **no comment**

Q12.1 Please specify the other presentations that should prompt targeted screening for FASD:

Enter any comments about targeted screening for FASD:

Targeted Screening in Other High Risk Groups

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

The following children should be screened for FASD:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q13. children of mothers attending alcohol treatment services	○	○	○	○	○	○
Q14. siblings of identified cases of FASD	○	○	○	○	○	○
Q15. children who are diagnosed with ADHD	○	○	○	○	○	○

All children should be screened for FASD when they enter:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q16. a child development service	○	○	○	○	○	○
Q17. child protection	○	○	○	○	○	○
Q18. foster care or adoptive placements (including kinship care)	○	○	○	○	○	○
Q19. a juvenile justice setting	○	○	○	○	○	○

Q20. Should any other groups receive targeted screening for FASD? ○ **no** ◉ **yes** ○ **no comment**

Q20.1 Please specify the other high risk groups that need to be screened for FASD:

Enter any comments about screening for FASD in high risk groups:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 4: Screening Providers

*This section examines **who** should provide screening for Fetal Alcohol Spectrum Disorders in Australia.*

Screening Providers

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

The following health professionals should screen for FASD:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. paediatricians	○	○	○	○	○	○
Q2. neonatologists	○	○	○	○	○	○
Q3. obstetricians	○	○	○	○	○	○
Q4. psychiatrists	○	○	○	○	○	○
Q5. general practitioners	○	○	○	○	○	○
Q6. nurse practitioners	○	○	○	○	○	○
Q7. midwives	○	○	○	○	○	○
Q8. child health nurses	○	○	○	○	○	○
Q9. school health nurses	○	○	○	○	○	○
Q10. community health nurses	○	○	○	○	○	○
Q11. occupational therapists	○	○	○	○	○	○
Q12. speech pathologists	○	○	○	○	○	○
Q13. social workers	○	○	○	○	○	○
Q14. psychologists	○	○	○	○	○	○
Q15. physiotherapists	○	○	○	○	○	○
Q16. Aboriginal health workers	○	○	○	○	○	○

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q17. All health professionals who screen for FASD require appropriate FASD-specific training	○	○	○	○	○	○

Enter any comments about which professionals should screen for FASD, or the training required for screening providers:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 5: Screening Methods Part 1: Prenatal Alcohol Exposure

This section examines the assessment of prenatal alcohol exposure during screening for Fetal Alcohol Spectrum Disorders in Australia.

Assessment of Prenatal Alcohol Exposure

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Assessment of prenatal alcohol exposure should identify and record the:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. number of standard alcoholic drinks consumed during a typical drinking occasion	○	○	○	○	○	○
Q2. frequency of drinking occasions	○	○	○	○	○	○
Q3. frequency of excessive (binge) drinking (5 or more standard drinks per occasion)	○	○	○	○	○	○
Q4. timing of alcohol intake during pregnancy	○	○	○	○	○	○

Q5. Should any other aspects of prenatal alcohol exposure be assessed and recorded? ○ **no** ◉ **yes** ○ **no comment**

Q5.1 Please specify the other aspects of prenatal alcohol exposure that should be assessed and recorded:

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q6. Alcohol exposure should be assessed at every antenatal visit	○	○	○	○	○	○
Q7. Alcohol exposure should be assessed alongside other lifestyle factors (e.g. diet)	○	○	○	○	○	○

Q8. Prenatal alcohol exposure can be effectively assessed using an informal approach (e.g. inquiring during a consultation)	○	○	○	○	○	○
Q9. Prenatal alcohol exposure should be assessed using a formal tool (e.g. AUDIT C)	○	○	○	○	○	○

Prenatal alcohol exposure should be assessed using one of the following tools:	**no**	**yes**	**unsure**	**not familiar with this tool**
Q10. AUDIT-C (3-item Alcohol Use Disorders Identification Test)	○	○	○	○
Q11. Lifescripts tool (includes 3-item AUDIT-C)	○	○	○	○
Q12. TWEAK (5-item Tolerance, Worry, Eye-opener, Amnesia, Cut down)	○	○	○	○
Q13. T-ACE (4-item: Tolerance, Annoyed, Cut down, Eye opener)	○	○	○	○

Q14. Should any other methods be used to assess prenatal alcohol exposure? ○ **no** ◉ **yes** ○ **no comment**

Q14.1 Please specify the other methods that should be used to assess prenatal alcohol exposure:

Enter any comments about the assessment of prenatal alcohol exposure in FASD screening:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 6: Screening Methods Part 2: Growth Deficit, Facial Anomalies and Birth Defects

This section examines the assessment of growth deficit, facial anomalies and birth defects during screening for Fetal Alcohol Spectrum Disorders in Australia.

Assessment of Growth Deficit

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. Growth should be assessed by comparing height and weight with population standards	○	○	○	○	○	○
Q2. Growth should be assessed by comparing weight to height ratio with population standards	○	○	○	○	○	○
Q3. Growth should be assessed by comparing weights over time (to identify decelerating weight over time)	○	○	○	○	○	○
Q4. Assessment of growth deficit should consider other factors that may affect growth (e.g. gestational age, parental size, gestational diabetes, nutritional status, illness)	○	○	○	○	○	○

Q5. Should any other methods be used to assess growth in FASD screening? ○ **no** ◉ **yes** ○ **no comment**

Q5.1 Please specify the other methods that should be used to assess growth in FASD screening:

Enter any comments about the assessment of growth in FASD screening:

Assessment of Characteristic Fetal Alcohol Syndrome (FAS) Facial Anomalies

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q6. The presence of the following characteristic FAS facial anomalies should be assessed: smooth philtrum, thin upper lip, and small palpebral fissures	○	○	○	○	○	○
Q7. Assessment of characteristic FAS facial anomalies should use appropriate anthropometric population standards for race and age where available	○	○	○	○	○	○

Q8. Do any other facial anomalies need to be assessed in FASD screening? ○ **no** ◉ **yes** ○ **no comment**

Q8.1 Please specify the other facial anomalies that need to be assessed in FASD screening?:

At the screening stage, characteristic FAS facial anomalies can be effectively assessed using:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q9. clinical observation	○	○	○	○	○	○
Q10. physical measurement of palpebral fissures	○	○	○	○	○	○
Q11. the University of Washington Lip-Philtrum Guide	○	○	○	○	○	○
Q12. the facial photographic screening tool	○	○	○	○	○	○

Q13. Should any other methods be used to assess characteristic FAS facial anomalies in FASD screening? ○ **no** ◉ **yes** ○ **no comment**

Q13.1 Please specify the other methods that should be used to assess characteristic FAS facial anomalies in FASD screening:

Enter any comments about the assessment of facial anomalies in FASD screening:

Assessment of Birth Defects

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q14. FASD screening should assess and record the presence of birth defects as part of the clinical examination	○	○	○	○	○	○

Enter any comments about the assessment of birth defects in FASD screening:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 7: Screening Methods Part 3: Central Nervous System Abnormalities

This section examines the assessment of central nervous system abnormalities during screening for Fetal Alcohol Spectrum Disorders in Australia.

Assessment of Central Nervous System (CNS) Abnormalities

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Assessment of CNS abnormalities in FASD screening may include:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. developmental milestones	○	○	○	○	○	○
Q2. motor and sensory function	○	○	○	○	○	○
Q3. cognition (IQ)	○	○	○	○	○	○
Q4. memory	○	○	○	○	○	○
Q5. academic achievement	○	○	○	○	○	○
Q6. executive functioning and abstract reasoning	○	○	○	○	○	○
Q7. adaptive behaviour	○	○	○	○	○	○
Q8. attention and hyperactivity	○	○	○	○	○	○
Q9. communication (receptive and expressive language)	○	○	○	○	○	○
Q10. social skills and social communication	○	○	○	○	○	○
Q11. hard and soft neurologic signs (including sensory-motor signs)	○	○	○	○	○	○
Q12. seizures that are not due to a postnatal insult or other postnatal process	○	○	○	○	○	○
Q13. head circumference	○	○	○	○	○	○
Q14. brain imaging	○	○	○	○	○	○

Q15. Do any other characteristics need to be assessed in FASD screening to identify CNS abnormalities?

○ **no** ◉ **yes** ○ **no comment**

Q15.1 Please specify the other characteristics that need to be assessed in FASD screening to identify CNS abnormalities:

The choice of tests for neuro-behavioural assessments should be guided by:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q16. the availability of valid and reliable instruments	○	○	○	○	○	○
Q17. clinician preference and experience	○	○	○	○	○	○
Q18. test appropriateness for patient age and cultural background	○	○	○	○	○	○

Enter any comments about the assessment of CNS abnormalities in FASD screening:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 8: Definition of Abnormal FASD Screening Findings

*This section examines how abnormal screening findings for Fetal Alcohol Spectrum Disorders are **defined**.*

Definition of Abnormal Screening Findings: Short palpebral fissures

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if outside your area of expertise.*

Short palpebral fissures:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. fissure length at or below the 10th percentile based on comparison with population standards, with physical or photographic measurement of length	○	○	○	○	○	○
Q2. fissure length below the 3rd percentile based on comparison with population standards, with physical or photographic measurement of length	○	○	○	○	○	○
Q3. positive finding based on visual assessment/clinical impression	○	○	○	○	○	○

Q4. Should any other criteria be used to define short palpebral fissures? ○ **no** ◉ **yes** ○ **no comment**

Q4.1 Please specify the other criteria that should be used to define short palpebral fissures:

Enter any comments about the definition of short palpebral fissures:

Definition of Abnormal Screening Findings: Thin upper lip and smooth philtrum

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if outside your area of expertise.*

Thin upper lip and smooth philtrum:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q5. positive finding based on the University of Washington Lip-Philtrum Guide (rank 4 or 5)	○	○	○	○	○	○
Q6. positive finding based on visual assessment/clinical impression	○	○	○	○	○	○

Q7. Should any other criteria be used to define thin upper lip and smooth philtrum? ○ **no** ◉ **yes** ○ **no comment**

Q7.1 Please specify the other criteria that should be used to define define thin upper lip and smooth philtrum:

Enter any comments about the definition of thin upper lip and smooth philtrum:

Definition of Abnormal Screening Findings: Growth deficit

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if outside your area of expertise.*

Growth deficit:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q8. 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)	○	○	○	○	○	○
Q9. prenatal or postnatal height or weight low for age/gestational age	○	○	○	○	○	○
Q10. disproportionately low weight-to-height ratio at or below the 10th percentile	○	○	○	○	○	○
Q11. disproportionately low weight-to-height ratio	○	○	○	○	○	○

Q12. Should any other criteria be used to define growth deficit? ○ **no** ◉ **yes** ○ **no comment**

Q12.1 Please specify the other criteria that should be used to define growth deficit:

Enter any comments about the definition of growth deficit:

Definition of Abnormal Screening Findings: Structural CNS abnormalities

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if outside your area of expertise.*

Structural CNS abnormalities:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q13. head circumference at or below the 10th percentile	○	○	○	○	○	○
Q14. head circumference below the 3rd percentile	○	○	○	○	○	○
Q15. 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	○	○	○	○	○	○

Q16. Should any other criteria be used to define structural CNS abnormalities? ○ **no** ◉ **yes** ○ **no comment**

Q16.1 Please specify the other criteria that should be used to define structural CNS abnormalities:

Enter any comments about the definition of structural CNS abnormalities:

Definition of Abnormal Screening Findings: Neurological CNS abnormalities

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if*

outside your area of expertise.

Neurological CNS abnormalities (and not due to other known causes):	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q17. presence of hard or soft neurological signs (including sensory-motor signs) based on clinical assessment	○	○	○	○	○	○
Q18. presence of a seizure disorder that is not due to a postnatal insult	○	○	○	○	○	○

Q19. Should any other criteria be used to define neurological CNS abnormalities? ○ **no** ◉ **yes** ○ **no comment**

Q19.1 Please specify the other criteria that should be used to define neurological CNS abnormalities:

Enter any comments about the definition of neurological CNS abnormalities:

Definition of Abnormal Screening Findings: Functional CNS abnormalities

*Please indicate your level of agreement with the following **minimum definitions of abnormality**, **or** select 'no comment' if outside your area of expertise.*

Functional domains may include (but are not limited to): cognitive or developmental deficits or discrepancies; executive functioning deficits; motor functioning delays; problems with attention or hyperactivity; social or communication problems; sensory problems; pragmatic language problems; memory deficits.

Functional CNS abnormalities (and not due to other known causes):	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q20. evidence of functional impairment on standard psychometric testing, with performance 2 or more standard deviations below the mean, assessed by qualified professionals	○	○	○	○	○	○
Q21. evidence of functional impairment on standard psychometric testing, with performance 1 or more standard deviations below the mean, assessed by qualified professionals	○	○	○	○	○	○

Q22. clinical judgement of functional impairment or deficit in domains where standardised measurements are not available	○	○	○	○	○	○
Q23. clinical judgement of functional impairment or deficit based on clinical observation and assessment	○	○	○	○	○	○

Q24. Should any other criteria be used to define functional CNS abnormalities? ○ **no** ◉ **yes** ○ **no comment**

Q24.1 Please specify the other criteria that should be used to define functional CNS abnormalities:

Enter any comments about the definition of functional CNS abnormalities:

Definition of Abnormal Screening Findings: Other considerations

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q25. Evidence of abnormality or dysfunction should be based on normative data	○	○	○	○	○	○
Q26. The population standards required for comparison during FASD screening are available in Australia	○	○	○	○	○	○
Q27. Evidence of abnormality or dysfunction should be based on valid and reliable standard assessment tools where available	○	○	○	○	○	○
Q28. A full diagnostic evaluation for FASD should occur outside standard criteria when health professionals have concerns or doubts about FASD screening results	○	○	○	○	○	○

Enter any comments about the definition of abnormal screening findings:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 9: Criteria for Conducting a Full Diagnostic Evaluation for FASD

There may be a number of combinations of abnormal screening findings (defined in the previous section) that could indicate the need for a full diagnostic evaluation for FASD. This section examines in general terms the combinations of abnormal screening findings that should prompt a full diagnostic evaluation for FASD in Australia.

Criteria for Prenatal Alcohol Exposure

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

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:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. less than 7 standard drinks per week, and no more than 2 standard drinks on any one day	○	○	○	○	○	○
Q2. less than 7 standard drinks per week, and between 3 and 4 standard drinks on any one day	○	○	○	○	○	○
Q3. 7 or more standard drinks per week, and no more than 2 standard drinks on any one day	○	○	○	○	○	○
Q4. 7 or more standard drinks per week, and between 3 and 4 standard drinks on any one day	○	○	○	○	○	○
Q5. binge drinking (5 or more standard drinks per occasion) less than once per week	○	○	○	○	○	○
Q6. binge drinking (5 or more standard drinks per occasion) once or twice per week	○	○	○	○	○	○
Q7. no level of prenatal alcohol exposure is alone sufficient to indicate the need for a full diagnostic evaluation for FASD	○	○	○	○	○	○

Enter any comments about the need for a full diagnostic evaluation for FASD based on alcohol exposure during pregnancy:

Criteria for Other Combinations of Screening Findings

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

In the absence of other known causes, a full diagnostic evaluation for FASD is required when there is evidence of:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q8. concern by a parent or foster parent that their child might have a FASD	○	○	○	○	○	○
Q9. all 3 of the characteristic FAS facial anomalies (smooth philtrum, thin vermillion border, and small palpebral fissures)	○	○	○	○	○	○
Q10. 2 of the characteristic FAS facial anomalies	○	○	○	○	○	○
Q11. the characteristic pattern of FAS facial anomalies (number unspecified)	○	○	○	○	○	○
Q12. 2 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality (structural, neurological or functional)	○	○	○	○	○	○
Q13. 2 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	○	○	○	○	○	○
Q14. 1 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality	○	○	○	○	○	○
Q15. 1 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	○	○	○	○	○	○
Q16. known or probable prenatal alcohol exposure, **and** 1 of the characteristic FAS facial anomalies, **and** a growth deficit **or** any CNS abnormality	○	○	○	○	○	○
Q17. known or probable prenatal alcohol exposure, **and** 1 of the characteristic FAS facial anomalies, **and** a growth deficit **and** any CNS abnormality	○	○	○	○	○	○
Q18. growth deficit **and** any CNS abnormality	○	○	○	○	○	○

Q19. known or probable prenatal alcohol exposure, **and** growth deficit **and** any CNS abnormality	○	○	○	○	○	○
Q20. known or probable prenatal alcohol exposure, **and** any CNS abnormality	○	○	○	○	○	○
Q21. 2 or more CNS abnormalities	○	○	○	○	○	○
Q22. known or probable prenatal alcohol exposure, **and** 2 or more CNS abnormalities	○	○	○	○	○	○
Q23. known or probable prenatal alcohol exposure, **and** 1 or more birth defects	○	○	○	○	○	○

Q24. Should the presence of any other combinations of findings indicate the need for a full diagnostic evaluation for FASD? ○ **no** ◉ **yes** ○ **no comment**

Q24.1 Please describe which other findings indicate the need for a full diagnostic evaluation for FASD:

Enter any comments about the criteria for a full diagnostic evaluation for FASD:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 10: Diagnostic Systems and Guidelines

*This section asks about your experience with **existing diagnostic guidelines** for Fetal Alcohol Spectrum Disorders.*

Existing Diagnostic Systems and Guidelines

Please respond to the questions below.

Are you familiar with the following diagnostic systems or guidelines for FASD:

Q1. Institute of Medicine guidelines (1996)	○ no ◉ yes

Q1.1 Have you used this system or guideline?	○ no ○ yes ○ unsure
Q1.2 Should this system or guideline be adopted as the standard for diagnosis in Australia?	○ no ○ yes ○ unsure ○ no comment
Q1.3 Are you aware of, or have you encountered, any limitations of this system or guideline?	○ no ◉ yes ○ unsure ○ no comment

Q1.3.1 Please describe these limitations:

Q2. University of Washington 4-Digit Diagnostic Code (2004)	○ no ◉ yes

Q2.1 Have you used this system or guideline?	○ no ○ yes ○ unsure
Q2.2 Should this system or guideline be adopted as the standard for diagnosis in Australia?	○ no ○ yes ○ unsure ○ no comment
Q2.3 Are you aware of, or have you encountered, any limitations of this system or guideline?	○ no ◉ yes ○ unsure ○ no comment

Q2.3.1 Please describe these limitations:

Q3. Centers for Disease Control and Prevention (CDC) guidelines (2004)	○ no ◉ yes

Q3.1 Have you used this system or guideline?	○ no ○ yes ○ unsure
Q3.2 Should this system or guideline be adopted as the standard for diagnosis in Australia?	○ no ○ yes ○ unsure ○ no comment
Q3.3 Are you aware of, or have you encountered, any limitations of this system or guideline?	○ no ◉ yes ○ unsure ○ no comment

Q3.3.1 Please describe these limitations:

Q4. Canadian guidelines (2005)	○ no ◉ yes

Q4.1 Have you used this system or guideline?	○ no ○ yes ○ unsure
Q4.2 Should this system or guideline be adopted as the standard for diagnosis in Australia?	○ no ○ yes ○ unsure ○ no comment
Q4.3 Are you aware of, or have you encountered, any limitations of this system or guideline?	○ no ◉ yes ○ unsure ○ no comment

Q4.3.1 Please describe these limitations:

Q5. Hoyme (an update based on Institute of Medicine) guidelines (2005)	○ no ◉ yes

Q5.1 Have you used this system or guideline?	○ no ○ yes ○ unsure
Q5.2 Should this system or guideline be adopted as the standard for diagnosis in Australia?	○ no ○ yes ○ unsure ○ no comment
Q5.3 Are you aware of, or have you encountered, any limitations of this system or guideline?	○ no ◉ yes ○ unsure ○ no comment

Q5.3.1 Please describe these limitations:

Q6. Do you use any other systems or guidelines for diagnosis?	○ no ◉ yes

Q6.1 Please describe this other system or guideline:

Q7. Please describe why you use, or don't use, the diagnostic systems or guidelines listed above, **or** select ☐ not my area of expertise

Enter any comments about the the adoption of existing FASD diagnostic systems or guidelines for use in Australia:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 11: Diagnostic Processes

*This section examines **general diagnostic processes** for Fetal Alcohol Spectrum Disorders in Australia.*

The Diagnostic Process for FASD

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. Exclusion of differential diagnoses is essential for the accurate diagnosis of Fetal Alcohol Spectrum Disorders	○	○	○	○	○	○
Q2. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to confirm the diagnosis of a FASD	○	○	○	○	○	○
Q3. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to exclude alternative diagnoses	○	○	○	○	○	○
Q4. With appropriate FASD-specific training, general practitioners can confirm the diagnosis of a FASD	○	○	○	○	○	○
Q5. With appropriate FASD-specific training, general practitioners can exclude alternative diagnoses	○	○	○	○	○	○
Q6. 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)	○	○	○	○	○	○

Q7. Should any other professionals confirm the diagnosis of a FASD and exclude alternative diagnoses? ○ **no** ◉ **yes** ○ **no comment**

Q7.1 Please specify the other professionals that should confirm the diagnosis of a FASD and exclude alternative diagnoses:

Enter any comments about who should confirm the diagnosis of a FASD and exclude alternative diagnoses:

Diagnostic Services

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q8. A multidisciplinary FASD assessment clinic should be available in major cities	○	○	○	○	○	○
Q9. Scheduled visits by FASD assessment teams to regional centres should be used to perform FASD screening and diagnosis	○	○	○	○	○	○
Q10. Scheduled visits by FASD assessment teams to regional centres should be used to support workforce training and development for FASD screening and diagnosis	○	○	○	○	○	○
Q11. Telehealth should be used by FASD assessment teams to support FASD screening and diagnosis	○	○	○	○	○	○

Enter any comments about the provision of FASD screening and diagnosis outside metropolitan regions:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 12: Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)

*A diagnosis of Fetal Alcohol Syndrome (FAS) is based on a combination of assessment findings. This section first examines **in general terms** the criteria required to diagnose FAS in Australia, and then how these criteria are defined.*

*If **all** of these questions are outside your expertise, please select ☐ not my area of expertise, and submit your responses at the bottom of the page.*

Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. 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	○	○	○	○	○	○
Q2. 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	○	○	○	○	○	○

Q3. Should any other combinations of diagnostic criteria be used to diagnose FAS? ○ **no** ◉ **yes** ○ **no comment**

Q3.1 Please specify the other combinations of diagnostic criteria that should be used:

Definition of Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)

Facial Anomalies

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Based on the general combination of criteria used to diagnose FAS above, FAS diagnostic criteria for facial anomalies should include:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q4. all 3 of the following characteristic FAS facial anomalies (short palpebral fissures, thin upper lip, smooth philtrum)	○	○	○	○	○	○
Q5. 2 or more of the following characteristic FAS facial anomalies (short palpebral fissures, thin upper lip, smooth philtrum)	○	○	○	○	○	○
Q6. 2 or more of the following characteristic FAS facial anomalies (short palpebral fissures, thin upper lip, smooth philtrum, flat midface)	○	○	○	○	○	○
Q7. 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)	○	○	○	○	○	○

Q8. Should any other diagnostic criteria be used for characteristic FAS facial anomalies? ○ **no** ◉ **yes** ○ **no comment**

Q8.1 Please specify the other diagnostic criteria that should be used:

Growth Deficit

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Based on the general combination of criteria used to diagnose FAS above, FAS diagnostic criteria for growth deficit should include:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q9. prenatal or postnatal growth deficit in height or weight at or below the 10th percentile	○	○	○	○	○	○
Q10. disproportionately low weight to height ratio at or below 10th percentile	○	○	○	○	○	○
Q11. disproportional low weight to height	○	○	○	○	○	○

Q12. low birth weight for gestational age	○	○	○	○	○	○
Q13. decelerating weight over time not due to nutrition	○	○	○	○	○	○

Q14. Should any other diagnostic criteria be used for growth deficit? ○ **no** ◉ **yes** ○ **no comment**

Q14.1 Please specify the other diagnostic criteria that should be used:

Central Nervous System (CNS) Abnormalities

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Based on the general combination of criteria used to diagnose FAS above, FAS diagnostic criteria for CNS abnormalities should include:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q15. at least 1 structural CNS abnormality (including decreased cranial size)	○	○	○	○	○	○
Q16. at least 1 of the following CNS abnormalities: structural (abnormal brain structure, including decreased cranial size), **or** neurological (hard or soft neurological signs)	○	○	○	○	○	○
Q17. 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)	○	○	○	○	○	○
Q18. 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						

Q19. Should any other diagnostic criteria be used for CNS abnormality? ○ **no** ◉ **yes** ○ **no comment**

Q19.1 Please specify the other diagnostic criteria that should be used:

Specific Definitions of Central Nervous System (CNS) Abnormalities

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

Based on the broad definitions of CNS abnormality for FAS above, decreased cranial size and functional performance abnormalities should be defined as:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q20. decreased cranial size at or below the 3rd percentile	○	○	○	○	○	○
Q21. decreased cranial size at or below the 10th percentile	○	○	○	○	○	○
Q22. global functional performance (cognitive or intellectual) below the 3rd percentile	○	○	○	○	○	○
Q23. performance for specific functional domains below the 3rd percentile	○	○	○	○	○	○
Q24. performance for specific functional domains below the 16th percentile	○	○	○	○	○	○
Q25. clinical judgement of functional impairment or deficit in domains where standardised measurements are not available	○	○	○	○	○	○
Q26. clinical judgement of functional impairment or deficit based on clinical assessment	○	○	○	○	○	○

Q27. Should any other definitions of abnormality be used for CNS findings? ○ **no** ◉ **yes** ○ **no comment**

Q27.1 Please specify the other definitions that should be used:

Enter any comments about the diagnostic criteria for FAS:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 13: Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders

*This section examines in general terms the criteria required to diagnose Fetal Alcohol Spectrum Disorders **other than Fetal Alcohol Syndrome** (FAS) in Australia.*

*If **all** of these questions are outside your expertise, please select ☐ not my area of expertise, and submit your responses at the bottom of the page.*

Part1. Diagnostic Criteria for Partial Fetal Alcohol syndrome (PFAS)

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

A diagnosis of Partial Fetal Alcohol Syndrome (PFAS) in the absence of FAS requires:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. 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)	○	○	○	○	○	○
Q2. 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)	○	○	○	○	○	○

Q3. Should any other criteria be used to diagnose Partial FAS? ○ **no** ◉ **yes** ○ **no comment**

Q3.1 Please specify the other combinations of diagnostic criteria that should be used:

Part 2. Diagnostic Criteria for Alcohol-related CNS Abnormalities in the absence of FAS i.e. Alcohol-related Neurodevelopmental Disorders (ARND) or Static Encephalopathy (alcohol exposed)

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

A diagnosis of Alcohol-related CNS Abnormalities in the absence of FAS i.e. Alcohol-related Neurodevelopmental Disorders (ARND) or Static Encephalopathy (alcohol exposed) requires:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q4. 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)	○	○	○	○	○	○
Q5. confirmed prenatal alcohol exposure, **and** evidence of decreased cranial size or abnormal brain structure	○	○	○	○	○	○
Q6. 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)	○	○	○	○	○	○

Q7. Should any other criteria be used to diagnose alcohol-related CNS abnormalities in the absence of FAS? ○ **no** ◉ **yes** ○ **no comment**

Q7.1 Please specify the other diagnostic criteria that should be used:

Part 3. Diagnostic Criteria for Alcohol-related Birth Defects in the absence of FAS

*Please indicate your level of agreement with the following statements, **or** select 'no comment' if outside your area of expertise.*

A diagnosis of Alcohol-related Birth Defects (ARBD) requires:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q8. 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)	○	○	○	○	○	○

Q9. Should any other criteria be used to diagnose alcohol-related birth defects? ○ **no** ◉ **yes** ○ **no comment**

Q9.1 Please specify the other diagnostic criteria that should be used:

	Strongly Agree	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q10. Alcohol-related birth defects is not sufficiently well defined to be a useful diagnostic category	○	○	○	○	○	○

Enter any comments about the diagnostic criteria for FASD other than FAS:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

Save Submit

Section 14: Acknowledgement and Feedback

Individual questionnaire responses will remain confidential and will not be linked to individual participant identities in any publications or reports. However, we would like to acknowledge the contribution of participants in publications arising from this study. Please indicate below if you wish to be acknowledged by name for your contribution.

Do you wish to be acknowledged by name as an expert participant in:	no	yes
Q1. the final project report	○	○
Q2. online project media	○	○

Enter any comments or feedback about this study:

To save your responses for later review click 'Save'.
*If you have finished this section and **answered all questions** please click 'Submit'.*

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FASD Questionnaire Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Demographic Information	Completed
2. Screening Programs	Completed
3. Targeted Screening	Completed
4. Screening Providers	Completed
5. Screening Methods Part 1: Prenatal Alcohol Exposure	Completed
6. Screening Methods Part 2: Growth Deficit, Facial Anomalies and Birth Defects	Completed
7. Screening Methods Part 3: Central Nervous System Abnormalities	Completed
8. Definition of Abnormal Screening Findings	Completed
9. Criteria for Conducting a Full Diagnostic Evaluation for FASD	Completed
10. Diagnostic Systems and Guidelines	Completed
11. Diagnostic Processes	Completed
12. Diagnostic Criteria for Fetal Alcohol Syndrome (FAS)	Completed
13. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders (FASD)	Completed
14. Acknowledgement and Feedback	Completed

Progress: 100% of sections complete. You have now finished the questionnaire.

Thank you very much for completing this questionnaire - we are extremely grateful for your contribution.

All participants will receive a summary of the group questionnaire results in the second questionnaire round.

This questionnaire was designed based on a systematic review of the international literature on FASD.
A list of the references reviewed can be found here.

Please log out when you are ready.

APPENDIX F2 ROUND 2 QUESTIONNAIRE

Welcome to the FASD Screening and Diagnosis Expert Questionnaire: Round 2

The purpose of this follow-up questionnaire is to provide feedback on the results of the first questionnaire, and seek your responses to a smaller number of questions on which group agreement has not been reached.

We are extremely grateful for your valuable contribution to the development of a screening and diagnostic instrument for FASD in Australia.

Instructions

- after you login you will be taken to the questionnaire contents page
    - each section of the questionnaire can be accessed from the contents page
- at the end of each section there are *'save'* and *'submit'* buttons:
    - clicking **'save'** indicates that you have not finished the section
    - clicking **'submit' finalises** your response and the section is considered complete
- this questionnaire will take around 20-25 minutes to complete, and does not need to be completed in a single sitting
    - **links at the top of each page** allow you to return to the contents page or log out at any time
    - a **progress bar** at the bottom of the contents page shows how close you are to completion of the questionnaire
- we do not expect all participants to be able to provide expert input in all areas of the questionnaire
    - the **'no comment'** option can be used to indicate that the question is outside your area of expertise
- also, the questions may not always reflect your beliefs about how FASD screening and diagnosis should be conducted in Australia
    - if so, **please let us know in the comment boxes provided**
- if you encounter any difficulties, or forget your password, please contact the project manager Heather Jones

Confidentiality statement

This project has been approved by the Human Research Ethics Committee of The University of Western Australia.
Study participants' identifying information is stored securely and separately from questionnaire responses to protect individual confidentiality.
See the full Telethon Institute for Child Health Research privacy statement here.

Login

Please login using your personal username and password supplied in your invitation email:

Username:

Password:

Login Reset

FASD Questionnaire Round 2 Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Screening Programs	Completed
2. Screening Methods: Assessment of Prenatal Alcohol Exposure	Completed
3. Definition of Abnormal Screening Findings	Completed
4. Criteria for Conducting a Full Diagnostic Evaluation	Completed
5. Diagnostic Criteria for FAS and PFAS	Completed
6. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders	Review
7. Existing Diagnostic Guidelines and Diagnostic Processes in Australia	Review
8. Acknowledgement and Feedback	Review

Progress: 63% of sections completed

Section 1: Screening Programs

Instructions:

This study aims to develop an instrument that can be used to improve the identification and or diagnosis of FASD in Australia.

We are using this Delphi process to identify an agreed starting point for instrument development.

Any tools developed on the basis of these findings will need further evaluation to establish their effectiveness in Australia.

Please consider the feedback and results on screening for Fetal Alcohol Spectrum Disorders in Australia presented in the boxes below, and complete questions Q1-Q13.

Summary of Findings - Screening

There was a high level of agreement ('strongly agree' or 'agree') that a broad range of health professionals could screen for FASD, and that all health professionals who screen for FASD require appropriate FASD-specific training.

Comments indicated support for universal screening strategies as an ideal and ethical approach, and support for targeted screening strategies as a more feasible and cost-effective approach. Comments also frequently

highlighted the importance of practical considerations including the availability of intervention services.

There was highest agreement with targeted screening in the presence of:

- **'a parent or foster parent who is concerned that their child might have a FASD'** *(99% agreement)*
- **'characteristic FAS facial anomalies'** *(97% agreement)*
- **'an alcohol-related event, illness or dependency in the birth mother'** *(96% agreement)*

Consistent with comments received, further research is required to evaluate the effectiveness of various targeted screening strategies.

Comments also frequently identified the need to define screening and clearly distinguish between screening and diagnostic assessments.

Some participants indicated a lack of support for the feasibility and effectiveness of screening solely for FASD. The need for health professionals to be more aware of the possibility of a FASD was thought by some participants to be more important and appropriate than conducting stand alone screening for FASD.

Enter any further comments about screening for FASD:

Screening at Birth

There was over 96% agreement that screening for FASD at birth should include the following 5 components:

- **prenatal alcohol exposure**

- **growth (birthweight, length and head circumference)**
- **characteristic FAS facial anomalies**
- **birth defects**
- **evidence of withdrawal from alcohol**

Participants also frequently suggested two additional components of screening at birth:

- **family history of FASD or developmental delay**
- **evidence of CNS dysfunction, including irritability, feeding difficulties, or other neurological signs**

Comments indicated that a stand-alone clinical assessment procedure for FASD screening at birth is not required, as all of the identified components of screening (perhaps apart from prenatal alcohol exposure) are routinely assessed at birth.

A screening checklist, using this routinely collected information and prenatal alcohol exposure, could be developed to screen for FASD at birth and identify which infants require a full diagnostic evaluation.

Previous responses indicated:

- **58% agreement that 'screening for FASD at birth should be universal'** *(you said 'Strongly Agree')*
- **69% agreement that 'screening for FASD at birth should be targeted'** *(you said 'Agree')*

Instructions:

Please take into account the information above and complete questions Q1-Q7 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

Screening for FASD at or around birth should <u>also</u> assess and record:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q1. family history of FASD or developmental delay	○	○	○	○	○	○
Q2. evidence of CNS dysfunction including irritability, feeding difficulties or other neurological signs	○	○	○	○	○	○

	Strongly Agree	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q3. Most of the information required for FASD screening at birth is routinely collected at birth	○	○	○	○	○	○
Q4. 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	○	○	○	○	○	○
Q5. 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	○	○	○	○	○	○
Q6. Screening for FASD at birth should be universal	○	○	○	○	○	○
Q7. Screening for FASD at birth should be targeted	○	○	○	○	○	○

Enter any further comments about screening for FASD at birth:

Screening in Childhood

There was over 90% agreement that screening for FASD in childhood should include the following components:

- **prenatal alcohol exposure**
- **growth (height and weight, prenatal and postnatal)**
- **characteristic FAS facial anomalies**
- **head circumference**
- **developmental delay**
- **neurological signs**
- **functional CNS abnormalities (e.g. cognition, behaviour, learning, sensory perception etc.)**
- **hearing and vision**
- **birth defects**

Participants also frequently suggested an additional component of screening in childhood:

- **family history of FASD, developmental delay, abuse or neglect**

Comments indicated that many of the agreed components of FASD screening in childhood are routinely conducted as part of a general clinical assessment of children with neurodevelopmental and other presentations.

Previous responses indicated:

- **48% agreement that 'screening for FASD in childhood should be universal'** *(you said 'Agree')*
- **79% agreement that 'screening for FASD in childhood should be targeted'** *(you said 'Strongly Agree')*

Instructions:

Please take into account the information above and complete questions Q8-Q13 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

Screening for FASD in childhood should <u>also</u> assess and record:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q8. family history of FASD, developmental delay, abuse or neglect	○	○	○	○	○	○

	Strongly Agree	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q9. 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	○	○	○	○	○	○
Q10. 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, etc.)	○	○	○	○	○	○
Q11. 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	○	○	○	○	○	○
Q12. Screening for FASD in childhood should be universal	○	○	○	○	○	○
Q13. Screening for FASD in childhood should be targeted	○	○	○	○	○	○

Enter any further comments about screening for FASD in childhood:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 2: Screening Methods: Assessment of Prenatal Alcohol Exposure

Instructions:

Please consider the feedback and results on the assessment of prenatal alcohol exposure presented in the boxes below, and complete questions Q1-Q4.

There was over 95% agreement with the 4 required components of prenatal alcohol exposure assessment:

- **the number of standard drinks consumed on a typical drinking occasion**
- **the frequency of drinking occasions**
- **the frequency of excessive drinking (5+ drinks)**
- **the timing of alcohol intake during pregnancy**

In addition:

- **93% agreed that alcohol use should be assessed alongside other lifestyle factors** *(you said 'no response')*
- **71% agreed that prenatal alcohol exposure should be assessed using a formal tool** *(you said 'no response')*
- **52% agreed that prenatal alcohol exposure can be effectively assessed using an informal approach** *(you said 'no response')*

Comments indicated that additional details about alcohol consumption should be assessed via informal

methods during the clinical interview; that general enquiry alongside lifestyle histories can obtain a detailed frequency, quantity and timing of alcohol use better than a tool; and that seeking additional information and using alternative assessment methods may be particularly important among high-risk groups.

Some participants also indicated the need to ensure that formal assessment tools are culturally appropriate.

While prenatal screening is not the focus of this study, participant comments emphasised the importance of universal prenatal assessment and documentation of prenatal alcohol exposure to enable intervention to decrease maternal intake and to follow-up prenatally exposed children.

Information:

Based on these findings, a suitable formal assessment tool for prenatal alcohol exposure may include the AUDIT-C.

The AUDIT-C is a 3-item tool that could be used to assess prenatal alcohol exposure at different times during pregnancy, and includes the above endorsed components of prenatal alcohol exposure assessment. The tool has a simple scoring method with predetermined response options.

The AUDIT-C questions are:

1. **How often do you have a drink containing alcohol?**
   (never, monthly or less, 2-4 times a month, 2-3 times a week, 4 or more times a week)
2. **How many drinks containing alcohol do you have on a typical day when you are drinking?**
   (1 or 2, 3 or 4, 5 or 6, 7 to 9, 10 or more)
3. **How often do you have six or more drinks on one occasion?**
   (never, less than monthly, monthly, weekly, daily or almost daily)

Instructions:

Please take into account the information above and complete questions Q1-Q4 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. 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	○	○	○	○	○	○
Q2. Prenatal alcohol exposure can be effectively assessed during a consultation using an informal approach	○	○	○	○	○	○
Q3. Information on alcohol use from family members, other health professionals or community members (if appropriate) should be sought if indicated	○	○	○	○	○	○
Q4. The AUDIT-C would be a useful tool for the formal assessment of prenatal alcohol exposure	○	○	○	○	○	○

Enter any further comments about the assessment of prenatal alcohol exposure:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 3: Definition of Abnormal Screening Findings

Instructions:

Please consider the feedback and results on the definition of abnormal screening findings presented in the boxes below, and complete questions Q1-Q8.

Functional Central Nervous System (CNS) Abnormalities

There was over 80% agreement that CNS assessment in FASD screening may include the following functional domains:

- **developmental milestones, motor and sensory function, cognition, memory, academic achievement, executive functioning and abstract reasoning, adaptive behaviour, attention and hyperactivity, communication (receptive and expressive language), social skills and social communication.**

Results indicated that formal assessment methods were favoured to define potential functional CNS abnormalities at the screening stage:

- **89% agreement with evidence of functional impairment on standard psychometric testing, with performance 2 or more standard deviations below the mean** *(you said 'no response')*
- **62% agreement with clinical judgement of functional impairment or deficit based on clinical observation and assessment** *(you said 'no response')*

However, comments indicated major concerns with the formal assessment of these functional CNS domains in

screening. Some participants indicated that screening must be simple and quick, with extensive clinical assessment only appropriate as part of the diagnostic assessment process.

Characteristic FAS Facial Anomalies

Similarly, the need to measure facial features was thought by some to be time consuming and not necessary during the screening stage.

However, responses also indicated that formal assessment methods should be used during screening:

- **69% agreement with the use of clinical observation to assess characteristic facial anomalies (smooth philtrum, thin upper lip, and small palpebral fissures)** *(you said 'no response')*
- **76% agreement with the use of physical measurement of palpebral fissures** *(you said 'no response')*
- **86% agreement with the use of the University of Washington Lip-Philtrum Guide** *(you said 'no response')*
- **73% agreement with the use of the facial photographic screening tool** *(you said 'no response')*

Formal assessment methods were also favoured to define characteristic FAS facial anomalies at the screening stage:

- **96% agreement with a positive finding of thin upper lip and smooth philtrum based on the University of Washington Lip-Philtrum Guide (rank 4 or 5)** *(you said 'no response')*
- **59% agreement with a positive finding of thin upper lip and smooth philtrum based on visual assessment and clinical impression** *(you said 'no response')*
- **68% agreement with fissure length at or below the 3rd percentile based on comparison with population references** *(you said 'no response')*

- **53% agreement with fissure length at or below the 10th percentile based on comparison with population references** *(you said 'no response')*
- **55% agreement with visual assessment/clinical impression of short palpebral fissures** *(you said 'no response')*

Comments indicated that the assessment of fissure length is diagnostic and not part of screening; that measurements by inexperienced assessors are unreliable; that there is a lack of appropriate normative data for comparison; and that assessment in older children can have negative psychosocial consequences.

Instructions:

Due to the lack of clear consensus on the most appropriate method to assess CNS abnormalities and characteristic FAS facial anomalies in FASD screening, please complete questions Q1-Q8 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

At the screening stage it is not necessary to formally assess and measure:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q1. suspected CNS abnormalities	○	○	○	○	○	○
Q2. suspected facial anomalies	○	○	○	○	○	○

At the screening stage, the following are acceptable indicators of possible CNS abnormalities (neurological, functional or structural):	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q3. clinical identification	○	○	○	○	○	○

Q4. parent or other credible third party report	○	○	○	○	○	○
Q5. results of previous relevant formal assessments (e.g. psychological report)	○	○	○	○	○	○

At the screening stage:	**Strongly Agree**	**Agree**	**Neither**	**Disagree**	**Strongly Disagree**	**No Comment**
Q6. 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	○	○	○	○	○	○
Q7. palpebral fissure length must be assessed using formal physical measurement and comparison with population references at the screening stage	○	○	○	○	○	○
Q8. 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	○	○	○	○	○	○

Enter any further comments about the definition of abnormal screening findings:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 4: Criteria for Conducting a Full Diagnostic Evaluation

Instructions:

Please consider the feedback and results on the criteria for conducting a full diagnostic evaluation presented in the boxes below, and complete questions Q1-Q3.

There was over 70% agreement with the following minimum criteria for conducting a full diagnostic evaluation:

BOX 1. CRITERIA FOR CONDUCTING A FULL DIAGNOSTIC EVALUATION

I. **concern by a parent or foster parent that their child might have a FASD** *(89% agreement, you said 'Strongly Agree')*

II. **2 or more of the characteristic FAS facial anomalies (smooth philtrum, thin vermillion border, and small palpebral fissures), or evidence of the characteristic pattern of FAS facial anomalies** *(77% agreement, you said 'Strongly Agree' & 72% agreement, you said 'Strongly Agree' respectively)*

III. **1 of the characteristic FAS facial anomalies, a growth deficit and any CNS abnormality (structural, neurological or functional)** *(85% agreement, you said 'Strongly Agree')*

IV. **known or probable prenatal alcohol exposure, 1 of the characteristic FAS facial anomalies, and a growth deficit or any CNS abnormality** *(93% agreement, you said 'Strongly Agree')*

V. **known or probable prenatal alcohol exposure and 1 or more CNS abnormalities** *(88% agreement, you said 'Strongly Agree')*

VI. **known or probable prenatal alcohol exposure and 1 or more birth defects** *(88% agreement, you said*

'Strongly Agree')

Comments indicated support for the use of sensitive criteria in order to reduce the risk of undetected cases.

Instructions:

To identify whether there is general agreement with the criteria for a full evaluation as displayed in Box 1 above, please complete question Q1, and then continue to Q2-Q3 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. If any of the combinations of criteria listed in Box 1 above are met, a full diagnostic evaluation is required	○	○	○	○	○	○

Enter any further comments about the criteria for conducting a full diagnostic evaluation:

'Significant prenatal alcohol exposure' as a criterion for conducting a full diagnostic evaluation

In addition to the criteria identified in Box 1, there was general agreement that evidence of prenatal alcohol exposure <u>on its own</u> at levels iv-vi below indicates the need for a full diagnostic evaluation:

i. **< 7 standard drinks per week and < 3 standard drinks on any one day** *(38% agreement, you said 'Disagree')*

ii. **< 7 standard drinks per week and 3 - 4 standard drinks on any one day** *(62% agreement, you said 'Disagree')*

iii. **7+ standard drinks per week and < 3 standard drinks on any one day** *(60% agreement, you said 'Agree')*

iv. **7+ standard drinks per week and 3 - 4 standard drinks on any one day** *(82% agreement, you said 'Strongly Agree')*

v. **5+ standard drinks per occasion less than once per week** *(79% agreement, you said 'Strongly Agree')*

vi. **5+ standard drinks per occasion once or twice per week** *(84% agreement, you said 'Strongly Agree')*

However, there was 46% agreement that 'no level of prenatal alcohol exposure is alone sufficient to indicate the need for a full diagnostic evaluation for FASD'. *(you said 'Strongly Agree')*

Reasons provided for a lack of support for significant prenatal alcohol exposure alone as a criterion for full assessment included that there are insufficient resources, and that the child's difficulties are the key.

Reasons provided in support of lower levels of prenatal alcohol exposure alone as a criterion for full assessment included that any exposure may be significant and should always lead to further assessment and monitoring, and that information about the exposure may not be accurate.

Comments also indicated that it is difficult to define the relevant level of exposure; that it is impossible to get this level of information in all settings; and that its absence should not preclude the need to consider FASD. Significant clinical suspicion of heavy prenatal alcohol exposure (e.g. a history of alcohol-related illness) was also considered by some as sufficient to warrant a full diagnostic evaluation.

Instructions:

To identify whether there is general agreement with the inclusion of significant prenatal alcohol exposure on its own as a criteria for full evaluation, and evaluate agreement with the general definition of significant prenatal alcohol exposure, please complete questions Q2-Q3 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q2. Evidence of significant prenatal alcohol exposure **on its own** is sufficient to require a full diagnostic evaluation	○	○	○	○	○	○
Q3. 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	○	○	○	○	○	○

Enter any further comments about prenatal alcohol exposure as a criteria for conducting a full diagnostic evaluation:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 5: Diagnostic Criteria for Fetal Alcohol Syndrome (FAS) and Partial FAS (PFAS)

Instructions:

Please consider the feedback and results on diagnostic criteria presented in the boxes below, and complete questions Q1-Q3.

Previous responses indicated agreement with the following specific definitions of the subcomponents of the FAS diagnostic criteria:

- **74% agreement with the need for all 3 characteristic facial anomalies**
- **80% agreement with prenatal or postnatal growth deficit in height or weight at or below the 10th percentile, and**
- **76% agreement with 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).

Responses also indicated agreement with the following specific definitions of CNS abnormalities:

- **88% agreement with decreased cranial size at or below the 3rd percentile,**
  79% agreement with global functional performance (cognitive or intellectual) below the 3rd percentile,

and
82% agreement with performance for specific functional domains below the 3rd percentile.

Previous responses indicated a similar level of agreement with the following two definitions for PFAS:

- **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) *(70% agreement, you said 'Strongly Agree')*
- **confirmed prenatal alcohol exposure, and**
  2 of the 3 characteristic FAS facial anomalies, 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) *(75% agreement, you said 'Strongly Agree')***.**

There was only 62% agreement with the general criteria for a FAS diagnosis as requiring all four of the following:

- **confirmed or unknown prenatal alcohol exposure**
  characteristic FAS facial anomalies
  growth deficit
  CNS abnormalities *(you said 'Strongly Agree')*

Instructions:

To clarify whether there is general agreement on the broad diagnostic criteria for FAS and PFAS, please complete questions Q1-Q3 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. A diagnosis of FAS requires 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	○	○	○	○	○	○

A diagnosis of Partial Fetal Alcohol Syndrome (PFAS) in the absence of FAS requires:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q2. 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, impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement)	○	○	○	○	○	○
Q3. 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)						

Enter any further comments about diagnostic criteria for FAS and PFAS:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 6: Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders

Instructions:

Please consider the feedback and results on the diagnostic criteria for other FASDs presented in the boxes below, and complete questions Q1-Q5.

Alcohol-related Neurodevelopmental Disorders (ARND) or Static Encephalopathy (alcohol exposed)

Previous responses indicated a similar level of agreement with two definitions of Alcohol-related Neurodevelopmental Disorders (ARND) or Static Encephalopathy (alcohol exposed) in the absence of FAS:

- **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) *(72% agreement, you said 'Strongly Agree')*
- **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) *(67% agreement, you said 'Strongly Agree')*

Diagnostic Criteria for Alcohol-related Birth Defects (ARBD)

Previous responses indicated 69% agreement with the definition of Alcohol-related Birth Defects in the absence of FAS:

- **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)** *(you said 'Strongly Agree')*

Responses also indicated:

- **56% agreement that alcohol-related birth defects is not sufficiently well defined to be a useful diagnostic category** *(you said 'Strongly Agree')*

Participant comments about the diagnostic criteria for ARBD included that it should take into account the level of alcohol exposure; that it is required and may become more concrete over time; that the diagnostic category cannot be medically confirmed; that it is not included in recent diagnostic guidelines for FASD; that it needs to be clinically useful; that the terminology is an inadequate reflection of the multifactorial and often unknown aetiology of birth defects; and that the labelling of complex problems can be dangerous and counterproductive.

Instructions:

To clarify whether there is general agreement on the diagnostic criteria for FASDs other than FAS, please complete questions Q1-Q5 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your

area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q1. 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	○	○	○	○	○	○

A diagnosis of Alcohol-related Birth Defects (ARBD) in the absence of FAS requires:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q2. confirmed prenatal alcohol exposure, **and** identification of alcohol-related birth defects on clinical examination (including cardiac, musculo-skeletal, renal, ocular, auditory or other malformations, including facial anomalies)	○	○	○	○	○	○
Q3. confirmed **significant** prenatal alcohol exposure, **and** identification of alcohol-related birth defects on clinical examination (including cardiac, musculo-skeletal, renal, ocular, auditory or other malformations, including facial anomalies)	○	○	○	○	○	○

A diagnosis of Alcohol-related CNS Abnormalities in the absence of FAS i.e. Alcohol-related Neurodevelopmental Disorders (ARND) or Static Encephalopathy (alcohol exposed) requires:	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment
Q4. 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, impulse control, social skills, receptive and expressive language, abstract reasoning, attention, memory or judgement, motor and sensory abnormalities)	○	○	○	○	○	○
Q5. 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)	○	○	○	○	○	○

Enter any further comments about the criteria for diagnosis of FASDs other than FAS:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 7: Existing Diagnostic Guidelines and Diagnostic Processes in Australia

Instructions:

Please consider the feedback and results about existing diagnostic guidelines and diagnostic processes in Australia presented in the boxes below, and complete questions Q1-Q4.

Existing Diagnostic Guidelines

Previous responses indicated:

- **51% were familiar with the University of Washington 4-Digit Diagnostic Code, and among these, 56% used the guidelines and 33% recommended they be adopted in Australia**
- **35% were familiar with the Canadian guidelines, and among these, 53% used the guidelines and 31% recommended they be adopted in Australia**
- **31% were familiar with the Centers for Disease Control and Prevention (CDC) guidelines, and among these, 43% used the guidelines and 9% recommended they be adopted in Australia**

Comments about the adoption of existing guidelines in Australia included that a standard national approach is required to understand the problem; that education and training for health professionals who are going to use the guidelines is essential; that the development of Australian guidelines that are appropriate for use in rural and remote Australia is required; that guidelines need to be valid, specific and sensitive; and that there must be good access to diagnostic, intervention and support services.

Enter any further comments about the adoption of existing guidelines for FASD screening and diagnosis in Australia:

Diagnostic Processes in Australia

Previous responses indicated:

- **90% agreement that a multidisciplinary FASD assessment clinic should be available in major cities**
- **77% agreement that scheduled visits by FASD assessment teams to regional centres should be used to perform FASD screening and diagnosis**
- **92% agreement that scheduled visits by FASD assessment teams to regional centres should be used to support workforce training and development for FASD screening and diagnosis**
- **80% agreement that telehealth should be used by FASD assessment teams to support FASD screening and diagnosis**

Comments indicated support for building local expertise in diagnosis and supporting existing services through the provision of ongoing training, screening and diagnostic tools, and assistance in complex cases. The involvement of multidisciplinary teams was recognised as best practice; however, screening was not considered to require the involvement of specialist assessment teams. An outreach service model was considered to provide little ongoing patient support and intervention.

Previous responses indicated:

- **74% agreement that evaluation by a general or subspecialist paediatrician or clinical geneticist is required to confirm the diagnosis of a FASD** *(you said 'no response')*

- **83% agreement that evaluation by a general or subspecialist paediatrician or clinical geneticist is required to exclude alternative diagnoses** *(you said 'no response')*
- **47% agreement that with appropriate FASD-specific training, general practitioners can confirm the diagnosis of a FASD** *(you said 'no response')*
- **30% agreement that with appropriate FASD-specific training, general practitioners can exclude alternative diagnoses** *(you said 'no response')*
- **84% agreement that 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)** *(you said 'no response')*

However, comments identified that resource constraints limit the provision of diagnostic services, particularly outside metropolitan areas where multidisciplinary teams are not available or practical. Several participants suggested that in rural areas general practitioners may have to be used to diagnose FAS; and that interest, education, training and experience can increase the capacity of non-specialists to confirm and exclude FASD in the absence of an ideal level of resources.

Instructions:

To clarify whether there is agreement on diagnostic processes for FASD, please complete questions Q1-Q4 below.

Please indicate your level of agreement with the following statements, or select 'no comment' if outside your area of expertise.

	Strongly Agree	Agree	Neither	Disagree	Strongly Disagree	No Comment

Q1. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to confirm the diagnosis of a FASD	○	○	○	○	○	○
Q2. Evaluation by a general or subspecialist paediatrician or clinical geneticist is required to exclude alternative diagnoses	○	○	○	○	○	○
Q3. With appropriate FASD-specific training, general practitioners in rural and remote settings can confirm the diagnosis of a FASD	○	○	○	○	○	○
Q4. With appropriate FASD-specific training, general practitioners in rural and remote settings can exclude alternative diagnoses	○	○	○	○	○	○

Enter any further comments about diagnostic processes in Australia:

*If you have finished this section and **answered all questions** please click 'Submit'. Click 'Save' if you have not finished.*

Save Submit

Section 8: Acknowledgement and Feedback

Thank you again for your valuable contribution to this project. We will notify all participants when the final report is available.

Individual questionnaire responses will remain confidential and will not be linked to individual participant identities in any publications or reports.

Please enter any comments or feedback about this study:

Participants have indicated the following preferences with respect to acknowledgement by name for their contribution to this project:

- **56% wish to be acknowledged in the final report** *(you said ' no response')*
- **49% wish to be acknowledged in online project media** *(you said ' no response')*

If you wish to change your preferences about acknowledgement, please update your responses at questions Q1-Q2 below:

Do you wish to be acknowledged by name as an expert participant in:	no	yes
Q1. the final project report	○	○
Q2. online project media	○	○

If you have finished this section please click 'Submit'. Click 'Save' if you have not finished.

Save Submit

FASD Questionnaire Round 2 Contents Page

Please complete all sections of the questionnaire below. Sections can be accessed through the links under 'Status'.

Section	Status
1. Screening Programs	Completed
2. Screening Methods: Assessment of Prenatal Alcohol Exposure	Completed
3. Definition of Abnormal Screening Findings	Completed
4. Criteria for Conducting a Full Diagnostic Evaluation	Completed
5. Diagnostic Criteria for FAS and PFAS	Completed
6. Diagnostic Criteria for Other Fetal Alcohol Spectrum Disorders	Completed
7. Existing Diagnostic Guidelines and Diagnostic Processes in Australia	Completed
8. Acknowledgement and Feedback	Completed

Progress: 100% of sections complete. You have now finished the questionnaire.

Thank you very much for completing this round 2 questionnaire - we are extremely grateful for your contribution.

This questionnaire was designed based on a systematic review of the international literature on FASD. A list of the references reviewed can be found here.

Please log out when you are ready.