Section E: Formulating a diagnosis

Information collected during the diagnostic assessment should be reviewed, ideally in a multi-disciplinary team context, to evaluate the strength of evidence to:

- Support a diagnosis of FASD with 3 sentinel facial features or a diagnosis of FASD with <3 sentinel facial features (Refer Table 1); or
- Consider whether the individual is at risk of FASD, requiring reassessment and/or further investigation; or
- Exclude other causes or conditions; and/or
- Assess the potential influence of other exposures and events.

> The yellow shaded sections on the FASD Diagnostic Assessment Form (Appendix A1) and the Summary Form (Appendix A2) summarise the clinical findings required to make a diagnosis of FASD.

Section F: Discussing the diagnosis and developing a management plan

After completing the diagnostic assessment, irrespective of the diagnosis, it is recommended that the health professional/s coordinating the diagnostic process:

- Discuss with individual/parents/caregivers the outcome of the medical assessment and any reports from other health professionals involved in the assessment.
- Discuss the diagnosis, as applicable, and develop a Management Plan, incorporating parent/caregiver and patient goals, referrals, management strategies and review dates (Appendix A3).
- Provide the individual/parents/caregivers with a written report.
- Discuss how this information may be important to share with relevant service providers and school staff. Parents/caregivers will need to provide consent for any reports to be sent directly to others; however the parent/caregiver may take their copy of the reports to the school or other organisations, to develop an appropriate plan and access services, for example through the education system or the National Disability Insurance Scheme.
- Provide contact details for follow-up communication with the clinic, if required.
- If FASD has been diagnosed, provided written information about FASD and provide contact details for NOFASD Australia - National Organisation for Fetal Alcohol Spectrum Disorder (NOFASD) Australia http://www.nofasd.org/ or phone 1300 306 238.

- For information and resources for individuals/parents /caregivers after a diagnostic assessment, including formulation of the management plan and referrals to therapy and other support services: see Appendix A7.
- Consider the need for referral for individuals or family members with alcohol use disorders, as appropriate.
- For information and resources for clinicians to support patients and their families after a diagnostic assessment: see Appendix A8.

Section G: Reporting a FASD diagnosis

Please note that FASD is a notifiable birth defect in some states (South Australia, Western Australia).

Western Australian Register of Developmental Anomalies

http://www.kemh.health.wa.gov.au/services/register_developmental_anomalies/

South Australian Birth Defects Registry

http://www.wch.sa.gov.au/services/az/other/phru/birthdefect.html

AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT FORM

PATIENT DETAILS

NAME	
Sex	☐ Female ☐ Male ☐ Other
Date of birth (DD/MM/YYYY)	/ / Age at assessment:
Racial/ ethnic background	
Preferred language	
Hospital number (if applicable)	
Referral source, date, provider number and contact details	
Name of person(s) accompanying patient	
Relationship (s) to the patient	
Patient's primary carer (select 1 or more)	☐ Birth mother ☐ Birth father ☐ Foster carer ☐ Adoptive parent/s ☐ Other
Birth mother's name	
Birth father's name	
Patient in care of	☐ Department of Child Protection ☐ Juvenile justice ☐ Not applicable
Consent form for assessment completed	☐ No ☐ Yes
Assessment Form completed by	
Place of assessment	
Completion of this form (DD/MM/YYYY)	/ /

History

Presenting concerns:
(Include concerns identified by referring doctor, parent, caregiver, teacher; strengths and needs; age-appropriate abilities e.g. behavioural regulation, memory and learning, social skills and motor control)

Obstetric history:

Developmental history:

Mental health and other behavioural problems:

Patient's medical history:

Social history: e.g. foster care, living arrangements.

MATERNAL ALCOHOL USE

Evidence of maternal alcohol use in the three months prior to and during pregnancy should be assessed, including any special occasions when a large amount of alcohol may have been consumed. The definition of a standard drink should be explained prior to administering the AUDIT-C (Q1-3). A Standard Drinks Guide can be downloaded.

http://www.health.gov.au/internet/alcohol/publishing.nsf/Content/drinksguide-cnt

Alcohol use in early pregnancy (if available)

a.	Was the pregnancy planned or unplanned? ☐ Planned ☐ Unplanned ☐ Unknown	
b.	At what gestation did the birth mother realise that she was pregnant? _____ (weeks) ☐ Unknown	
c.	Did the birth mother drink alcohol before the pregnancy was confirmed? ☐ Yes ☐ No ☐ Unknown	
d.	Did the birth mother modify her drinking behaviour on confirmation of pregnancy? ☐ Yes ☐ No ☐ Unknown If Yes please specify:	
e.	During which trimesters was alcohol consumed? (tick one or more) ☐ None ☐ 1st ☐ 2nd ☐ 3rd ☐ Unknown	

AUDIT-C Reported alcohol use (if available)

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

Unknown	Never [skip Q2+Q3]	Monthly or less	2-4 times a month	2-3 times a week	4 or more times a week
☐	☐ $_0$	☐ $_1$	☐ $_2$	☐ $_3$	☐ $_4$

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

Unknown	1 or 2	3 or 4	5 or 6	7 to 9	10 or more
☐	☐ $_0$	☐ $_1$	☐ $_2$	☐ $_3$	☐ $_4$

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

Unknown	Never	Less than monthly	Monthly	Weekly	Daily or almost daily
☐	☐ $_0$	☐ $_1$	☐ $_2$	☐ $_3$	☐ $_4$

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

Scores: 0=No exposure 1-4= Confirmed exposure   5+= Confirmed high-risk exposure

Other evidence of exposure

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

☐ No   ☐ Yes (identify below, including source of information)

☐ Alcohol dependency (specify)

☐ Alcohol-related illness or hospitalisation (specify)

☐ Alcohol-related injury (specify)

☐ Alcohol-related offence (specify)

☐ Other (specify)

Information from records: e.g. medical records, court reports, child protection records.

Is there evidence that the birth mother's partner has ever had a problem associated with alcohol misuse or dependency?

☐ No   ☐ Yes (identify below, including source of information)

Alcohol exposure summary

Source of reported information on alcohol use: ☐ Birth mother   ☐ Other (specify)

In your judgement what is the reliability of the information on alcohol exposure: ☐ Unknown ☐ Low ☐ High

In your judgement was there high-risk consumption of alcohol during pregnancy? ☐ Unknown ☐ Yes ☐ No

Prenatal alcohol exposure: ☐ Unknown exposure ☐ No exposure ☐ Confirmed exposure ☐ Confirmed-high risk exposure

OTHER EXPOSURES

Assess evidence of adverse prenatal and postnatal exposures and events that need to be considered.

Prenatal

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

☐ Nicotine (e.g. cigarettes, inhalers, e-cigs and chewed tobacco) (specify)

☐ Marijuana (specify)

☐ Heroin (specify)

☐ Cocaine (specify)

☐ Amphetamines (specify)

☐ Other non-prescription drugs (specify)

☐ Anti-convulsants (specify)

☐ Other prescription drugs (specify)

☐ Don't know

☐ None

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

Other prenatal risk summary:

☐ No known risk ☐ Unknown risk ☐ Some risk ☐ High risk

Postnatal

Specify other physical or medical risk factors and assess risk based on your clinical judgement: (e.g. prematurity, history of abuse or neglect, serious head injury, meningitis or other medical conditions that lead to brain damage, child substance abuse)

Specify other psychosocial risk factors and assess risk (e.g. emotional abuse, early life trauma, parental separation or incarceration, drug and alcohol use in the household; overcrowding, socio-economic disadvantage):

Postnatal risk summary:

☐ No known risk ☐ Unknown risk ☐ Some risk ☐ High risk

GROWTH

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

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

Growth reference chart used: ☐ CDC   ☐ WHO   ☐ Other (specify)

Postnatal		Height		Weight	
Date	Age	cm	percentile	kg	percentile

Growth reference chart used: ☐ CDC   ☐ WHO   ☐ Other (specify)

Parental height (if available)

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

Specify factors that may explain growth parameters: (e.g. nutritional or environmental neglect, genetic condition, prematurity, other drugs, nicotine)

Growth summary

Was an unexplained deficit in height or weight < 3rd percentile identified at any time? ☐ Yes   ☐ No
_____
If Yes ☐ height or weight ≤10th and >3rd percentile   ☐ height or weight ≤3rd percentile

SENTINEL FACIAL FEATURES

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

Palpebral Fissure Length (PFL)

			Right PFL		Left PFL		Mean PFL	
Date	Age	Assessment method	mm	Z score (SD)	mm	Z score	mm	Z score*
		☐ direct measure　☐ photo analysis						
		☐ direct measure　☐ photo analysis						
PFL reference chart used:　☐ Stromland　☐ Clarren　☐ Other								

Philtrum

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

Upper lip

Date	Age	Assessment method	UW Lip-Philtrum Guide 5-point rank
		☐ direct measure　☐ photo analysis	
		☐ direct measure　☐ photo analysis	
		☐ direct measure　☐ photo analysis	
Lip-Philtrum Guide[†] used:　☐ Guide 1. Caucasian　☐ Guide 2. African American			

Sentinel Facial Features Summary

Number of Sentinel Facial Features (PFL 2 SD or more below the mean, philtrum rank 4 or 5, upper lip rank 4 or 5):
☐ 0　　☐ 1　　☐ 2　　☐ 3

OTHER PHYSICAL FINDINGS

Dysmorphic facial features (please specify)

Other birth defects - major or minor (please specify)

Other medical conditions:

Hearing impairment:　☐ No　☐ Not tested　☐ Yes (specify)

Vision impairment:　☐ No　☐ Not tested　☐ Yes (specify)

Known syndrome or genetic disorder (please specify):

Other (please specify):

Investigations:

Chromosomal microarray:　☐ No　☐ Result pending　☐ Yes (specify result)

Fragile X testing:　☐ No　☐ Result pending　☐ Yes (specific result)

Other investigations as indicated: Full blood count, ferritin, metabolic screen, creatinine kinase, lead, and thyroid function (Specify):

*University of Washington Palpebral Fissure Length Z-score calculator: http://depts.washington.edu/fasdpn/htmls/diagnostic-tools.htm#pfl

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

NEURODEVELOPMENTAL DOMAINS

1 BRAIN STRUCTURE / NEUROLOGY DOMAIN

BRAIN STRUCTURE

Occipitofrontal Circumference (OFC)

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

*correct for gestational age when < 2 years old

If OFC < 3rd percentile, is it explained by other aetiologies e.g. infection, metabolic or other disease?

No   ☐ Yes (specify)

Imaging

CNS imaging performed: ☐ No   ☐ Yes (specify image modality and date)

Specify any structural abnormalities:

If yes, are they explained by other aetiologies e.g. injury, infection, or metabolic or other disease? ☐ No   ☐ Yes (specify)

NEUROLOGY

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

Seizure disorder

Seizure disorder present: ☐ No   ☐ Yes (specify)

If yes, are they explained by other aetiologies e.g. injury, infection, or metabolic or other disease? ☐ No   ☐ Yes (specify)

Other neurological diagnoses e.g. cerebral palsy, visual impairment, sensorineural hearing loss

Other abnormal neurological diagnoses present: ☐ No   ☐ Yes (specify)

If yes, are they explained by other aetiologies e.g. injury, infection, or metabolic or other disease? ☐ No   ☐ Yes (specify)

Brain Structure/ Neurology domain summary

Evidence of brain structure/neurology abnormalities of presumed prenatal origin that are unexplained by other causes?
    ☐ No   ☐ Yes   ☐ Not assessed

FUNCTIONAL NEURODEVELOPMENTAL DOMAIN SUMMARIES

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

2. MOTOR SKILLS

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation
Other information:				
Motor Skills impairment: ☐ None	☐ Some	☐ Severe	☐ Not assessed	

3. COGNITION

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation
Other information:				
Cognition impairment: ☐ None	☐ Some	☐ Severe	☐ Not assessed	

4. LANGUAGE

(Expressive and Receptive)

Test/subtest name	Age/Date	Score	%ile/SD	Interpretation
Other information:				
Language impairment ☐ None	☐ Some	☐ Severe	☐ Not assessed	

5. ACADEMIC ACHIEVEMENT

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation

Other information:

Academic achievement impairment	☐ None	☐ Some	☐ Severe	☐ Not assessed

6. MEMORY

Test/subtest name	Age /Date	Score	%ile/SD	Interpretation

Other information:

Memory impairment	☐ None	☐ Some	☐ Severe	☐ Not assessed

7. ATTENTION

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation

Other information:

Attention impairment	☐ None	☐ Some	☐ Severe	☐ Not assessed

8. EXECUTIVE FUNCTION, INCLUDING IMPULSE CONTROL AND HYPERACTIVITY

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation

Other information:

Executive function, including impulse control and hyperactivity impairment

☐ None ☐ Some ☐ Severe ☐ Not assessed

9. AFFECT REGULATION

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation

Other information:

Affect regulation impairment: ☐ None ☐ Some ☐ Severe ☐ Not assessed

10. ADAPTIVE BEHAVIOUR, SOCIAL SKILLS, OR SOCIAL COMMUNICATION

Test/subtest name	Age/ Date	Score	%ile/SD	Interpretation

Other information:

Adaptive behaviour, social skills, or social communication impairment

☐ None ☐ Some ☐ Severe ☐ Not assessed

NEURODEVELOPMENTAL DOMAINS SUMMARY

Number of neurodevelopmental domains with evidence of severe impairment:

☐ None ☐ 1 ☐ 2 ☐ 3 or more (specify)_____

DIAGNOSIS:

For derivation of the Australian FASD diagnostic categories, please refer to the Australian FASD Diagnostic Criteria and FASD Diagnostic Pathway Algorithm below (also see Table 1 and Figure 1 in the Guide). Record the diagnosis below.

Indicate as applicable:

11 | P a g e

☐   FASD with 3 sentinel facial features

☐   FASD with < 3 sentinel facial features

☐   At risk of FASD

☐   Incomplete assessment e.g. further investigation/information needed

☐   Other diagnoses (with or without FASD)

Clinical notes:

AUSTRALIAN FASD DIAGNOSTIC ASSESSMENT SUMMARY FORM

PATIENT DETAILS

NAME	
Sex	☐ Female ☐ Male ☐ Other
Date of birth (DD/MM/YYYY)	/ / Age at assessment:
Racial/ethnic background	
Hospital number (if applicable)	

ALCOHOL EXPOSURE SUMMARY

Source of reported information on alcohol use: ☐ Birth mother ☐ Other (specify)

In your judgement what is the reliability of the information on alcohol exposure: ☐ Unknown ☐ Low ☐ High

In your judgement was there high-risk consumption of alcohol during pregnancy? ☐ Unknown ☐ Yes ☐ No

Prenatal alcohol exposure: ☐ Unknown exposure ☐ No exposure ☐ Confirmed exposure ☐ Confirmed-high risk exposure

SENTINEL FACIAL FEATURES SUMMARY

Number of Sentinel Facial Features (PFL 2 SD or more below the mean, philtrum rank 4 or 5, upper lip rank 4 or 5):

☐ 0   ☐ 1   ☐ 2   ☐ 3

NEURODEVELOPMENTAL DOMAINS SUMMARY

Neurodevelopmental Domain	Impairment			
1 Brain structure/Neurology	☐ No	☐ Yes	☐ Not assessed	
2 Motor Skills	☐ None	☐ Some	☐ Severe	☐ Not assessed
3 Cognition	☐ None	☐ Some	☐ Severe	☐ Not assessed
4 Language	☐ None	☐ Some	☐ Severe	☐ Not assessed
5 Academic achievement	☐ None	☐ Some	☐ Severe	☐ Not assessed
6 Memory impairment	☐ None	☐ Some	☐ Severe	☐ Not assessed
7 Attention	☐ None	☐ Some	☐ Severe	☐ Not assessed
8 Executive function, including impulse control and hyperactivity	☐ None	☐ Some	☐ Severe	☐ Not assessed
9 Affect regulation	☐ None	☐ Some	☐ Severe	☐ Not assessed
10 Adaptive behavior, Social Skills, or Social Communication	☐ None	☐ Some	☐ Severe	☐ Not assessed

Number of neurodevelopmental domains with evidence of severe impairment

☐ None ☐ 1   ☐ 2   ☐ 3 or more (specify) _____

Other Prenatal or Post-natal risk/exposure

Other prenatal risk summary:	☐ No known risk	☐ Unknown risk	☐ Some risk	☐ High risk
Postnatal risk summary:	☐ No known risk	☐ Unknown risk	☐ Some risk	☐ High risk

Growth summary

Was an unexplained deficit in height or weight < 3rd percentile identified at any time? ☐ Yes ☐ No

For derivation of the Australian FASD diagnostic categories, please refer to the Australian FASD Criteria and the FASD Diagnostic Pathway Algorithm below (also see Table 1 and Figure 1 in the Guide).   Record the diagnosis below.

Indicate as applicable:

☐   FASD with 3 sentinel facial features
☐   FASD with < 3 sentinel facial features
☐   At risk of FASD
☐   Incomplete assessment e.g. further investigation/information needed
☐   Other diagnoses (with or without FASD)

Clinical notes: