Australian Paediatric Surveillance Unit
PROTOCOL SHEET- FETAL ALCOHOL SYNDROME

Background

Fetal alcohol syndrome (FAS) was first identified in the 1970's and has been described as a preventable tragedy[1,2,3,4]. FAS is caused by maternal alcohol consumption during pregnancy and represents the severe end of the spectrum of the effects of exposure to alcohol *in utero*. Children with FAS display a wide range of physical defects and disabilities, however the cardinal features are:

- *Minor cranio-facial abnormalities*
- *Prenatal and/or postnatal growth deficiency*
- *Evidence of damage to the central nervous system.*

Most epidemiological studies on FAS have been performed in the USA and Canada where prevalence estimates range from 0.3-7.2 per 1000 live births[5,6]. The National Perinatal Statistics Unit which collects information on congenital malformations in Australia, does not include FAS. The Australian prevalence is unknown and there is currently no mechanism for national data collection. Under-recognition of FAS may occur due to the complexity of the disorder leading to difficulty in diagnosis, the frequent delay between exposure and symptom manifestation and the difficulty in confirming alcohol exposure.

Objectives

1. To increase clinicians' awareness of FAS by provision of information on clinical features and diagnostic criteria
2. To estimate the birth prevalence of FAS in Australian children seen by paediatricians
3. To estimate the incidence of FAS (new cases identified) in children <15 years over the study period
4. To determine the sex, age at diagnosis, clinical features and co-morbidity in FAS
5. To attempt to establish, retrospectively, alcohol intake during pregnancy in mothers of affected children
6. To describe the epidemiology of FAS in terms of geography, socio-economic status and ethnicity
7. To document use of other harmful substances eg. drugs, by mothers of children with FAS
8. To describe use of health services by children with FAS
9. To use information provided by paediatricians to categorise the severity of alcohol-related abnormalities

CASE DEFINTION AND REPORTING INSTRUCTIONS

Any child under the age of 15 years seen in the previous month with newly diagnosed fetal alcohol syndrome, suspected fetal alcohol syndrome or partial fetal alcohol syndrome (as defined below)

Fetal Alcohol Syndrome - Alcohol exposure confirmed
Evidence of prenatal alcohol exposure in association with:
All characteristic cranio-facial abnormalities ***and*** prenatal and/or postnatal growth deficiency ***and*** structural abnormalities or dysfunction of the central nervous system.

Suspected Fetal Alcohol Syndrome – Alcohol exposure not confirmed
No confirmed evidence of prenatal alcohol exposure but presence of:
All characteristic cranio-facial abnormalities ***and*** prenatal and/or postnatal growth deficiency ***and*** structural abnormalities or dysfunction of the central nervous system.

Partial Fetal Alcohol Syndrome – Alcohol exposure confirmed
Evidence of prenatal alcohol exposure in association with:
All characteristic cranio-facial abnormalities ***and*** structural abnormalities or dysfunction of the central nervous system

or

Some characteristic cranio-facial abnormalities ***and*** prenatal and/or postnatal growth deficiency ***and*** structural abnormalities or dysfunction of the central nervous system

The clinical features associated with FAS are listed below:

Cranio-facial abnormalities	Prenatal and/or postnatal growth deficiency	Central nervous system manifestations
Characteristic features Short palpebral fissures Long smooth philtrum Thin upper lip Other features High arched eyebrows Ptosis Flat midface Short, upturned nose	Birth weight < 3rd centile for gestational age Post natal growth deficiency Height and weight <3rd centile **or** Weight <3rd & height < 10th centile **or** Weight <10th & height <3rd centile	Microcephaly (head circumference < the 3rd centile for age) Structural brain abnormality e.g. cerebellar hypoplasia Persistent neurological signs eg. impaired fine or gross motor function poor coordination abnormal muscle tone sensorineural hearing loss Cognitive impairment IQ <60 Other (not included as part of the definition) speech or language delay Behavioural problems e.g. ADHD Emotional problems

Note that a large number of **other congenital defects** including defects of the eye, heart and kidney have been associated with alcohol exposure in pregnancy but their frequency is not known.

The study investigators will be using a FAS diagnostic guide to objectively record the severity of the features of FAS present in each case reported. This includes measurement of the palpebral fissure length and the degree of lip thinness and philtrum smoothness. Enclosed is the **5-point pictorial Lip-philtrum guide** to help objectively quantify lip thinness and philtrum smoothness. During assessment, the lips should be gently closed with no smile. Please grade the appearance according to the Lip-philtrum guide. The maximum **palpebral fissure length** (from the outer to the inner canthus) should be measured with the child looking upwards while the head is held level.

An overview of the FAS diagnostic guide is available on http://www.depts.washington.edu/fasdpn

Investigators

Dr Carol Bower	Principal Research Fellow, TVW Telethon Institute for Child Health Research, Perth, WA
Dr Elizabeth Elliott	Consultant Paediatrician, Children's Hospital at Westmead, NSW; A/Prof, Department of Paediatrics and Child Health, University of Sydney
Dr Eric Haan	ClinicalGeneticist, Women's and Children Hospital, Adelaide, SA
Dr Ingrid Bucens	Consultant Paediatrician, Royal Darwin Hospital, Darwin, NT
Dr Alison Leversha	Community Paediatrician, Westkids, Auckland, NZ
Dr Anne Morris	Paediatric Fellow, Children's Hospital at Westmead, NSW
Dr Greta Ridley	Scientific Officer, Australian Paediatric Surveillance Unit

Any questions should be directed to Dr Anne Morris, APSU (02) 98452200
Email **annem3@chw.edu.au**

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