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Impacts of alcohol use in pregnancy

The role of the GP

BACKGROUND

Fetal alcohol syndrome (FAS) is a preventable cause of developmental delay and growth failure.

OBJECTIVE

This article discusses the clinical features of fetal alcohol spectrum disorders (FASD) and the role of the general practitioner in prevention and management.

DISCUSSION

Early diagnosis of and intervention for problems associated with FAS reduce adverse long term outcomes. Most health professionals have limited knowledge of FASD and lack confidence in the diagnosis and management of children with FASD. General practitioners have an important role in identifying women and children at risk of harm from alcohol and arranging referral for assessment and management when necessary. Educational materials for health professionals are currently under development.

Alcohol is teratogenic and its ingestion during pregnancy can result in miscarriage, stillbirth, prematurity, birth defects, and problems of growth and development. Fetal alcohol spectrum disorder (FASD) is an umbrella term used to encompass some of the conditions that result from fetal exposure to alcohol including fetal alcohol syndrome (FAS), alcohol related birth defects (ARBD), and alcohol related neurodevelopmental disorder (ARND). These conditions, which are associated with lifelong problems, are entirely preventable.¹

Fetal alcohol spectrum disorders

Fetal alcohol syndrome

Children with FAS have characteristic facial features (*Figure 1*), pre- and/or post-natal growth failure, and structural and/or functional abnormalities of the central nervous system (*Table 1*). Children with FAS may have a range of other birth defects, difficulties with learning and memory, poor executive functioning, and behavioural and emotional problems. Fetal alcohol syndrome is a lifelong condition and children with FAS are at high risk of secondary disabilities in adulthood, partly as a result of educational failure. These include drug and alcohol abuse, mental health problems, inappropriate sexual behaviour,

unemployment, and contact with the law.² Fetal alcohol syndrome costs the USA \$3.6 billion per annum.³

Few data on FAS are available in Australia, where the reported birth prevalence ranges from 0.06–0.68 per 1000 live births overall.⁴ Rates are consistently higher in subgroups of indigenous children: 2.76/1000 live births in Western Australia,⁵ and 4.7/1000 live births in the Northern Territory.⁴ In the USA, FAS has a birth prevalence of 0.5–2.0/1000 live births overall, but 9.8/1000 live births in native Americans. Rates in Europe are similar to those in Australia, however reported rates in some communities (eg. Cape Province in South Africa) are much higher (46/1000 live births).⁶ It is likely that the low rate of FAS in Australia is due to under diagnosis and one reason for this may be lack of knowledge about FAS and under-recognition of the disorder by health professionals.

Alcohol related birth defects

A number of major and minor birth defects have been associated with alcohol exposure, and these may be diagnosed in children with confirmed prenatal alcohol exposure but without a diagnosis of FAS (*Table 2*). There are no data on the prevalence of ARBD in Australia.

Alcohol related neurodevelopmental disorder

Children with confirmed exposure to alcohol during pregnancy but without a diagnosis of FAS may have a range of cognitive, emotional and behavioural problems (Table 3). There are no data on the prevalence of ARND.

Alcohol use in pregnancy

Just under half of all pregnancies in Australia are unplanned.⁷ Considering current and increasing rates of risky and binge drinking in women,⁸ and the relationship between alcohol use, unprotected sex, and unplanned pregnancy, it is likely that unintentional fetal alcohol exposure occurs frequently in early pregnancy. A Western Australian study showed that 80% of women

consumed alcohol in the 3 month period before becoming pregnant and 58% drank in at least one trimester during pregnancy. In the first trimester, 14% of women surveyed drank at levels outside the National Health and Medical Research Council (NHMRC) guidelines for pregnancy.⁷ The highest risk to the fetus is during the first trimester, but the fetus is at risk of harm from alcohol exposure throughout pregnancy.⁹

Why should GPs ask about alcohol use in pregnancy?

General practitioners are ideally placed to ask women of childbearing age about alcohol in pregnancy, and then to assess and advise

about its effects. However, in a survey of 1143 WA health professionals, including 170 GPs, only 67% of GPs said they routinely asked about alcohol use in pregnancy; 24% asked sometimes; and 30% asked if there were maternal risk factors including smoking and drug use. Few GPs (31%) said they routinely gave information on the consequences of maternal alcohol use for the fetus, and 17% said they did not provide such information at all.¹⁰

Making a diagnosis of FAS

Diagnosis of FAS relies on history and physical examination (Table 1) and exclusion of other diagnoses. However, the diagnosis of FAS is challenging for a number of reasons. There is no definitive test for FAS; it is a diagnosis of exclusion; and there is overlap between the features of FAS and some other syndromes. After puberty, the characteristic facial features may become more difficult to distinguish (Figure 1). Currently, there are at least three different diagnostic criteria in use which – although similar – have points of conflict.^{1,11,12} Another barrier to diagnosis is that many children with FAS are in alternative care (eg. foster care, kinship care) and details of the mother’s antenatal history and findings from the infant’s early examinations are frequently unknown. Also, it may be difficult to accurately confirm prenatal alcohol exposure retrospectively.

Nevertheless, FASD should be considered if: a child has poor pre- and/or post-natal growth, or any of the neurological dysfunctions or facial characteristics or other features listed in Table 1–3, and information on prenatal alcohol intake should be sought.

Diagnosis is best made by a multidisciplinary team to assess the child in medical, developmental, and psychological domains. Ideally, such a team consists of a developmental paediatrician, psychologist, speech pathologist, and occupational therapist. Other team members might include a geneticist, child psychiatrist, drug and alcohol worker and cultural liaison worker. No specific FAS diagnostic clinics exist in Australia. However, developmental paediatricians and child development services are appropriate services for referral of children with suspected FASD.

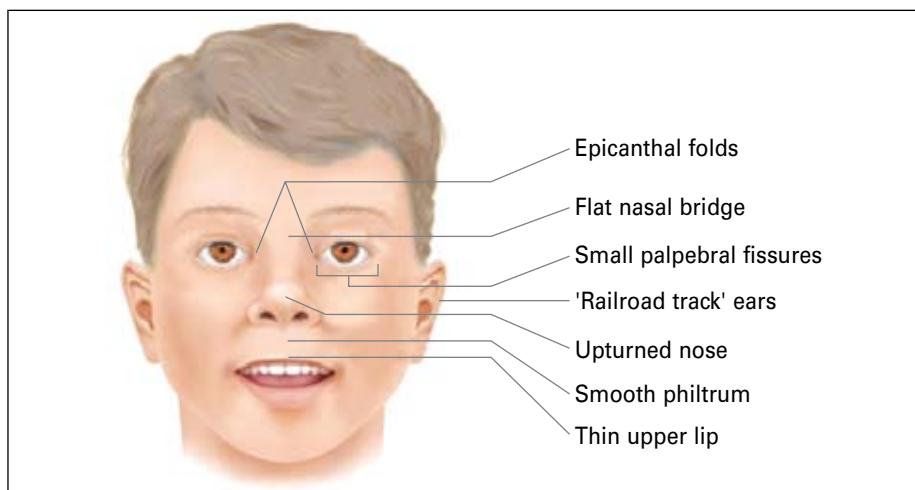


Figure 1. Facial features of fetal alcohol syndrome

Table 1. Diagnostic features of FAS¹¹

Alcohol exposure during pregnancy

Confirmed or unknown

Growth deficiency

Pre- or post-natal height and/or weight \leq 10th percentile

Key facial features

- smooth philtrum
- thin vermilion border (ie. upper lip)
- small palpebral fissures (\leq 10th percentile according to age and racial norms)

Neurological dysfunction/damage

Structural

- head circumference \leq 10th percentile
- structural brain abnormality on imaging

Neurological

- neurological problems not due to a postnatal event

Functional

- performance below the individual’s age

GPs knowledge about and experience with FAS

Few GPs feel confident about diagnosing or managing FAS and only 20% of GPs surveyed in WA correctly identified the four essential diagnostic features. Although only 14% of these GPs had diagnosed a child with FAS, 35% of GPs had on occasion suspected that a child had FAS but had not made the diagnosis. Over half of the GPs (56%) were concerned that making the diagnosis would stigmatise the child and their family. Most GPs surveyed requested

Table 2. Alcohol related birth defects¹

Confirmed maternal alcohol exposure **and** at least two of the facial features in Table 1 **and** one or more of the following structural defects:

Cardiac

- atrial septal defects
- ventricular septal defects
- aberrant great vessels
- conotruncal heart defects

Skeletal

- pectus carinatum/excavatum
- scoliosis radioulnar synostosis
- vertebral segmentation defects
- large joint contractures

Renal

- aplastic/hypoplastic/dysplastic kidneys
- 'horseshoe' kidneys
- ureteral duplications

Eyes

- strabismus
- ptosis
- refractive errors
- retinal vascular anomalies
- optic nerve hypoplasia

Ears

- conductive hearing loss
- sensorineural hearing loss
- 'railroad track' ears

Minor anomalies

- hypoplastic nails
- short fifth digits
- clinodactyly of fifth fingers
- camptodactyly
- 'hockey stick' palmar creases
- epicanthal folds
- low nasal bridge
- midface hypoplasia

Table 3. Alcohol related neurodevelopmental disorder¹

Confirmed maternal alcohol exposure **and** at least one of the following:

Structural

- head circumference \leq 10th percentile
- structural brain abnormality on imaging

Behavioural or cognitive abnormalities inconsistent with developmental level that cannot be explained by genetic predisposition, family background or environment alone

Impairment in performance of complex tasks

- complex problem solving
- planning
- judgment
- abstraction
- arithmetic

Higher level receptive and expressive language deficits**Disordered behaviour**

- difficulties in personal manner
- emotional lability
- motor dysfunction
- poor academic performance
- deficient social interaction

education materials for themselves (75%) and their patients (69%), including diagnostic checklists and referral resources to assist them in managing FAS.¹⁰

Why diagnose FAS?

Fetal alcohol syndrome is likely underdiagnosed and under-reported in Australia, and health professional surveys^{10,13} identified a number of barriers to making the diagnosis. Health professionals do not routinely ask about alcohol use in pregnancy, have poor knowledge about FAS, have concerns about stigmatising the child and family, feel ill equipped to manage FAS, believe some carers would refuse referral, and identify a lack of both services and effective interventions for children.

However, there are compelling reasons for making the diagnosis. For the child, accurate diagnosis informs and enables early intervention which leads to a 2–4 times reduction in the risk of secondary disabilities in adulthood.² For the family, diagnosis is important. It provides an explanation for the child's problems, leads to better understanding and more appropriate expectations of the child, and may improve access to educational and social services and government allowances. Making the diagnosis

of FAS, ARBD or ARND also provides an opportunity for prevention of damage to future children. At a public health level, diagnosis can improve reporting of incidence and prevalence and thus allows research, advocacy and planning for health, educational and social services. Potential negatives of making the diagnosis such as maternal guilt and blame always need to be addressed sensitively as part of assessment and management.

Interventions for FASD

Although, there are no specific diagnostic services in Australia for FASD, existing paediatric and child development services can provide an assessment and coordinate care. There are few published studies evaluating specific interventions for children with FASD, although several randomised controlled trials of cognitive behavioural and family therapy are under way in North America. Thus, recommendations for therapy should be based on the specific problems identified during the child's assessment. Interventions required may include physiotherapy, speech and occupational therapy, remedial education, behaviour management, family therapy and support, psychological therapy, specialised paediatric therapy, hearing

Table 4. NHMRC Australian alcohol guideline 11¹⁴**Women who are pregnant or might soon become pregnant**

11.1 may consider not drinking at all

11.2 most importantly, should never become intoxicated

11.3 if they choose to drink – over a week, should have less than 7 standard drinks, **and** on any one day, no more than 2 standard drinks (spread over at least 2 hours)

11.4 should note that the risk is highest in the earlier stages of pregnancy, including the time from conception to the first missed period

and vision services, and pharmacotherapy for specific problems. Referral to drug and alcohol services may be required for the mother.

Alcohol policy for pregnancy

It is acknowledged that maternal use of large amounts of alcohol in early pregnancy puts the fetus at most risk. However, there is a lack of consensus about the risk to the fetus associated with low to moderate levels of maternal alcohol consumption in both early and late pregnancy. Differences in interpretation of the available evidence are reflected in policies developed by governments and professional bodies. The NHMRC Australian alcohol guideline 11 currently advises that: 'Women who are pregnant or might soon become pregnant may choose not to consume alcohol during pregnancy, and if they do drink should not become intoxicated and should drink at low risk levels'¹⁴ (Table 4).

This guideline is currently under review. It has variable support, with few government agencies or professional bodies providing advice consistent with the guideline.¹⁵ Most government,^{16–20} professional,^{21–23} and nongovernment^{24,25} bodies in Australia either provide an abstinence message or recommend abstinence as the safest choice. The variable support for guideline 11 and the inconsistency of dissemination of policy across Australia potentially creates confusion and may lessen the impact of the message. In the WA survey only 17% GPs gave advice entirely consistent with NHMRC guideline 11. However, 88% recommended that women consider not drinking at all and 33% recommended to women not to become intoxicated during pregnancy.¹⁰

The trend in policy in other English speaking countries is toward abstinence,^{26–30} although

some policies recognise that risk is dose related. In the United Kingdom women are advised that occasional, small amounts of alcohol (around 1–2 units per week) pose a low risk to the fetus.^{27,28,30} It is argued that an abstinence message has the potential to create undue fear and/or guilt which may lead women to seek termination despite a low level of risk to the fetus.^{31,32} While the Canadian authorities promote an abstinence message, they also advise that women who have used small amounts of alcohol should be reassured that the risk of harm to the fetus is minimal and relative to the amount consumed.³³

The role of the GP

General practitioners have an important role in the diagnosis and prevention of FASD. They should ask about and document alcohol use in pregnancy; assess the level of risk of women's alcohol consumption; advise women of childbearing age, including pregnant women, that no alcohol is the safest choice if pregnant or trying to get pregnant; reassure women who have consumed small amounts of alcohol before they knew they were pregnant that the risk of harm to the fetus is low; assist women to stop or reduce alcohol consumption; and arrange for referral for women to appropriate services when needed. General practitioners should carefully examine any exposed child at birth and refer children with problems to a paediatric service for assessment.

Resources

Carers wanting more information can be referred to the National Organisation of Fetal Alcohol Syndrome and Related Disorders (www.nofasard.org). Information for health professionals is available at www.rhef.com.au/. Education materials for health professionals are currently being developed by the Telethon Institute for Child Health Research in Western Australia.

Summary of important points

- Alcohol use in pregnancy may cause birth defects, growth failure, developmental delay and learning difficulties.
- Early diagnosis of and intervention for problems associated with FAS reduce adverse long term outcomes.
- Health professionals have an important role in the prevention of FASD
- Health professionals should provide information to women about the potential risks to the fetus from alcohol consumption during pregnancy.

Conflict of interest: none declared.

References

1. Hoyme KE, May PA, Kalberg WO, et al. A practical clinical approach to diagnosis of Fetal Alcohol Spectrum Disorders: clarification of the 1996 Institute of Medicine criteria. *Pediatrics* 2005;115:39–47.
2. Streissguth AP, Bookstein FL, Barr HM, Sampson PD, O'Malley K, Young JK. Risk factors for adverse life outcomes in fetal alcohol syndrome and fetal alcohol effects. *J Dev Behav Pediatr* 2004;25:228–38.
3. Lupton C, Burd L, Harwood R. Cost of fetal alcohol spectrum disorders. *Am J Med Genet C Semin Med Genet* 2004;127:42–50.
4. Harris KR, Bucens IK. Prevalence of fetal alcohol syndrome in the top end of the Northern Territory. *J Paediatr Child Health* 2003;39:528–33.
5. Bower C, Silva D, Henderson TR, Ryan A, Rudy E. Ascertainment of birth defects: the effect on completeness of adding a new source of data. *J Paediatr Child Health* 2000;36:574–6.
6. Elliott EJ, Bower C. FAS in Australia: fact or fiction? *J Paediatr Child Health* 2004;40:8–10.
7. Colvin L, Payne J, Parsons D, Kurinczuk JJ, Bower C. Alcohol consumption during pregnancy in nonindigenous west Australian women. *Alcohol Clin Exp Res* 2007;31:276–84.
8. Australian Bureau of Statistics. Alcohol consumption in Australia: a snapshot, 2004–05. Canberra: Australian Government, 2006.
9. Coles CD. Critical periods for prenatal alcohol exposure: evidence from animal and human studies. *Alcohol Health Res World* 1994;18:22–9.
10. Payne J, Elliott E, D'Antoine H, et al. Health professionals' knowledge, practice and opinions about fetal alcohol syndrome and alcohol consumption in pregnancy. *Aust N Z J Public Health* 2005;29:558–64.
11. Bertrand J, Floyd LL, Weber MK. Guidelines for identifying and referring persons with fetal alcohol syndrome. *MMWR Recomm Rep* 2005;54:1–14.
12. Astley SJ. Comparison of the 4-digit diagnostic code and the Hoyme diagnostic guidelines for fetal alcohol spectrum disorders. *Pediatrics* 2006;118:1532–45.
13. Elliott EJ, Payne J, Haan E, Bower C. Diagnosis of foetal alcohol syndrome and alcohol use in pregnancy: a survey of paediatricians' knowledge, attitudes and practice. *J Paediatr Child Health* 2006;42:698–703.
14. National Health and Medical Research Council. Australian Alcohol Guidelines: health risks and benefits. Canberra:

- National Health and Medical Research Council, 2001.
15. Commonwealth of Australia. National clinical guidelines for the management of drug use during pregnancy, birth and the early developmental years of the newborn. Sydney: Commonwealth Department of Health and Ageing, 2006.
 16. New South Wales Health. Women and alcohol. Available at www.alcoholinfo.nsw.gov.au/family_wellbeing/pregnancy [Accessed December 11 2006].
 17. Northern Territory Department of Health and Community Services. Alcohol and other drugs. In: Northern Territory Department of Health and Community Services, editor. The Public Health Bush Book. Darwin: Northern Territory Government, 2006.
 18. South Australian Department of Health. Alcohol can affect your unborn child. Available at www.health.sa.gov.au/PREGNANCY/DesktopDefault.aspx?tabid=187 [Accessed December 12 2006].
 19. Victorian Government Health Information. Alcohol and young people. Available at www.health.vic.gov.au/drugs/alcohol/youth/faq.htm [Accessed December 12 2006].
 20. Queensland Health. Strategic policy for Aboriginal and Torres Strait Islander children and young people's health 2005–2010. Brisbane: Strategic Policy Branch, Queensland Health, 2005.
 21. Harris M, Bailey L, Bridges-Webb C, et al. Guidelines for preventive activities in general practice. Melbourne: The Royal Australian College of General Practitioners, 2005.
 22. Australian Medical Association. AMA highlights dangers of alcohol during pregnancy. Available at www.ama.com.au/web.nsf/doc/WEEN-6FSVM4 [Accessed December 12 2006].
 23. The Royal Australasian College of Physicians. Alcohol policy: using evidence for better outcomes. Sydney: The Royal Australasian College of Physicians, 2005.
 24. National Organisation for Fetal Alcohol Syndrome and Related Disorders. Alcohol and pregnancy brochure. Available at www.nofasard.org/ [Accessed December 12 2006].
 25. Australian Drug Foundation. Alcohol, other drugs and pregnancy. Available at www.adf.org.au/store/search.asp?keywords=Alcohol%2C+Other+Drugs+and+Pregnancy+&SearchType=Keywords&Format= [Accessed December 13 2006].
 26. New Zealand Ministry of Health. A national drug policy for New Zealand 1998–2003. Wellington: New Zealand Ministry of Health, 1998.
 27. Department of Health England. The pregnancy book. London: National Health System, Department of Health, 2001.
 28. Health Scotland. Alcohol: safe in pregnancy? Available at www.hebs.scot.nhs.uk/readysteadybaby/pregnancy/health.htm [Accessed December 13 2006].
 29. United States Surgeon General. Advisory on alcohol use in pregnancy. Available at www.hhs.gov/surgeon-general/pressreleases/sg02222005.html [Accessed December 13 2006].
 30. The Royal College of Obstetricians and Gynaecologists. Alcohol consumption and the outcomes of pregnancy. London: The Royal College of Obstetricians and Gynaecologists, 2006.
 31. Australian National Council on Drugs. Fetal alcohol syndrome national workshop 2002. Canberra: Commonwealth Department of Health and Ageing, 2003.
 32. Koren G. Alcohol consumption in early pregnancy. How much will harm a fetus? *Can Fam Physician* 1996;42:2141–3.
 33. Health Canada. Nutrition for a healthy pregnancy – national guidelines for the childbearing years. Ottawa: Health Canada, 1999.

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Epidemiology of alcohol use. Alcohol consumption during pregnancy is common. The prevalence of alcohol consumption prior to and during pregnancy highlights the need to educate all reproductive age women about the potential harms of alcohol exposure on the developing fetus. As alcohol is a teratogen that impacts fetal growth and development at all stages of pregnancy, national guidelines and medical societies from multiple countries recommend complete abstinence during pregnancy [8-13]. Abstaining from alcohol while pregnant eliminates the risk of alcohol-related birth defects and developmental disabilities, including resultant neurocognitive and behavioral problems [9]. The effect of alcohol abuse even before pregnancy may cause harmful impact on the glucose levels of the offspring. The effects of alcohol use during pregnancy on an unborn child are well known, including possible birth defects and learning and behavior problems. However, it is not known whether a mother's alcohol use before conception also could have negative effects on her child's health and disease susceptibility during adulthood," said principal investigator Dipak Sarkar, Ph.D., DPhil, a distinguished professor at Rutgers University in New Brunswick, N.J., and directo