Fetal alcohol syndrome (FAS) and FAS spectrum deficits (FASD) are the most common and completely preventable forms of developmental disabilities and birth defects in Canada. The term FAS was first used in 1973 by Jones and Smith to describe a group of children born to alcoholic mothers, and included the following abnormalities:

- Growth retardation;
- Characteristic facial features; and
- Central nervous system involvement.¹

The estimated incidence of FAS in Canada is between 0.5 and three per 1,000 live births.² It is now recognized that prenatal alcohol exposure can cause a spectrum of deficits, with FAS being the most readily recognizable. Other conditions associ-
Fetal alcohol exposure have been referred to as fetal alcohol effects (FAE), partial FAS (PFAS), alcohol-related birth defects (ARBD) and alcohol-related neurodevelopmental disorder (ARND). All of these terms refer to situations where the patient has a history of alcohol exposure during gestation, and some, but not all, of the findings of FAS. The estimated incidence of conditions within the FASD category is 10 cases per 1,000 live births. Despite increased public awareness of FAS and its diagnosis during pregnancy increased from 12.4% to 16.3% between 1991 and 1995. Moreover, frequent drinking during pregnancy, defined as seven or more drinks per week, increased during the same time period from 0.8% to 3.5%. In the face of this increasing pattern of maternal alcohol consumption, it is imperative all women be screened for alcohol use. Appearance, culture or socioeconomic status cannot identify a pregnant woman who consumes alcohol. Several screening tools have been developed for use during pregnancy, including CAGE (Cut down, Annoyed by criticism, Guilty about drinking, Eye-opener drinks) and the T-ACE (Tolerance level for alcohol, Annoyed by criticism, Cut down, Eye-opener drinks). The T-ACE has advantages, including the ability to use it with a variety of cultural groups, and its higher sensitivity and specificity for assessment of peri-conceptual heavy drinking. The following are additional areas that might indicate the presence of a high-risk maternal drinking pattern:

- The presence of a heavy drinking partner — most women drink with their partners;
- Past history of sexual or physical abuse — a study of 80 birth mothers of children with FAS revealed 95% were physically or sexually abused during their lifetime;
- The presence of mental health disorders. In the

### Summary

**Fetal Alcohol Syndrome**

- The growth pattern characteristic of FAS likely presents in the prenatal period, and persists as a consistent impairment over time. By contrast, growth deficiency due to postnatal influences is likely to present as periodic fluctuations in the growth curve.
- Language deficits result from a global impairment in executive functioning, due to prenatal alcohol exposure. The understanding of complex language and figures of speech is markedly deficient.
- Reasoning and memory deficits are related to basic impairments in executive functioning of the brain and become more noticeable when the FAS-affected person reaches school age.
- Management in the primary-care setting begins with screening for the key manifestations of FAS and FASD disorders, and facilitating an early and accurate diagnosis by a multidisciplinary team that can address primary and secondary disabilities.
- FAS is 100% preventable through consistent application of primary prevention strategies, systematic screening of all pregnant women and timely provision of interventions.
- The facial features that are most closely associated with FAS are short palpebral fissures, a thin upper lip and a smooth philtrum. The facial features of FAS become less noticeable in adolescence and adulthood.
- Prenatal alcohol exposure causes global impairment in cognitive abilities related to executive functioning, and specific speech and language deficits. The understanding of complex language and figures of speech is markedly deficient.
same study of 80 birth mothers, 96% had one to
10 mental health disorders, the most prevalent
being post traumatic stress disorder (77%) and
phobia (44%);7,8
• A polysubstance abuse profile; and
• Social isolation and lack of social support.
Interventions should be tailored to deal with
alcohol abuse and the other risk factors identified
above. In most provinces, there is no waiting period
for pregnant women in need of addictions treatment.
Studies indicate that supportive counseling and/or a
case-management approach can help 60% to 80% of
pregnant women reduce their alcohol intake before
the third trimester, and 35% to 50% will stop heavy
drinking.5

Once a diagnosis of FAS is made, intensive case
management must be offered that addresses the
medical and social needs of the birth mother and her
family to prevent future children from being affect-
ed. The risk of recurrence of FAS in families with
one affected child is 771 per 1,000 live births.5 A
lay-advocate approach that supports the mother and
facilitates access to resources has been shown to be
effective in maintaining abstinence and promoting
contraceptive use in high-risk women.9 Similar pro-
grams are now running in many major centers in
Canada (See the FAS/FAE Information Service:
Canadian Centre for Substance Abuse Web site:
www.ccsa.ca/fasgen.htm)

Fetal Alcohol Syndrome

Spectrum of Alcohol Damage and Diagnostic Categories

Alcohol-related effects: ARBD and ARND.
When a pregnant woman consumes alcohol, she
does not drink alone. Alcohol is a known terato-
gen, causing a spectrum of damage that disrupts
fetal development in all three trimesters. Alcohol
and its metabolites interfere with DNA synthesis,
cell division, as well as cell migration and devel-

dopment. Exposure in the first trimester affects
organ development and craniofacial development.
Structural brain abnormalities are most common,
followed by cardiac abnormalities, especially sep-
tal defects.2 The whole range of ARBDs is pre-
sented in Table 1.10 Exposure in the second
trimester leads to an increased rate of spontaneous
abortions. Exposure to alcohol in the third
trimester has a more severe effect on birth weight.

ARND can manifest as either structural changes
(e.g., microcephaly, agenesis of the corpus callo-
sum or cerebellar hypoplasia) or functional deficits
affecting behavior and cognition (Table 1).10

FAS diagnostic categories. FAS with confirmed
alcohol exposure was first defined in 1973. Researcher
s and clinicians have struggled to find
consistent terminology to describe the spectrum of
effects and the individual criteria that should be
included in the diagnosis. In 1996, the US institute
of Medicine published new diagnostic categories,
which are summarized in Table 2.10,11

FAS without confirmed alcohol exposure is
used to describe children who have the growth,
facial and central nervous system (CNS) charac-
teristics of FAS, but there is no way to accurately
verify the mother’s use of alcohol.

The term partial FAS is applied to the patient
with a confirmed history of prenatal alcohol expo-
sure, who has some, but not all, of the characteris-
tics of FAS. Partial does not mean the condition is
less severe than full FAS. Many patients diag-
nosed as having partial FAS would have been des-
ignated as having FAE under previous diagnostic
systems. The use of the term FAE has been dis-
couraged by its originator, Dr. Sterling Clarren,
since it is non-specific and encompasses a broad
range of conditions that have varying severity and
outcomes.12 A common misconception is that FAE
is a less severe form of FAS. Although the patient
designated as having FAE may not have all of the
physical abnormalities of FAS, the cognitive and

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behavioral impairments and, therefore, life-long disabilities, are similar in severity.

### Primary Features and Disabilities of FAS: Growth, Face and Brain

The growth pattern characteristic of FAS likely presents in the prenatal period, and persists as a consistent impairment over time. By contrast, growth deficiency due to postnatal influences is likely to present periodic fluctuations in the growth curve. Patients with severe growth impairments due to alcohol exposure present with height and weight percentiles less than the third percentile. Those with moderate impairment fall between the third and tenth percentile. The most consistent features in the FAS facial phenotype include small palpebral fissures (reflecting small eyes), a smooth philtrum and a thin upper lip. Criteria and norms have now been established by Astley and Clarren, and they allow for more accurate case definitions. A manual and CD-ROM have been developed to provide clinicians with specific information on how to examine and measure the face.

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### Table 1

**Diagnostic Criteria for Alcohol-Related Effects**

1. **Alcohol-related birth defects**
   - **Cardiac:** Atrial septal defects, ventricular septal defects, aberrant great vessels, tetralogy of Fallot.
   - **Skeletal:** Hypoplastic nails, shortened fifth digits, radioulnar synostosis, joint 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, retinal 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.

2. **Alcohol related neurodevelopmental disorder**
   - Presence of A and/or B.
     - A. Evidence of CNS abnormalities in at least one of the following:
       - Decreased cranial size at birth
       - Structural brain abnormalities (e.g., microcephaly, 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
     - B. Evidence of a complex pattern of behavior 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 judgment.

### Table 2

#### Diagnostic Criteria for FAS

1. **FAS with confirmed maternal alcohol exposure**
   
   **A.** Confirmed maternal alcohol exposure.
   
   **B.** Evidence of characteristic pattern of facial anomalies including short palpebral fissures and abnormalities in the premaxillary zone (e.g., flat upper lip, flattened philtrum, and flat midface).
   
   **C.** Evidence of growth retardation in at least one of the following:
      - Low birth weight for gestational age
      - Decelerating weight over time not due to other identified causes
      - Disproportional low weight to height
   
   **D.** Evidence of CNS abnormalities in at least one of the following:
      - Decreased cranial size at birth
      - Structural brain abnormalities (e.g., microcephaly, cerebellar hypoplasia)
      - Neurological hard or soft signs (as age appropriate), such as impaired fine motor skills, neurosensory hearing loss, poor tandem gait, poor hand-eye coordination

2. **FAS without confirmed maternal alcohol exposure**
   
   B, C, and D above

3. **Partial FAS with confirmed maternal alcohol exposure**
   
   **A.** Confirmed maternal alcohol exposure.
   
   **B.** Evidence of some components of the pattern of characteristic facial anomalies and either C or D or E below.
   
   **C.** Evidence of growth retardation, 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 abnormalities in at least one of the following:
      - Decreased cranial size at birth
      - Structural brain abnormalities (e.g., microcephaly, cerebellar hypoplasia)
      - Neurological hard or soft signs (as age appropriate), such as impaired fine motor skills, neurosensory hearing loss, poor tandem gait, poor hand-eye coordination.
   
   **E.** Evidence of a complex pattern of behavior 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 judgment.

1. 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.

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This cluster of minor facial abnormalities is very specific to alcohol exposure on Day 20 post-conception, and is likely related to excessive cell death in the midline of the developing embryo.

Several practice points emerge from this finding:
- The likelihood of an FAS diagnosis is greatest when the three features of small palpebral fissures, smooth philtrum and thin upper lip, are present;
- The more severe the facial manifestations, the greater the probability of midline and other structural CNS defects; and
- If there are no facial features, but there is a definitive history of prenatal alcohol exposure, another diagnosis within the alcohol-related spectrum needs to be explored.

Figure 1 shows the major and associated features of FAS. Other conditions that have similar facial characteristics include Fragile X Syndrome, Velocardiofacial Syndrome and Fetal Dilantin Syndrome.

Figure 2 shows a graphic representation of the brain manifestations of prenatal alcohol exposure. This photograph has been used widely in FAS public awareness campaigns, and compares the brain of a child who died at birth due to severe FAS to that of a healthy newborn.

The severity of brain dysfunction can be placed on a continuum from subtle neurobehavioral deficits to obvious structural abnormalities. The primary functional brain disabilities of FAS can be organized according to a mnemonic “ALARM.”
- Adaptive Functioning
- Language
- Attention
- Reasoning
- Memory

The level of adaptive functioning and the ability to live independently is often less than half the patient’s chronological age. Less than 10% of adults with FAS live independently, due to their impairment in social and life skills, although many will have low-average or average intellectual abilities.

Language deficits result from a global impairment in executive functioning, due to prenatal alcohol exposure. The understanding of complex language and figures of speech is markedly deficient. The use of language, therefore, as a social or behavioral mediator, is poor and further compromises the adaptive functioning of the FAS-affected individual. Traditional tests of language often do not detect the deficits seen in FAS. As a result, a speech and language pathologist familiar with testing the social use of language and language pragmatics, should be consulted.

The majority of children with FASD have attentional difficulties and would fulfill diagnostic criteria for ADHD. However, the pattern of attention deficit is different in FASD in the following ways:
- Attentional problems have an earlier onset and are severe at a younger age;
- FASD conditions have more complex comorbidities than are typically seen with ADHD, including cognitive deficits, verbal learning disorders, disruptive behavior disorders and early onset of sexually impulsive behavior; and
- Many children with FAS respond poorly to methylphenidate for treatment of attentional symptoms.

Reasoning and memory deficits are also related to basic impairments in executive functioning of the brain and become more noticeable when the FAS-affected person reaches school age. Patients with FAS are often slow to learn new skills and do not learn from past experiences. Problems are also seen in the areas of math and telling time, due to deficits in visual spatial memory. Patients with FAS are often slow to learn new skills, do not learn from past experiences, often get lost and lose things easily. Intelligence quotient (IQ) testing will not detect reasoning and memory deficits seen in FAS. More specialized neuropsychological tests are required.
Each component of the presentation of FAS can range in severity, according to the patient’s age and the particular impact the alcohol has on each component of the diagnosis. It is, therefore, easy to see why no two individuals with FAS will present with the same constellation of abnormalities and disabilities. In response to these limitations, Astley and Clarren have developed a new diagnostic system where growth, facial phenotype, CNS dysfunction and alcohol exposure vary along separate continua.\textsuperscript{13}

The “Four-Digit Diagnostic Grid” allows the features of growth, face, brain and alcohol use to be ranked independently on a four-point Likert scale, with one reflecting complete absence of FAS features and four reflecting a strong presence of FAS features. Many diagnostic teams in Canada and the US are now using this approach.

**Secondary Disabilities, Co-existing Conditions and Protective Factors**

The secondary disabilities of FAS arise after birth as a result of the neurological deficits and come at a high cost to the individual, her family and society. The estimated cost per case of FAS is between $2.5 million and $3 million.\textsuperscript{10}

Common secondary disabilities include:

- Mental health disorders — depression and suicide threats and attempts are most common;
- Disrupted school and employment experience;
- Trouble with the law;
- Inappropriate sexual behavior and involvement in the sex trade;\textsuperscript{10} and
- Addictions.

![Facies in Fetal Alcohol Syndrome](image)

**Figure 1.** Facial characteristics of FAS.

![Figure 2. This photograph has been used widely in FAS public awareness campaigns, and compares the brain of a child who died at birth due to severe FAS to that of a healthy newborn.](image)

**Figure 2.** This photograph has been used widely in FAS public awareness campaigns, and compares the brain of a child who died at birth due to severe FAS to that of a healthy newborn.
Fetal Alcohol Syndrome

FAS usually occurs within a constellation of co-existing prenatal and postnatal, social, and medical comorbidities. Hence, some common co-existing conditions that must be considered include:
• Attachment disorders due to multiple caregivers and placements; and
• Signs of abuse and neglect.

The impact of these secondary conditions may be reduced significantly by the following protective factors:
• Early diagnosis, preferably before age six;
• Appropriate interventions for primary and secondary disabilities; and
• Placement in a stable and nurturing environment that is non-abusive.10

Key Points in Diagnosing FAS Across the Lifespan

FAS is a clinical diagnosis that can have varying physical and developmental manifestations across a patient’s lifespan. A multidisciplinary team approach is recognized as a best practice standard for making the diagnosis and planning interventions. Figure 3 outlines the primary and secondary multidisciplinary teams.

Key practice points to consider when making the diagnosis of FAS:
• Facial features, such as the smooth philtrum and thin upper lip may not be present in the newborn period and are most pronounced between eight months and eight years;
Fetal Alcohol Syndrome

- Facial features and growth delay diminish in adolescence;
- In infancy and early childhood (0 to five years), key features include delayed developmental milestones, poor sleeping patterns, problems with feeding and problems adapting to change;
- From ages six to 11, significant difficulties with academic achievement, attention and behavior difficulties and problems with social and adaptive functioning become apparent; and
- In adolescence and adulthood, secondary disabilities, including problems with independent living, social integration, psychiatric disorders and involvement with the justice system become evident.10

Management

Management in the primary-care setting begins with screening for the key manifestations of FAS and FASD disorders, and facilitating an early and accurate diagnosis by a multidisciplinary team that can address primary and secondary disabilities. It is important to recall that FAS is a diagnosis for two — when a child is diagnosed with FAS, the biological mother is also diagnosed with, at least, a past misuse or problem with alcohol. Biological parents should be identified and supported, so future pregnancies are not alcohol-affected.

Additional ongoing management strategies include:
- Support to caregivers and educators through the provision of resources and information (see the patient information sheet at the end of this article);18,19
- Routine screening of patients with FAS and related conditions for the primary brain disabilities — Adaptive functioning, Language, Attention, Reasoning and Memory (ALARM) and advocacy for early and intensive intervention;
- Identify advocates for the affected individual who can act as an “external brain” (e.g., protector or custodian);
- If medications are necessary, provide close follow-up, monitor for side effects and begin at a lower starting dose; and
- Regular surveillance for onset of secondary disabilities, linked to aggressive intervention.10

In summary, the primary-care physician has an integral role in the effective prevention, diagnosis and management of FAS and related conditions. FAS is 100% preventable through consistent application of primary prevention strategies, systematic screening of all pregnant women and timely provision of interventions.

References

Note: Suggested additional reading is noted with an asterix (*).
15. Fetal Alcohol Syndrome Tutor: Medical Training Software. Interactive software that assists in the screening and diagnosis of fetal alcohol syndrome. FAS Diagnostic and Prevention Network, Department of Laboratory Medicine, and the Office of Continuing Medical Education, University of Washington, Seattle, WA, 1999.*

Internet Resources

Alberta Medical Association: Clinical Practice Guidelines
www.albertadoctors.org

Alcohol Related Birth Injury Site (ARBI)
www.arbi.org/index.html

www.mcf.gov.bc.ca/child_protection/fasfae_index.htm

For Good News
See Page 130 & 131

BC Ministry of Education: Teaching Children with FAS
www.bced.gov.bc.ca/specialed/fas/contents.htm

FAS/FAE Information Service: Canadian Centre on Substance Abuse
www.ccsa.ca/fasgen.htm

Health Canada
www.healthcanada.ca/fas

Seattle University of Washington FAS Diagnostic and Prevention Web site