GUIDELINES FOR DIAGNOSIS

Identifying fetal alcohol spectrum disorder in primary care

Fetal alcohol spectrum disorder (FASD) is a term that encompasses the wide range of physical, mental and behavioural effects that can occur when a person is exposed to ethanol during gestation. The diagnoses under the FASD umbrella are fetal alcohol syndrome (FAS), partial FAS (p-FAS) and alcohol-related neurodevelopmental disorder (ARND). The term FASD is not intended for use as a clinical diagnosis.

Like other developmental disabilities, FAS-related conditions require comprehensive assessment by a team of trained specialists. This has left many primary care physicians unclear of their role in making FASD-related diagnoses in clinical practice. This article highlights the salient messages for primary care physicians from a comprehensive guideline on FASD diagnosis that is included with this issue of *CMAJ* as a supplement and available online at www.cmaj.ca/content/vol172/5_suppl/index.shtml.[1]

Rationale for early diagnosis

Early diagnosis is associated with better long-term outcomes. Targeted interventions in childhood have been shown to reduce the risk for later secondary disabilities, such as dependent living, confinement, and addiction and mental health concerns.[2]

A diagnosis in early childhood may also serve as a clinical biomarker for unrecognized maternal mental health and addiction concerns and provide an opportunity to offer interventions, support and counselling for the birth mother. This, in turn, may decrease rates of maternal–child separation, facilitate recognition and intervention for other affected siblings, and prevent subsequent children from being similarly affected.[3]

Therefore, an FASD diagnosis should result in far more than simply providing patients and their families with a label; it should provide the impetus for early developmental interventions, family support and preventive measures to limit subsequent health problems.

The diagnosis of FAS has not been monitored consistently on a provincial or national basis, which has resulted in significant underreporting and, therefore, an inadequate allocation of resources. A standardized interdisciplinary approach to early diagnosis is essential for more accurate surveillance of FASD.[4]

The FASD diagnostic process

The diagnostic process for FASDs is complex, given the wide range in the type and severity of the possible effects of prenatal alcohol exposure. The best way to ensure an accurate diagnosis is through a comprehensive, multidisciplinary assessment that involves physicians (generally pediatricians, clinical geneticists, psychiatrists or neurologists) and educational psychologists or neuropsychologists, joined by other developmental specialists as indicated by the age or presentation of the patient (e.g., speech or language pathologists, occupational therapists and physiotherapists).

The criteria for FASD diagnoses are shown in Table 1 (a formal 4-Digit Diagnostic Code is presented in the supplement). Box 1 outlines the key domains of the central nervous system

Table 1: FASD diagnostic criteria

Criterion	FAS	p-FAS	ARND
Growth impairment*	Yes	No	No
Facial anomalies: (1) Short palpebral fissures (2) Smooth or flattened philtrum (3) Thin upper lip	All 3 are present	2 of the 3 are present	None are present
Brain injury	Minimum of 3 CNS domains impaired†	Minimum of 3 CNS domains impaired†	Minimum of 3 CNS domains impaired†
Prenatal alcohol exposure	Confirmed or unconfirmed	Confirmed	Confirmed

Note: FAS = fetal alcohol syndrome, p-FAS = partial FAS, ARND = alcohol-related neurodevelopmental disorder, CNS = central nervous system.
*Prenatal or postnatal or both.
†See Box 1.

Box 1: Key domains assessed for CNS deficit*

- Hard and soft neurological signs
- Brain structure (including microcephaly)
- Cognition
- Communication
- Academic achievement
- Memory
- Executive functioning and abstract reasoning
- Adaptive behaviour, social skills, social communication
- Attention span, activity level, distractibility

*A deficit is defined as abnormality at 2 standard deviations below the mean. All domains are generally assessed by registered psychologists, speech or language pathologists or occupational therapists except neurological signs, which are assessed by specialist physicians or by the specialists already listed.

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(CNS) that are assessed in the diagnostic process. Most of the domains are generally tested by registered psychologists, speech or language pathologists or occupational therapists. To make a diagnosis of brain injury, expert consensus has recommended that there be abnormalities in at least 3 domains at 2 standard deviations below that expected in the normal population.

The role of the primary care physician

The primary care physician is involved in screening for alcohol use in pregnant women, referral for diagnosis, follow-up and linking patients to the community resources.

Screening

Tools for screening for alcohol misuse in women that have been found to be both sensitive and specific include CRAFFT (Box 2) for adolescent women and the modified CAGE (T-ACE and TWEAK; Table 2) for pregnant and nonpregnant women. Additional training to ask questions related to alcohol use during pregnancy in a consistent, sensitive and culturally appropriate manner may be required.

When a patient is found to be misusing alcohol, the primary care physician is in the ideal role to obtain additional information regarding quantity and maximum frequency of alcohol use, to validate the screening, to provide support and counselling when trained in early brief intervention or motivational interviewing techniques, and to make referrals when indicated. National Institute on Alcohol Abuse and Alcoholism guidelines recommend that any woman who drinks more than 7 drinks per week or more than 3 drinks on any given day in the past month should be further assessed for risk of alcohol-related problems. Recent data suggest that prenatal exposure to small amounts of alcohol — as low as 1 standard drink per day, where a standard drink contains about 0.5 oz of ethanol alcohol (e.g., 360 mL [12 oz.] of beer, 150 mL [5 oz.] of wine or 45 mL [1.5 oz.] of spirits) — has some neurobehavioural effects on the offspring.[6] Since the precise lower threshold for ethanol embryopathy is unknown, pregnant women should be advised to abstain from all alcohol use if possible.

Who should be referred for multidisciplinary assessment?

Patients should be referred for assessment in the following situations:

1. presence of 3 facial anomalies characteristic of prenatal alcohol exposure (see Table 1) and evidence of significant

Box 2: Screening questionnaire for diagnosing problem drinking in adolescent women: CRAFFT*

C	Have you ever ridden in a CAR driven by someone (including yourself) who was high or had been using alcohol or drugs?
R	Do you ever use alcohol or drugs to RELAX, feel better about yourself, or fit in?
A	Do you ever use alcohol or drugs while you are by yourself, ALONE?
F	Do you ever FORGET things you did while using alcohol or drugs?
F	Does your family or FRIENDS ever tell you that you should cut down on your drinking or drug use?
T	Have you ever gotten into TROUBLE while you were using alcohol or drugs?

*Each question on the CRAFFT list is given a score of 1. A cut point of 2 provides moderate sensitivity (70%) and excellent specificity (94%) for identifying alcohol use disorders in adolescents. Any positive answer on the CRAFFT list requires further assessment.

Table 2: Screening questionnaires for diagnosing problem drinking

TWEAK (score of 3 or more indicates heavy or problem drinker)	
T (tolerance)	How many drinks* does it take before you begin to feel the first effects of alcohol? (3 or more drinks = 2 points)
W (worried)	Have close friends or relatives worried or complained about your drinking in the past year? (Yes = 2 points)
E (eye-opener)	Do you sometimes take a drink in the morning when you first get up? (Yes = 1 point)
A (amnesia)	Has a friend or family member ever told you about things you said or did while you were drinking that you could not remember? (Yes = 1 point)
K (kut down)	Do you sometimes feel the need to cut down on your drinking? (Yes = 1 point)
T-ACE (score of 2 or more indicates heavy or problem drinker)	
T (tolerance)	How many drinks does it take to make you feel high? (3 or more drinks = 2 points)
A (annoyed)	Have people annoyed you by criticizing your drinking? (Yes = 1 point)
C (cut down)	Have you ever felt you ought to cut down on your drinking? (Yes = 1 point)
E (eye-opener)	Have you ever had a drink in the morning to steady your nerves or to get rid of a hangover? (Yes = 1 point)

Note: TWEAK has a sensitivity of 79% and a specificity of 83%; T-ACE has a sensitivity of 70% and a specificity of 85%.[5]
*A standard drink is commonly defined as one containing 15 g of alcohol (e.g., 360 mL [12 oz.] of beer, 150 mL [5 oz.] of wine or 45 mL [1.5 oz.] of spirits).

prenatal exposure to alcohol at levels known to be associated with physical and developmental effects;
2. presence of 1 or more facial anomalies, growth impairment and known or probable significant prenatal alcohol exposure;
3. presence of 1 or more facial anomalies, 1 or more CNS deficits (see Box 1) and known or probable significant prenatal alcohol exposure;
4. presence of 1 or more facial anomalies, growth impairment and 1 or more CNS deficits;
5. CNS deficits and known or probable significant prenatal alcohol exposure.

Accurate identification of facial anomalies characteristic of prenatal alcohol exposure may be challenging for many clinicians (figures showing these anomalies are provided in the supplement). Although it is unnecessary that facial anomalies be identified for patient referral, the family physician's information on pre- and postnatal growth and on maternal health and risk factors for alcohol intake during pregnancy are essential for accurate diagnosis. It is vital, therefore, that the referral include as much information as possible regarding alcohol and other potential teratogen exposure or genetic factors to assist with diagnostic accuracy and to consider and exclude other confounding or comorbid conditions.

Conclusion

The diagnostic assessment for prenatal alcohol exposure is a diagnosis for the affected individual, their mother, and other possibly affected family members. The primary care physician is a catalyst for prevention of continued exposure and subsequent affected siblings, referrals for diagnostic assessment, and links to services and resources to improve outcome.

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