Summary Paper

Learning Objectives
- Describe and define the updated diagnostic nomenclature;
- Understand and be able to apply the new diagnostic recommendations for FASD into the clinical setting;
- Identify and list criteria for infant/small children and adults that has been newly presented as it relates to FASD diagnosis; and
- Outline and describe the 10 domains identified in the neuropsychological assessment.

Synopsis
The goal of this presentation is to provide an overview of the Canadian diagnostic guideline revision projects, with a specific focus on infant and adult diagnostic criteria and recommendations, and introduction to the updated neuropsychological assessment domains. These guidelines intend to address the concerns and gaps identified by the larger FASD community, with representation from applicable facets of health and social service disciplines. Ultimately, there was unanimous agreement that the diagnostic process continues to involve a comprehensive bio-psychosocial approach including history, physical examination and neurodevelopmental assessment that require a multidisciplinary approach.

Introduction
In 2005, the Fetal Alcohol Spectrum Disorder (FASD): Canadian Guidelines for Diagnosis [1] was published as a supplement to the Canadian Medical Association Journal. The field has since evolved and additional evidence, expertise, and experience are now available necessitating a revision of the 2005 guidelines with the objectives to address new knowledge, as well as specific gaps and inconsistencies in several key areas.

The Public Health Agency of Canada tasked the Canada Fetal Alcohol Spectrum Disorder Research Network (CanFASD) to lead the revision and update of the 2005 guidelines. An expert steering committee was established to review the literature, lead national and international expert consultations, and draft a survey for all diagnostic centres across Canada that would evaluate the utility of the 2005 guidelines. These updated recommendations supercede the 2005 guidelines and respond to an important clinical question, which is to provide current evidence-based recommendations for the diagnostic approach to disabilities associated with prenatal alcohol exposure, as well as clarifications related to diagnostic terminology.

FASD diagnoses require the services of a multidisciplinary assessment team who provides case management, coordination (e.g., nurse, social worker), medical history and examination (e.g., paediatricians, psychiatrists, family physicians or other health care providers with FASD diagnostic expertise and training), and age appropriate neurodevelopmental assessments (e.g., psychologists, speech and language pathologists and occupational therapists). Some settings use additional health, education and social service professionals to meet the needs of their particular population. Teams can be physical or virtual.

Guideline Development
The updated and revised diagnostic guidelines for FASD have attempted to address the identified limitations leading to improved clarity and consistency. A multidisciplinary approach continues to be the standard for collecting precise data that will provide information about the incidence and prevalence of FASD. This information continues to be of paramount importance for informing prevention and intervention strategies and policy.
**KEY UPDATES**

**Prenatal Alcohol Exposure**

At this time, the threshold of alcohol exposure known to be associated with adverse neurobehavioural effects is 7 or more standard drinks per week, or any episode of drinking 4 or more drinks on the same occasion. The DSM-5, which includes proposed criteria for an alcohol-related diagnosis as a condition for further study, recommends a similar threshold. Because the effect sizes seen with a single binge episode are relatively small, a threshold of 2 binge episodes is recommended as a minimum for diagnosis. These recommendations are tentative, and may become outdated as more data becomes available.

Minimal drinking, below the threshold described above, has not been shown to be associated with neurobehavioural effects in humans, but also has not been studied thoroughly enough to be considered safe. There is no known safe amount of PAE, and public health agencies should advise women to abstain from alcohol while pregnant. A variety of maternal and fetal factors such as maternal age and weight, rate and pattern of alcohol consumption, levels of drinking prior to pregnancy, and nutrition status [19-21] can also mediate the impact of a given dose of alcohol on brain development.

**Diagnostic Categories and Criteria**

**FASD with Sentinel Facial Features**

- Simultaneous presentation of the 3 sentinel facial features (short palpebral fissures, smooth philtrum and thin upper lip)
  - Short palpebral fissures, at or below the 3rd percentile (2 standard deviations below the mean);
  - Smooth or flattened philtrum, 4 or 5 on the 5-point Likert scale of the University of Washington Lip-Philtrum Guide [22, 23]; and
  - Thin upper lip (rank 4 or 5 on the Lip-Philtrum Guide).
- Evidence of impairment in 3 or more of the following central nervous system (CNS) domains:
  - Motor Skills
  - Neuroanatomy /Neurophysiology
  - Cognition
  - Language
  - Academic Achievement
  - Memory
  - Attention
  - Executive Function, including Impulse Control
  - Affect Regulation
  - Adaptive Behaviour, Social Skills, or Social Communication
- Confirmation of prenatal alcohol exposure should be documented if available. This diagnosis should not be made when prenatal alcohol exposure is confirmed absent or at a level definitely below that known to be associated with physical and/or developmental effects.
- Growth impairment and other alcohol-related birth defects should be documented if present.
- Hereditary, prenatal and postnatal factors that may influence developmental outcome should be recorded.
**OR**

**FASD without Sentinel Facial Features**

- Evidence of impairment in **3 or more** of the following central nervous system domains:
  - Motor Skills
  - Neuroanatomy /Neurophysiology
  - Cognition
  - Language
  - Academic Achievement
  - Memory
  - Attention
  - Executive Function, including Impulse Control
  - Affect Regulation
  - Adaptive Behaviour, Social Skills, or Social Communication
- Confirmation of prenatal alcohol exposure is required, with the estimated dose at a level known to be associated with neurobehavioural effects.
- Growth impairment and other alcohol-related birth defects should be documented if present.
- Hereditary, prenatal and postnatal factors that may influence developmental outcome should be recorded.

**At Risk for Neurodevelopmental Disorder and FASD, Associated with Prenatal Alcohol Exposure**

**Designation Criteria**

- Confirmation of prenatal alcohol exposure, with the estimated dose at a level known to be associated with neurobehavioural effects;
- CNS Criteria for the diagnostic categories described above are not met;
- Some indication of neurodevelopmental disorder in combination with another plausible explanation as to why the neurodevelopmental assessment results failed to meet the criteria for significant impairment (i.e., patient was too young; incomplete assessment in adolescents or adults due to social circumstances);
- Growth impairment and other alcohol-related birth defects should be documented if present;
- Hereditary, prenatal and postnatal factors that may influence developmental outcome should be recorded.

**This designation:**

- May also be considered for individuals with the 3 sentinel facial features of FASD, who do not yet have documentation or evidence for the requisite 3 or more CNS criteria including true microcephaly.
- Should not be considered when prenatal alcohol exposure is confirmed absent.

For this designation, an individual still may have FASD, but it cannot be determined at this time. A full re-assessment, including the neurodevelopmental assessment, **must** be performed at a later date, as appropriate. Individuals in this category should receive the same services as those with a diagnosis of FASD, as required to address their current needs. An at-risk designation can be withdrawn if the individual does not show a true neurodevelopmental...
disorder in later years. It is important to emphasize that an at-risk designation must not be a holding pattern; appropriate services to address current and emerging needs should be recommended and accessed.

**Growth**

In North America, FAS was ”discovered” because a group of children were referred to a clinic for growth deficiency, and who were later found to have the other features of what is now known as FAS. At that time, growth deficiency became one of the defining features of FAS. Since then, the importance of growth in the overall presentation of alcohol-related effects has been debated. The predictive value of growth deficiency especially in the absence of documented prenatal alcohol exposure has been queried. Recent evidence [24], plus clinical experience suggest that growth is neither sensitive nor sufficiently specific to indicate an FASD diagnosis. Other contemporary diagnostic approaches have relaxed the criterion for growth deficiency in making the diagnosis, although not removing it entirely. Following an analysis of historical clinical reports, basic science, and clinical research, the committee supported the recommendation to remove growth as a diagnostic criterion.

**Terminology**

The terminology associated with FASD has evolved over the years and has generated some challenges. The recommended adoption of FASD as a diagnostic term reflects an attempt to focus more on the effects of prenatal alcohol exposure on brain and behaviour (neurodevelopment) and to simplify the nomenclature.

**Special Considerations in the Neurodevelopmental Assessment of Infants and Young Children**

- Infants and young children who have confirmed prenatal alcohol exposure, but not all three sentinel facial features and who are too young for a comprehensive neurodevelopmental assessment can be designated *At Risk for Neurodevelopmental Disorder and FASD, Associated with Prenatal Alcohol Exposure*. A complete neurodevelopmental assessment should be recommended at an age-appropriate time to determine if the child meets the diagnostic criteria for FASD.

- Young children with all 3 sentinel facial features and microcephaly should be diagnosed with *FASD with Sentinel Facial Features*. This reflects the fact that children with all 3 facial features and microcephaly have a very high baseline risk of neurodevelopmental disorder [76, 113]. Young children with all 3 sentinel facial features but without microcephaly should also be referred to clinical geneticist and designated as “*At Risk for Neurodevelopmental Disorder Associated with Prenatal Alcohol Exposure and FASD*”. A complete neurodevelopmental assessment should be recommended at an age-appropriate time.

**Special Considerations When Diagnosing Adolescents and Adults**

- The diagnostic criteria for FASD are the same for adults as for younger individuals.

- When it is not possible to obtain a formal adaptive behaviour measure or when there is no suitable informant, historical or current information, derived from a file review may be used as a proxy.

- The recommendations following the diagnostic assessment must address basic and immediate needs of the client, and assist them in accessing the supports and services they need.
References


