Guideline for the Diagnosis of Fetal Alcohol Syndrome (FAS)

This guideline was developed by a working group based on best available evidence and from a province-wide survey of physicians. The development of this guideline was funded as part of the Alberta FAS Initiative and in cooperation with the Prairie Province FAS Initiative.

DEFINITIONS

♦ FASD (Fetal alcohol spectrum disorder) is an umbrella term used that includes:
  • Fetal Alcohol Syndrome (FAS) with Confirmed Maternal Alcohol Exposure
  • Fetal Alcohol Syndrome (FAS) without Confirmed Maternal Alcohol Exposure
  • Partial Fetal Alcohol Syndrome (PFAS) with Confirmed Maternal Alcohol Exposure
  • Alcohol-Related Birth Defects (ARBD)
  • Alcohol-Related Neurodevelopmental Disorder (ARND)

GOALS

This guideline is intended to:
♦ assist health care professionals to recognize the disorders associated with fetal alcohol exposure.
♦ promote early (infancy and preschool) and accurate diagnosis.
♦ prevent secondary disabilities through early diagnosis.
♦ prevent future FAS children in affected families by offering interventions to families which will enable them to abstain from alcohol use when planning or during pregnancy.

SECONDARY DISABILITIES:
♦ mental health problems
♦ disruptive school experience
♦ trouble with the law
♦ inappropriate sexual behaviour
♦ drug/alcohol problems

PROTECTIVE FACTORS:
♦ early diagnosis before age 6
♦ stable and understanding caregiver in a non-abusive environment.
♦ access to resources for person’s with disabilities.

RECOMMENDATIONS

♦ The standard for diagnosis of FAS includes the following clinical indicators (Tables 1 and 2):
  • a history of maternal alcohol consumption during pregnancy;
  • prenatal and/or postnatal growth retardation
  • neurodevelopmental and behavioral characteristics
  • characteristic facial features (see diagram in background section)

♦ Primary care providers should refer any child, adolescent or adult suspected to have FAS to an appropriate specialist such as: a pediatrician, psychiatrist, psychologist, for further assessment.

♦ Once a diagnosis has been made:
  • specific advice and contraceptive counselling can prevent further births of alcohol affected children.
  • aggressive intervention measures with the help of a multidisciplinary team can improve the outcome for the individual; and
  • provide information and support to family/caregivers.

A multi-disciplinary team for care and management could include, at minimum, two or three professionals, depending on need and availability within the area; and could be comprised of the following professionals: physicians, nurses, psychologists, speech pathologists, occupational therapists, educators, and social workers. (Refer to Figure 2 on back cover)

Virtual teams can be created in regions where distance is an obstacle to diagnosing the FAS child (refer to appendix 1)

The above recommendations are systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances. They should be used as an adjunct to sound clinical decision making.
BACKGROUND ON DIAGNOSIS

History of Maternal Drinking

Establishing the history of alcohol consumption is one of the most difficult issues in diagnosing FAS. The pregnant woman who consumes alcohol is not always easily identified. Patients usually are not forthright about their drinking habits nor are they necessarily able to recall the precise quantities and timing of their drinks. However, in the absence of a specific biomarker to detect alcohol exposure, the history remains pivotal in the diagnosis.

The challenge for the physician is to identify women who are drinking alcohol during pregnancy. Problem drinkers cannot be identified by appearance or by socioeconomic characteristics. A systematic drinking history is essential and should be obtained from all patients during the initial history and in subsequent prenatal care.

Taking a history of maternal drinking can be helped by specific screening tools, included in the Prevention of FAS Recommendations.

Physical and Neurological Features and Characteristics

In the most severely affected children, FAS can be diagnosed at birth, however, the characteristic physical features are most pronounced between eight months and eight years of age. Facial abnormalities observed in affected children are the key cluster of physical features of FAS. As the child approaches adolescence, the typical facial features become less pronounced. In some adults, facial characteristics have become so normalized that early childhood photographs must be used to confirm diagnosis. Some authors suggest that FAS may not be recognized until postnatal growth retardation and developmental delay become apparent. Abnormalities in neurodevelopment and behavior are usually evident. Alcohol related birth defects (cardiac, skeletal, renal, ocular, auditory) occasionally occur as well (Table 2).

No single feature alone can be used to diagnose FAS

Growth failure

Alcohol exposure in-utero can cause growth failure either apparent at birth or postnatally.

Facial features

No single facial feature is diagnostic of FAS, but the constellation of short palpebral fissure, smooth philtrum and thin vermilion upper lip are characteristic features.

Neurodevelopmental and behavioral characteristics

FAS results in abnormalities of cognition, language, and behavior. The expression of these abnormalities changes from birth to adulthood.

In infancy and early childhood (0-5 years) they include delayed developmental milestones, poor sleep/wake cycle, attentional deficits, impulsivity, and difficulty adapting to change.

From ages 6 to 11, the following may also appear: significant learning difficulties, cognitive delay, an inability to appreciate cause and effect, and poor understanding of social expectations. Executive function deficits can include: going with strangers, breaking rules (repeatedly), not learning from previous mistakes, having difficulty with time and money, and giving in to peer pressure.

In adolescence and adulthood, these difficulties lead to problems with independent living, competitive employment, social integration, and involvement with the legal system.
Concomitant secondary disabilities, including mental health disorders, problems of substance abuse, and behavior disorder are most obvious during adolescence and adulthood.

Differential Diagnosis

Other medical, psychosocial, and psychiatric conditions/disorders may present similarly to FAS. Usually they can be differentiated by an adequate history and investigation. However, co-morbidities with FAS is a common occurrence.

In an individual presenting with a behaviour disorder or Attention Deficit Disorder, it is important to consider the maternal alcohol use history and a diagnosis.

The diagnostic criteria outlined in Tables 1 and 2 cover the full spectrum of the continuum, recognizing that it is sometimes impossible to confirm maternal drinking. Rather than excluding these cases, it is imperative that the diagnostic criteria enable professionals to make a diagnosis identifying specific areas of difficulty.

In Conclusion

The diagnosis of FAS relies on a composite of specific physical, psychological and behavioral tests. Specific programs or services for the individual and the caregiver are required for accurate diagnosis and appropriate long-term management.

REFERENCES


TOWARD OPTIMIZED PRACTICE (TOP) PROGRAM

The successor to the Alberta Clinical Practice Guideline (CPG) program, TOP is an initiative directed jointly by the Alberta Medical Association, Alberta Health and Wellness, the College of Physicians and Surgeons, and Alberta’s Health Regions. The TOP Program promotes appropriate, effective and quality medical care in Alberta by supporting the use of evidence-based medicine.

TO PROVIDE FEEDBACK

The Working Group for FAS is a multidisciplinary team composed of family physicians, obstetricians, pediatricians, geneticists, Community Medicine specialists, midwives, representatives from AADAC, Alberta Family and Social Services, Health Canada, the Alberta CPG Program, the Reproductive Care Committee, the NECHI Institute, and the public.

The Working Group encourages your feedback. If you need further information or if you have difficulty applying this guideline, please contact:
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Primary Team = for all patients  
Secondary Team = on consultation for individual needs  

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