Fetal Alcohol Spectrum Disorders: An Overview for Pediatric and Adolescent Care Providers

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Fetal alcohol spectrum disorder (FASD) is a term used to describe the spectrum of conditions associated with prenatal alcohol exposure. These are characterized by facial dysmorphia, growth deficits and central nervous system abnormalities. FASDs are the most common preventable cause of intellectual disability in the United States and have high financial costs. Therefore, efforts at prevention are paramount. When an individual with an FASD goes undiagnosed and when appropriate interventions are not instituted, secondary disabilities such as substance abuse, school dropout, and criminal involvement are common with corresponding suffering endured by both the affected individual and the family. The diagnostic process opens up access to existing tools and resources, including the new American Academy of Pediatrics (AAP) FASD algorithm for the evaluation of FASDs, the new AAP FASD toolkit and evidence-based interventions specific to FASDs. Pediatric and adolescent clinicians are challenged to participate in the continuum of care from FASD prevention to identification, diagnosis, and management, including provision of supportive services for families in order for clinicians to make a difference in this 100% preventable disorder.

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Definition of Terms

The term Fetal alcohol spectrum disorder (FASD) describes the spectrum of effects that can be observed in individuals who were exposed to alcohol in utero. As demonstrated in human and animal studies, effects of prenatal alcohol exposure include facial abnormalities, growth deficits, and central nervous system abnormalities. The exposure to alcohol affects the individual's development, learning, and cognition, producing characteristic ways of thinking and behaving.[1] Developmental and behavioral problems in individuals with an FASD reflect alcohol's effect on the brain, and deficits can range from mild cognitive deficits to profound intellectual disability, from subtle effects on memory, executive function, and adaptive behavior to severe behavioral problems that result from poor self-regulation. At one extreme end of the spectrum of disorders arising from prenatal alcohol exposure is fetal alcohol syndrome (FAS). FAS is characterized by facial, growth, and CNS abnormalities in the background of alcohol exposure[2] (Table 1). FAS is diagnosed by the triad of (1) characteristic facial features: short palpebral fissures or lateral eye openings (normograms for palpebral fissures are available from various sources),[3,4] smooth philtrum, and thin vermillion border of upper lip (rank 4–5 on the University of Washington Lip–Philtrum Guide) (Fig. 1), (2) growth deficiency with a height or weight at or below the tenth percentile at any point of the child's life, and (3) evidence of central nervous system abnormality, which is structural, neurological, or functional (e.g., microcephaly, seizures, cognitive or learning deficits, and behavioral dysregulation), etc.[5] (Fig. 2). This is a narrow set of criteria that may miss a number of patients who have other physical and neurocognitive effects from in utero alcohol exposure but do not have the classic triad essential for the diagnosis of FAS. In fact, children who experienced prenatal alcohol exposure but do not meet full criteria for FAS often have similar cognitive and behavioral characteristics.[6]

Other disorders that have been identified within the spectrum include Alcohol-Related Neurodevelopmental Disorder (ARND), Partial Fetal Alcohol syndrome (PFAS), and Alcohol-Related Birth Defects (ARBD).[7] ARND is characterized by evidence of CNS neurodevelopmental abnormalities, including at least one of

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TABLE 1. Criteria for FAS Diagnosis[a]

A. Requires all three of the following findings:
   1. Documentation of all three facial abnormalities (smooth philtrum, thin vermillion, and small palpebral fissures).
   2. Documentation of growth deficits.
   3. Documentation of CNS abnormality.
B. Facial dysmorphia: Based on racial norms, individual exhibits all three characteristic facial features:
   1. Smooth philtrum (University of Washington Lip–Philtrum Guide rank 4 or 5)
   2. Thin vermillion border (University of Washington Lip–Philtrum Guide rank 4 or 5)
   3. Small palpebral fissures (at or below tenth percentile)
C. Growth problems: Confirmed prenatal or postnatal height or weight, or both, at or below the tenth percentile, documented at any one point in time (adjusted for age, sex, gestational age, and race or ethnicity).
D. Central nervous system abnormalities:
   Structural
   Head circumference (OFC) at or below the tenth percentile adjusted for age and sex.
   Clinically significant brain abnormalities observable through imaging.
   Neurological
   Neurological problems not due to a postnatal insult or fever or other soft neurological signs outside normal limits.
   Functional
   Performance substantially below that expected for an individual's age, schooling, or circumstances, as evidenced by the following:
   Global cognitive or intellectual deficits representing multiple domains of deficit (or significant developmental delay in younger children) with performance below the third percentile (2 standard deviations below the mean for standardized testing) OR
   Functional deficits below the 16th percentile (1 standard deviation below the mean for standardized testing) in at least three of the following domains:
      Cognitive or developmental deficits or discrepancies
      Executive functioning deficits
      Motor functioning delays
      Problems with attention or hyperactivity
      Poor social skills
      Other (e.g., sensory problems, pragmatic language problems, or memory deficits)
E. Maternal alcohol exposure
   1. Confirmed prenatal alcohol exposure
   2. Unknown prenatal alcohol exposure

[a]**Adapted with permission from Bertrand et al.[5]**

the following: structural brain abnormalities, such as microcephaly or abnormalities on brain imaging; neurologic hard and soft signs such as deficits in fine motor skills; neurosensory hearing loss; poor tandem gait; and poor eye–hand coordination). The term Neurodevelopmental Disorder Associated with Prenatal Alcohol Exposure (ND-PAE) is the new terminology that has been proposed in the Diagnostic and Statistical Manual (DSM)-V and the criteria for the ND-PAE diagnosis are listed in the appendix of DSM-V under conditions for further study and under Other Specified Neurodevelopmental Disorder 315.8. The proposed criteria include that the patient was "exposed to alcohol at any time during gestation, including prior to pregnancy recognition and the exposure level was more than minimal" along with presence of neurocognitive impairment, impairment in self-regulation, and impairment in adaptive functioning.[8,9] Partial FAS (PFAS) is diagnosed when there is confirmed alcohol exposure in utero and evidence of a characteristic pattern of facial anomalies as well as either growth retardation, CNS abnormalities, or cognitive abnormalities that are characteristic of full blown FAS. ARBDs are diagnosed when there is confirmed exposure to alcohol in utero and birth defects associated with alcohol exposure. It is now known that relying on facial features alone for the identification of children with an FASD underrepresents the population of all alcohol-exposed pregnancies and that severity of atypical facial features may not directly correlate with severity of neurobehavioral effects. A study by Mattson et al.[10] showed that in children with history of heavy prenatal exposure to alcohol, similar levels and patterns of cognitive deficits were seen regardless of meeting facial criteria for FAS. In fact, it is now well known that the most devastating effect of alcohol exposure in utero is its impact on the brain.[11]

Epidemiology

Prenatal alcohol exposure is the most common nonhereditary cause of intellectual disability in the United States.[12] FAS has a prevalence rate of 0.2–1.5 cases per 1000 births across various populations in the United