

Fetal Alcohol Spectrum Disorders *An Overview*

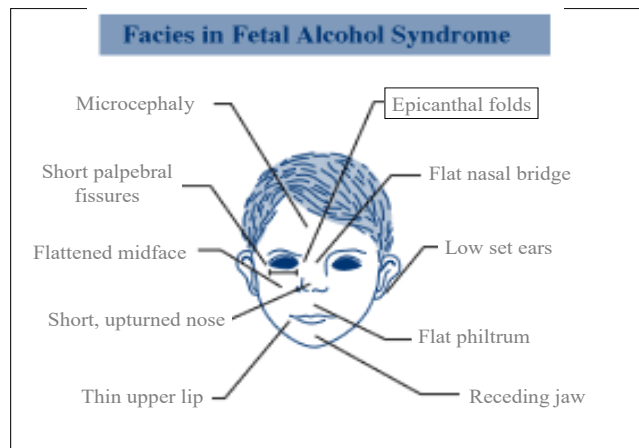
Fetal Alcohol Syndrome (FAS) is the original name given to a cluster of physical and mental defects present from birth that is the direct result of a woman drinking alcoholic beverages while pregnant. Infants and children with FAS have signs in three categories:

1. Growth deficiencies
2. Central nervous system problems
3. Changes in facial structure

Growth deficiencies

In the United States, the average birth weight of babies born at full term (38 to 42 weeks gestation) is 7 pounds 8 ounces, with a normal range down to 5 pounds 8 ounces. Babies born to mothers who use alcohol have an average birth weight of around 6 pounds and are more likely to weigh less than 5 pounds 8 ounces. As children with FAS grow older, they tend to continue to be small – short and underweight – for their age. To meet the FAS diagnostic guidelines set for growth criteria, a child must have either reduced weight *or* height (at or below 10th percentile on standard growth charts) at birth *or* at any point in time after birth.

Changes in facial features



Prenatal alcohol exposure can lead to overall flattening of the middle portion of the face. As a result, children with FAS exhibit the following facial features:

- epicanthal folds (extra skin folds coming down around the inner angle of the eye)
- short palpebral fissures (small eye openings)
- a flattened elongated philtrum (no groove or crease running from the bottom of the nose to the top of the lip)
- thin upper lip
- small mouth with high arched palate (roof of the mouth)



- small teeth with poor enamel coating
- low set ears

These changes can vary in severity, but usually persist over the life of the child. Most people will not recognize any differences when they see the child, but physicians and other practitioners with experience working with children prenatally exposed to alcohol will be able to detect the changes.

A problem arises when clinicians rely too heavily on changes in facial structure to recognize the child affected by prenatal alcohol exposure. In animal studies, pregnant rats given alcohol on days 7 or 8 after conception had newborns with facial features typical of FAS. However, giving the pregnant rats alcohol on days 1 through 6, or on day 9 or any time beyond did not affect the facial features in any way. Thus, there appears to be a very narrow window of alcohol exposure that can affect children's facial features.

Children with Fetal Alcohol Syndrome also may have a variety of malformations of major organs, especially the heart, kidneys, eyes, and ears. Children with prenatal alcohol exposure frequently have vision problems; many have an eye that turns inward (i.e., esotropia, or a "lazy eye"). In addition, the children can have a predisposition to ear infections and a high rate of partial or complete hearing loss (i.e., eighth nerve deafness), so a thorough hearing exam is recommended in the first year of life and should be repeated annually based on the child's speech and language development.

Central nervous system problems

Problems in the central nervous system (the brain) can present as structural, neurological, or functional changes:

- Structurally, a small head circumference (at or below 10th percentile) at birth or at any time thereafter indicates poor brain growth. For example, the average head size of term infants at birth is 35 centimeters, while the head size of a baby with FAS often is less than 33 centimeters.
- Neurologically, damage can lead to seizures, problems in coordination, difficulty with motor control, or several "soft" neurological deficits.
- Functionally, the average IQ in children with FAS is about 68, as compared to that of the general population the average IQ is 100. Alcohol-exposed children, with or without the characteristic facial features or growth retardation, have consistently lower IQ scores than non-exposed children. Even alcohol-exposed children with a "normal IQ" demonstrate difficulty with behavioral regulation, impulsivity, social deficits, and poor judgment, causing problems in day-to-day management in the classroom and home. In fact, children with FASD exhibit a wide range of functional difficulties much more common than intellectual impairment; these difficulties include the following characteristics:
 - learning disabilities
 - poor school performance
 - diminished executive functioning (e.g., organization of tasks, understanding cause and effect, following several steps of directions)
 - clumsiness



- poor balance
- problems with writing or drawing
- Behaviorally, many of the children have a short attention span, and often are described as impulsive and hyperactive.

From a brain structure perspective, prenatal alcohol exposure not only can cause the child to have a small brain overall, but also can stunt the growth of individual parts of the brain. This damaged growth may be present regardless of the child's facial features. Problems with the formation and development of different parts of the brain can result in a wide range of behavioral and learning deficits. Many children with prenatal alcohol exposure have trouble moving information between different brain regions; they cannot effectively use information to self-direct their behavior or to think in the abstract. They may have trouble learning new information and recording it in the brain — and then have even more difficulty retrieving the information already learned. Like Jonny, the child may learn his multiplication tables one day, but forget them the next.

Other parts of the brain also can be affected, impairing the child's ability to coordinate planned motor movements, resulting in impulsive movement and clumsiness. Reduction in the size of the cerebellum in the back part of the brain, for example, produces difficulties with balance and arousal and may be a source of sleep/wake problems. Again, it is important to remember that such problems occur not only in children with the abnormal facial features associated with full expression of FAS, but also in alcohol-exposed children who "look normal."

Another way alcohol can affect brain functioning is by disrupting sugar metabolism in the brain, which may lead to a clinical pattern of distractibility and impulsiveness. To make matters worse, many alcohol-exposed children are simultaneously exposed to their mother's gestational use of tobacco, cocaine, methamphetamine, heroin, marijuana, and other drugs. These substances affect the dopamine, serotonin, and norepinephrine systems in the fetal brain, causing changes in the composition of chemicals that control behavior, thought, emotions, and movement. Thus, the exposed child has trouble responding appropriately to the world around him and looks "out of sync" in the classroom and at home. The difficulty in evaluating and diagnosing children with prenatal alcohol exposure often lies in separating the effects of the alcohol exposure from the effects of the tobacco or other drugs to which the child was exposed.

Problems in diagnosis

Originally, if a woman drank alcohol during pregnancy and gave birth to a child who showed partial or no apparent expression of physical features characteristic of alcohol exposure, her child was said to have fetal alcohol effects (FAE). These children may have had minimal to moderate facial changes or no changes at all, but usually they had some problems with intellectual, behavioral, or emotional development. These difficulties were known to have an impact on learning and long-term development, though just how extensively FAE affected the child was less clear.

More recently, research has demonstrated that children with prenatal alcohol exposure may have significant structural and functional changes in the brain, even though they lack visible physical effects of



the alcohol exposure. Currently, the preferred terminology for children who have been exposed to alcohol but who do not meet criteria in all three diagnostic categories is alcohol-related neurodevelopmental disorder (ARND). In April 2004, a group of federal agencies developed a consensus definition of fetal alcohol spectrum disorders (FASD):

[A]n umbrella term describing the range of effects that can occur in an individual whose mother drank during pregnancy. These effects may include physical, mental, behavioral, and/or learning disabilities with possible lifelong implications.¹

Diagnostic terminology in daily use mainly focuses on FAS or ARND, both of which fall within the larger continuum of effects seen in children with FASD. FASD is not meant to serve as a diagnostic term, but rather a unifying one to help us appreciate the many ways in which prenatal alcohol exposure can become manifest in the affected individual.

Diagnostic dilemmas for parents and physicians

If diagnosis of alcohol-affected children were as easy as the terminology implies, we could move on from this discussion to other more important topics. But the truth is, there is great controversy as to how and when to diagnose children whose mothers drank alcohol during pregnancy. The key barrier to diagnosis is the lack of information regarding maternal alcohol use during pregnancy. Recent studies in general populations of pregnant women in Iowa report that about 25% of the women have drunk some amount of alcohol during gestation, with the highest risk population frequently comprising middle class, well-educated women. However, prenatal care providers often are reluctant to address their patients' drinking, and as a result alcohol use continues to lead to FASD—one of the most commonly missed complications of pregnancy.

Another difficulty in diagnosis relates to the lack of documentation of prenatal alcohol use by the mother and by the lack of physical features among most alcohol-exposed children. Normal growth patterns and normal facial features interfere with appropriate diagnosis. In fact, a recent study showed that 85% of children with FASD are misdiagnosed. The most common mistaken diagnoses range across a broad of array – from Attention Deficit Hyperactivity Disorder to Oppositional Defiant Disorder to Bipolar Disorder, and so on.

The clarification of these diagnostic issues is important for all those who care for children, especially pediatricians whose role is to recognize, early, children who may be at risk from prenatal exposure to alcohol as well as foster and adoptive parents who must advocate for the child to ensure access to early intervention programs. Without a diagnosis of alcohol-related risk, many children will not be deemed eligible for early intervention and school-based treatment programs, nor will insurance companies pay for related health care interventions. Parents and caregivers thus find themselves in a position of advocating for children whose behavior difficulties are not deemed “bad enough” to receive services.

¹ Bertrand J, Floyd RL, Weber MK, et al. National Task Force on FAS/FAE. Fetal Alcohol Syndrome: Guidelines for Referral and Diagnosis; 2004; Atlanta, GA: Centers for Disease Control and Prevention.