Do We Need the Term "FAE"?

ABBREVIATIONS. FAS, fetal alcohol syndrome; FAE, fetal alcohol effect.

Fetal alcohol syndrome (FAS) was first recognized as a distinct clinical entity by Jones and Smith in 1973.' In their first reports,[2,3] all affected children had been born to severely alcoholic women, and had in common problems in three major categories:

1. prenatal and/or postnatal *growth deficiency;*
2. abnormal brain function reflected in *mental deficit;* and
3. a distinctive pattern of mild facial *dysmorphology*.

Later psychological studies revealed a pattern of *behavioral aberration,* which is quite common in affected children, but has not been shown to be unique to FAS.[4,5]

As is usually the case with newly described clinical syndromes, diagnosticians soon began to realize that they were encountering children with some, but not all the classical signs of FAS. Typically, the maternal history indicated moderate to severe gestational alcohol abuse and the child showed developmental delay and behavioral abnormalities, but the characteristic facial anomalies were absent and growth and development were variably affected. Because a diagnosis of FAS demanded the presence of all three primary diagnostic criteria, (growth deficiency, CNS dysfunction, and physical characteristics)[6] a term was needed to refer to children with what seemed to be form *fruste* FAS, and references to "suspected fetal alcohol effects" began to appear in the literature.[7-9] This was not intended to be a diagnosis, but only a "bookmark," suggesting that the abnormalities seen in the child were compatible with those caused by prenatal alcohol exposure, but that the pattern was not sufficiently complete to permit definition of FAS.

Unfortunately, within a few years after its introduction, the designation fetal alcohol effect (FAE) began to be applied more or less indiscriminately to children with a variety of problems, even those with simple growth deficiency or isolated behavioral aberration, based almost entirely on the knowledge (or suspicion) that their mothers drank alcohol during pregnancy. Not only clinicians, but concerned teachers, social workers, and foster parents, seeking expla-

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nations for the problems in children under their care, seized upon the "diagnosis" of FAE. Some health agencies followed suit, accepting FAE as a medical condition making a child eligible for financial assistance and educational intervention. In the absence of definitive laboratory tests or other means of confirming that in utero alcohol exposure is the cause of the child's problems, the "diagnosis" of FAE can be neither proven nor refuted, yet it is used to make decisions ranging from insurance coverage to sentencing for capital crime.

Although the general criteria for diagnosing FAS have been clearly delineated and are widely accepted, clinicians and investigators remain frustrated by the lack of a meaningful definition of FAE. The original Fetal Alcohol Study Group of the Research Society on Alcohol in their 1980 report suggested that FAE encompassed "Any conditions thought to be secondary to alcohol exposure in utero."[10] Clearly, such a definition allows for wide divergence in interpretation and has little relevance in the clinical setting. In fact, the *known* "effects" of prenatal alcohol exposure are those making up FAS. Each of the individual components is nonspecific, and only their combination with each other allows definition of the diagnosis.

For example, the diagnostic physical features of FAS all are minor anomalies or structural variants that can be found as isolated characteristics in normal individuals and families. Each of these nonspecific variants fits the criteria for polygenic (or multifactorial) inheritance: they show a wide spectrum of variability and occur with higher frequency in more closely related individuals, but do not follow a classic Mendelian inheritance pattern. The significance of such minor physical features lies in their association with one another to form a recognizable pattern that helps define a specific syndrome. The dysmorphic characteristics seen in FAS, when combined with growth and mental/behavioral aberrations, paints a unique picture that has been reported only in children prenatally exposed to alcohol. No such consistent pattern exists for FAE.

The term and concept of FAE does have validity in one application; in human population studies and animal research in which the independent variable is maternal alcohol consumption during pregnancy. If measurable differences can be found when a group of offspring with documented prenatal alcohol exposure is compared with an otherwise identical but unexposed population, it is justifiable to suggest that alcohol caused that difference. Important contributions have been made using these techniques in the areas of growth[11,12] and intellectual development.[13] It must be emphasized, however, that such effects represent statistical correlations and not final proof of alcohol as the causative agent.

The evaluation of a specific patient in the clinical setting is quite a different matter. The greatest concern centers on individuals with behavioral difficulties and learning disorders. Hyperactive children and adolescents with conduct disorders often are suspected of FAE, even in the absence of knowledge about maternal alcohol intake, because such behavior forms a part of FAS. As yet, however, a specific psychological/behavioral phenotype *unique* to those prenatally exposed to alcohol has not been defined, and attribution of the aberrant conduct to in utero alcohol exposure remains problematic.

Several unfortunate consequences may result from inappropriately using the term FAE:

1. Presupposition that alcohol is the major (or only) cause of the child's problems may end the search for other possible causes such as psychosocial deprivation and abuse;
2. Educators and care providers may base their expectations for the child's performance on that of children with FAS;
3. Women are stigmatized for having damaged their children by drinking during pregnancy when it is by no means certain that they have done so;
4. Clinicians become frustrated by the imprecision of the "diagnosis" of FAE and thus disregard any possible contribution of alcohol exposure to their patients' problems; and
5. Efforts to learn the real magnitude of the problem of prenatal alcohol damage are frustrated by overdiagnosis.

On the other hand, foster and adoptive parents have raised the objection that without a "diagnosis" such as FAE, a child may not quality for special education programs, Social Security payments, and other benefits. Some state and local social service agencies do indeed require a medical diagnosis from a specified list as justification for provision of services. We believe that it is unfair that a mother must be stigmatized to obtain services for her child; such decisions should be based on demonstrated patient need, rather than arbitrary categories of diagnosis. The results of specific physical, psychological, and behavioral testing form the best basis for such decisions, and pediatricians armed with this information can be effective advocates for their patients in helping to obtain appropriate services as well as helping change the present inadequate qualification categories.

We propose abandoning the clinical use of the term fetal alcohol effect with its implications of causation, and urge simple recording of the verifiable conclusions concerning the individual patient. For patients referred because of suspicion of FAS, a three-axis scheme seems appropriate. The first entry refers to exposure status, the second to presenting problems, and the third to definitive diagnosis. If prenatal alcohol exposure has taken place, but FAS cannot be substantiated, the exposure still should be indicated, and any nonspecific abnormalities or problems noted.

Thus, the problem list for a growth-deficient newborn with no other physical stigmata whose mother admits to drinking during pregnancy would read: 1) prenatal alcohol exposure; 2) intrauterine growth retardation; and 3) diagnosis deferred.

In a similar fashion, an adolescent for whom gestational alcohol exposure is suspected but not confirmed might have a problem list reading: 1) questionable prenatal alcohol exposure; 2) normal growth, microcephaly, and learning deficits; and 3) inadequate evidence to define the possible contribution of alcohol to these problems.

Obviously, if information later came to light that would add further specificity, the "diagnosis" could be changed as needed.

This paper is simply a call for accuracy and a more conservative approach to diagnostic terminology. A diagnosis that implies causation should not be applied unless the relationship can be proven. Until it is known which features in individuals prenatally exposed to alcohol are uniformly and exclusively caused by that exposure, we suggest reporting only objective descriptors unless the "full" fetal alcohol syndrome can be confirmed.

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REFERENCES

1. Jones KL, Smith DW, Ulleland CN, Streissguth Al'. Pattern of malformation in offspring of chronic alcoholic mothers. Lancet. 1973;1: 1267-1271
2. Jones KL, Smith DW. Recognition of the fetal alcohol syndrome in early infancy. Lancet. 1973;2:999-1001
3. Jones KL, Smith DW, Streissguth Al', Myrianthopoulos NC. Outcome in offspring of chronic alcoholic women. Lancet. 1974;1:1076-1078
4. Streissguth Al'. The behavioral teratology of alcohol: performance, behavioral and intellectual deficits in prenatally exposed children. In: West J, ed. *Alcohol and Brain Development. New* York, NY: Oxford University Press, Inc; 1986:3-44
5. Streissguth Al', Barr HM, Sampson PD. Alcohol use during pregnancy and child development: a longitudinal, prospective study of human behavioral teratology. In: Greenbaum CW, Auerbach JG, eds. Longitu*dinal Studies of Children at Psychological Risk: Cross-National* Perspectives. Norwood, NJ: Ablex; 1992:174-200
6. Sokol RJ, Clarren SK. Guidelines for use of terminology describing the impact of prenatal alcohol on the offspring. *Alcohol Clin Exp Res.* 1989; 13:597-598
7. Clarren SK, Smith DW. The fetal alcohol syndrome. N *Engl* J Med. 1978;298:1063-1067
8. Smith DW. Fetal drug syndromes: effects of ethanol and hydantoins. Pediatr *Rev.* 1979;1:165-172
9. Smith DW. Fetal alcohol syndrome and fetal alcohol effects. Neurobehav Toxicol Teratol 1981;3:127
10. Rosetti HL. A clinical perspective of the fetal alcohol syndrome. *Alcohol Clin Exp Res.* 1980;13:118
11. Kyllerman M, Aronson M, Sabel KG, Karlberg E, Sandin B, Olegard R. Children of alcoholic mothers (growth and motor performance compared to matched controls). *Acta* Paediatr *Scand.* 1985;74:20-26
12. Day NL, Robles N, Richardson G, et al. The effects of prenatal alcohol use on the growth of children at three years of age. *Alcohol Clin Exp Res.* 1991;15:67-71
13. West JR, ed. *Alcohol and Brain Development. New* York, Oxford University Press; 1986