Introduction

The three papers by Astley, O’Brien and Mattson, and O’Connor reviewing the current knowledge base of Fetal Alcohol Spectrum Disorders (FASD) reveal the ongoing challenges that face researchers and clinicians, in particular the accurate diagnosis and meaningful differentiation of children who are prenatally-exposed to alcohol.

The diagnosis of Fetal Alcohol Syndrome (FAS) is based on three criteria: growth retardation, central nervous system (CNS) impairment, and characteristic facial dysmorphology. However, as acknowledged in all of the papers, most children with prenatal exposure to alcohol do not fully manifest these criteria but demonstrate a wide range of less specific physical changes and neurodevelopmental deficits. To address this issue, diagnostic terminology has been expanded to include a wide range of signs and symptoms that fall under FASD. Although this terminology accomplishes what it was designed to do – encompass all children with prenatal alcohol exposure – its use has muddied the waters and led to even more confusion around the diagnosis of FAS. In addition, an inexact diagnostic approach complicates attempts to address the neurobehavioural profile and the clinical needs of children affected by prenatal alcohol exposure.

Research and Conclusions

Diagnostic classification within FASD

Over the years, the relationship between functional impairment and the physical manifestations of prenatal alcohol exposure has remained unclear. In 2001, Astley and Clarren found that alcohol-exposed children with...
more severe facial phenotypes demonstrated more impaired levels of cognitive, neurodevelopmental and visual motor functioning. Most recently, magnetic resonance studies of children with fetal alcohol spectrum disorders demonstrated increased brain damage that correlated with more severe facial dysmorphology and more severe neurodevelopmental dysfunction. On the other hand, Mattson, et al. found that the degree of the severity of neurodevelopmental deficits evident in children with prenatal alcohol exposure is independent of the physical features associated with FAS. Further investigation of the connection between physical and imaging findings and the severity of dysfunction is necessary; however, accurate results depend upon a clear and consistent diagnostic schema across the fetal alcohol spectrum.

A recent study compared neurodevelopmental functioning among children with fetal alcohol syndrome (FAS), pFAS (partial fetal alcohol syndrome) and alcohol-related neurodevelopmental disorder (ARND). Clinical criteria for placement in each of the diagnostic groups included confirmed prenatal exposure to alcohol, documentation of all three criteria for facial dysmorphology, and past or current growth impairment (height or weight) below the third percentile. In addition, in order to meet criteria for abnormal central nervous system functioning, children had to have a head circumference below the third percentile, evidence of global cognitive functioning below the third percentile, and/or three domains of neurodevelopmental functioning greater than two standard deviations below the normed mean, to ensure that children were not misclassified. It should be noted that this paper relied on third percentile rather than tenth percentile cut-off for definition of growth impairment and microcephaly, as opposed to the recommendations from the Centers for Disease Control and Prevention (CDC) and the Institute of Medicine (IOM) which rely on the tenth percentile as the defining cutoff.

The children who met tightly-defined physical criteria for a diagnosis of FAS were significantly different neurodevelopmentally from alcohol-exposed children who did not meet all criteria, while children with pFAS and ARND were similar in all neurodevelopmental domains that were tested. Children with FAS exhibited the most impaired level of general intelligence, significantly-worse language-based memory, and significantly-poorer functional communication skills. All three groups of alcohol-affected children demonstrated executive functioning deficits, but the FAS group of children performed significantly worse on sequencing and shift tasks than either the pFAS or ARND groups. Similar to previous studies of behaviour in children with FASD, there was a high rate of diagnosis of Attention Deficit Hyperactivity Disorder (ADHD) in each of the three groups: 60% of children with FAS, 88.9% of children with pFAS, and 76.9% of children with ARND met DSM-IV criteria for a diagnosis of ADHD.

In her paper, Dr. Astley makes a strong case for clear and precise measurement and classification of children with prenatal alcohol exposure. However, although the 4-digit code is well suited for use in specialty clinics and research programs, the reality is that most practitioners find it cumbersome when using it in the field. O'Brien and Mattson’s paper is a case in point. The authors use the term “FASD” to review the literature on neurobehavioural profiles, since most studies do not differentiate what specific diagnostic groups within the fetal alcohol spectrum the various studies are examining.

Secondary disabilities

Streissguth and her colleagues defined the primary disabilities of FAS as those directly related to intrauterine alcohol toxicity. The notion of secondary disabilities was used to “encompass the measurable difficulties that people with FAS/FAE (fetal alcohol effects) face as they mature…” In a four-year study of
secondary disabilities, the researchers found that over 90% of the subjects had mental health problems.\(^{18}\)

However, opinions regarding the etiology of mental health disorders in alcohol-exposed children vary widely. O’Connor et al\(^{19,20}\) suggested that the high proportion of children with mood disorders was due to the damage sustained by the *basal ganglia* and *cerebellum*. On the other hand, O’Connor et al.\(^{21,22}\) also posited that in addition to a genetic predisposition for depression and difficult temperaments that are a result of their prenatal exposure to alcohol, the development of optimal relationships between mother and child are impeded by the mothers’ alcohol use and mood disorder. The role of out-of-home placement in increasing alcohol-exposed children’s risk for mental health disorders\(^{23}\) raises another confounding factor, since most research has documented an increased prevalence of psychopathology among children in the foster care system.\(^{24-26}\)

A new study by our group\(^{27}\) examines the prevalence of mental health disorders among children with FAS and ARND in out-of-home placement as compared to a similar group of children who had no prenatal exposure to alcohol. Not surprisingly, the rates of mental health disorders in both groups of children were markedly higher than rates found in the general population of children in the United States. The prevalence of mental health disorders among children in the alcohol-exposed cohort were similar to those rates found in previous studies of alcohol-exposed children.\(^{18-23}\) ADHD was the most common mental health disorder, occurring in almost 75% of the alcohol-exposed group, significantly higher than the 58% rate found among the non-exposed group. The study found only 19% of the alcohol-exposed children met criteria for a mood disorder, a rate similar to that of Fryer et al. (18%),\(^{23}\) while the rate of mood disorders was significantly higher among the non-exposed children in our sample (37.5%).

In examining the relative contributions of factors related to prenatal alcohol exposure as opposed to child welfare factors related to being in out-of-home placement, logistic regressions revealed that children with a history of physical or sexual abuse, regardless of prenatal alcohol exposure history, had the highest rates of anxiety disorders, and environmental factors, such as number of placements and length of time in the current placement, were more predictive of mood disorder than the biological toxicity of alcohol exposure. Much further research is needed in this arena, especially work that includes appropriate comparison groups that can help differentiate biological and environmental factors that explain mental health disorders in children with prenatal alcohol exposure.

**Policy Recommendations and Implications**

There is a great deal of ongoing discussion and controversy regarding the diagnostic criteria for FAS, pFAS, ARND and other components across the fetal alcohol spectrum. The IOM\(^{12}\) guidelines, as clarified by Hoyme,\(^ {28}\) utilize a cutoff for abnormal growth as the tenth percentile, require only two of three facial abnormalities, and are nonspecific as to central nervous system functioning. Current recommendations regarding facial criteria from the CDC\(^ {11}\) are more stringent than those of the IOM but set the threshold for abnormal CNS functioning only at greater than 1 standard deviation below the mean in three domains. In addition, thresholds for growth impairment (weight, height, head circumference) are set by the CDC at only the tenth percentile.

Based on current knowledge, I would recommend that the term pFAS be eliminated and alcohol-exposed children be diagnosed as having FAS or ARND. Confirmation of prenatal alcohol exposure should be required in all cases. Growth retardation should be defined as current or past weight and/or height less than the third
percentile, rather than the tenth percentile. A finding of facial dysmorphology should require abnormal measurements of the upper lip (rank 4 or 5) and the philtrum (rank 4 or 5) and shortened palpebral fissures greater than two standard deviations below the mean.\textsuperscript{6,29} This is consistent with the CDC’s\textsuperscript{11} and Astley’s\textsuperscript{6-8,30} recommendations, but more stringent than the guidelines issued by the IOM.\textsuperscript{12} To qualify as having evidence of central nervous system abnormalities, the child’s current head circumference should be below the third percentile for age and gender rather than the tenth percentile as currently recommended, and/or the child should demonstrate functional deficits of global cognitive delays with performance below the third percentile on standardized testing or three or more domains of neurodevelopmental functioning more than two standard deviations below the normed mean on standardized measures of cognitive, executive, memory, adaptive, motor, attentional, social skills, or sensory functioning.

Children with documented prenatal alcohol exposure who meet all physical criteria for growth impairment and facial dysmorphology as well as neurodevelopmental deficits would be assigned a diagnosis of FAS. Children with documented prenatal alcohol exposure who do not meet all growth and/or facial criteria but who meet criteria for neurodevelopmental deficits would be classified as ARND. Utilizing two diagnoses – FAS and ARND – with strict criteria delineated for all to follow would create a common language and diagnostic schema that would be suitable in the clinical setting while establishing a consistency with ongoing research. This would be invaluable in further work to determine the neurobehavioural profile for the two groups.

Given the lack of clear evidence regarding the relationship between prenatal alcohol exposure and mental health problems, I would suggest we use more precise terminology and speak of mental health disorders that co-occur in children with FAS and ARND. Labeling mental health disabilities as “secondary” implies a more direct cause and effect association than can be documented through current research. Rather, the correlation is more likely the result of environmental factors that act on the biologically vulnerable brain of the child with FASD. From a policy perspective, eliminating the use of the term “secondary disabilities” would stress the importance of early identification and intervention for alcohol-exposed children at risk for abuse and neglect and the need to support and enhance the stability of the children’s placements whether they be in the biologic home or in out of home placement. This, in turn, would perhaps decrease the rates of mental health disorders in this population of high-risk children.

References


