

DIFFERENTIATING FULL FETAL ALCOHOL SYNDROME FROM INCOMPLETE FETAL ALCOHOL SYNDROME – STILL A CHALLENGE

Brittany Sauv , Graduate Student, Division of Clinical Pharmacology, The Hospital for Sick Children

A CRITICAL REVIEW of “Characteristics of children who have full or incomplete fetal alcohol syndrome.” *Kvigne VL, Leonardson GR, Neff-Smith M, Brock E, Borzelleca J, Welty TK. J Pediatr 2004; 145(5):635-40.*

Varying degrees of structural anomalies, behavioural and neurocognitive disabilities resulting from in utero alcohol exposure characterizes Fetal Alcohol Spectrum Disorder (FASD).¹ Difficulty in diagnosing even full blown Fetal Alcohol Syndrome (FAS) leads to numerous undetected cases.² Diagnosing incomplete FAS or FASD cases presents an even greater diagnostic challenge because often the physical signs are much more subtle or missing.³ Since Jones, Smith and colleagues first described the malformations among fetal alcohol exposed children in detail and provided diagnostic criteria for the condition they termed FAS,^{4,5,6} several thousand articles on the effects of prenatal alcohol exposure among human subjects and on a variety of laboratory animal have been published.⁷ There are two commonly used diagnostic criteria for the evaluation of children potentially affected by fetal alcohol exposure, namely the Institute of Medicine criteria and the Washington criteria,³ however even these are still being altered or clarified.⁸

The specific aim of Kvigne et al was to describe the clinical features, hospitalizations and social service intervention rates of American Indian children with full or incomplete FAS.

The authors compared two retrospective cohorts. Children who were exposed to a noxious influence during pregnancy and breastfeeding using the International Classification of Diseases, Ninth Revision, Clinical Modification code 760.71 were identified from four Northern Plains Indian Health Service Hospitals. Using their medical records, 43 children were diagnosed with FAS based on the following criteria: (1) prenatal alcohol exposure or maternal history of alcohol consumption; (2) FAS diagnosed or a suspected

case noted by a physician; (3) one or more facial features attributed to FAS; (4) any growth deficiency in head circumference, weight or height less than 10th percentile of age group; and (5) central nervous system (CNS) impairment. Thirty-five children with 1 to 4 characteristics were defined as having incomplete FAS. Comparing each group to its separate control group, two individual analyses of data were reported. In addition, FAS and incomplete-FAS groups were compared. Both groups had similar mean age range (4 to 21 years). A matched analysis was performed using corrected McNemar, X² and t tests. In addition, odds ratios and 95% confidence limits were calculated.

Kvigne et al presented growth deficiencies, dysmorphic facial features, CNS dysfunctions, muscular, cardiac and skeletal problems separately. They also looked at rates of hospitalizations for both groups based on the number and days of hospitalization. The rates of Social Service involvement for both groups were based on removal of the children from their home and the rate of placement in foster care. Most of the characteristics reported were not novel as most have been previously published. Common characteristics associated with FAS were seen in both groups.

Growth deficiency, defined as any measurement in height, weight or head circumference below the 10th percentile for age, occurred in a 100% of the children with FAS and occurrence was significantly higher than among children with incomplete FAS. Both groups had significantly higher rates of growth deficiency than their control groups.

The common dysmorphic facial features reported in both studies such as long, flat philtrum, low nasal bridge, short palpebral fissures, thin upper lip, ear malformations, flattened maxilla and epicanthal folds were reported in both FAS and incomplete FAS with roughly the same rate.

The main CNS problems were behavioural problems, developmental delay, speech/language

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delay and microcephaly. These were significantly more prevalent in both groups compared to their controls, but children with FAS had significantly higher rate than children diagnosed with incomplete FAS.

The study is the first to show that children with incomplete FAS display gross and fine motor development delays. Muscular, cardiac and skeletal problems such as trembling, hypotonia and clinodactyly and their occurrence in FAS or incomplete FAS have been previously described by other studies.^{9,10,11}

Children from both groups spent significantly more time in the hospital than did the control children. As supported by previous data these children were commonly hospitalized for otitis media,¹² pneumonia,¹³ failure to thrive¹⁰ and neglect.¹⁴ Feeding difficulties, dehydration, anemia and child sexual abuse in FAS have not been previously reported as a common cause for hospitalization.

The major problem of the study is the criteria used to place the children in the FAS or incomplete FAS group. For diagnosing partial FAS, the Institute of Medicine requires confirmed history of maternal drinking for all children who do not display all three characteristics associated with full-blown FAS (growth retardation, facial dysmorphism and CNS neurodevelopment abnormalities). Kvigne et al only required that the children possess one to four characteristics from the aforementioned criteria and do not explicitly require a history of maternal drinking for incomplete FAS. Without history of maternal drinking, the pathology of the features associated with alcohol fetal exposure may be due to a number of other diseases such as: deLange syndrome, Noonan syndrome, PKU embryopathy, Toluene embryopathy, X-linked mental deficiency or fetal hydantoin syndrome.¹⁵

Clearly, the study is limited by the fact that it is based on medical records and this was noted as a limitation by the authors. However, one might argue that in a retrospective study there is less likelihood of biased diagnosis while looking at each case.

There is little data in this paper that allows further understanding of the characteristics involved in FAS and incomplete FAS as most of the characteristics have been previously reported. However, the comparison between the children

with full FAS and those with incomplete FAS provides some insight on characteristics which are more common in the FAS cases as compared to the incomplete FAS cases. After noting the difficulty of diagnosing FAS and the lack of evidence supporting diagnostic criteria for incomplete FAS, it seems unfortunate that the authors did not make this a larger portion of their discussion.

While most of the information presented has already been established, it strengthens previous studies and deepens the understanding of the severity of FASD. The physical disabilities, large rate of hospitalization and social service intervention display the high individual and societal costs associated with FAS. The authors use the study to justify the need to expand and develop community programs to foster these children throughout their lives.

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