FETAL ALCOHOL SPECTRUM DISORDERS (FASD)

Piyadasa Kodituwakku Comments on Sandra and Joseph Jacobson and Susan Astley on FAS/FAE

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Introduction

Neurodevelopmental disorders are associated with unique patterns of motor, cognitive, and social skills. For example, people with Prader-Willie syndrome display a characteristic pattern of cognitive competencies (e.g., unusual skills with jigsaw puzzles and weakness in auditory processing) and behaviour (e.g., insatiable appetite). It therefore seems reasonable to ask whether there may be a signature neurobehavioural profile associated with prenatal alcohol exposure.

Establishing a unique neurobehavioural profile for people exposed to alcohol in utero would be a tremendous aid in diagnosis, treatment planning and epidemiological research. Epidemiological studies of fetal alcohol syndrome have shown that only one-third of the babies born to mothers who drank heavily during pregnancy presented with full-blown FAS. Moreover, the majority of people who were exposed to alcohol in the womb do not display the classic dysmorphic features of fetal alcohol syndrome, clinicians have had to rely on neurocognitive findings to identify FAS patients. However, research has revealed that impairment of social-cognitive functioning is just as prevalent among FAS children without dysmorphic features it is among those with dysmorphic features.

A. Research Context: Sandra and Joseph Jacobson

Sandra Jacobson and Joseph Jacobson’s overview of neurobehavioural findings in alcohol-exposed children clearly shows that researchers have made significant advances in delineating their cognitive and social-emotional functioning. The authors highlight recent research findings related to three principal areas of functioning: hyperactivity and attention, learning and memory, and social-emotional functioning. They conclude
that alcohol-exposed children display a distinctive pattern of cognitive disabilities with significant difficulties in arithmetic, executive function, acquisition of new information, and social behaviour.

However, although this pattern of difficulties is often found in children with prenatal alcohol exposure, it has not yet been determined whether it is unique to this group. Indeed, children with nonverbal learning disabilities also present with deficient skills in arithmetic, attention, and social skills. Further, the Jacobson overview does not include other research findings that might be used in the development of policies and treatment plans. For one thing, children with prenatal alcohol exposure are deficient in their ability to alter their behavioural responses to changes in reinforcement (affective set shifting) and these deficits predict parent-rated behaviour problems. Even though mildly affected children are not linguistically impaired, those diagnosed with FAS presented with marked deficits in this area. Researchers have reported also deficient performances in tests of visual motor integration, and of visual memory.

Implications for Policy and Provision of Services

A number of the points that have been underscored in the Jacobson overview have significant implications for policy and the provision of services. First, the offspring of mothers who drank recreationally during pregnancy present with patterns of social cognitive deficits similar to those seen in children with FAS. This observation brings us to the question of whether there is a safe threshold for alcohol consumption during pregnancy. Although researchers have gathered evidence from large-scale studies that a threshold might exist, this information is not necessarily generalizable since the deleterious effects of alcohol on the fetus are known to vary depending upon a multitude of factors. Therefore, abstinence from alcohol during pregnancy seems to be the safest course of action.

Second, neuropsychological studies of alcohol-affected children have yielded useful information for school-based interventions. For example, the finding that alcohol-exposed children are deficient in their executive functions indicates that these children have difficulty in guiding goal-directed behaviour using information held in their working memory. Strategies based on external visual guidance may prove to be helpful in the management of their behaviour. Moreover, findings from a prospective study in Seattle indicate that specific variables are associated with better outcomes in alcohol-affected children. These variables include living in a stable and nurturing home, receiving an early diagnosis, and never having been subjected to violence. Findings such as these have substantial implications for early intervention and for the placement of children in stable homes. Since adverse conditions in the family negatively impact on cognitive development in children, family interventions should be held as essential policy and service imperatives, along with individual treatment for alcohol-exposed children.

But because the roots of conduct disorder in alcohol-exposed children are not yet fully understood, further research is required to provide data that is vitally needed for the development of specific interventions for preventing violent behaviours in these children.

B. Research Context: Susan Astley

Jones and Smith introduced the term fetal alcohol syndrome (FAS) to describe a pattern of abnormalities found in a group of children born to alcoholic mothers. The defining characteristics of the syndrome included growth

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retardation, evidence of central nervous system dysfunction, and a characteristic pattern of minor anomalies of the face. In the years following the publication of Jones and Smith’s paper, clinicians found significant variability in the expression of the syndrome, ranging from the classic form to a few minor anomalies. With this discrepancy in mind, Clarren and Smith introduced the diagnostic nomenclature, suspected fetal alcohol effects (FAE), to denote the partial expression of the syndrome. Unfortunately, this diagnosis became an all-purpose label for any behavioural dysfunction in children with suspected histories of prenatal alcohol exposure. In point of fact, one cannot necessarily assume a causal link between alcohol exposure and cognitive dysfunction in these children since the latter is influenced by multiple factors. Thus, FAE became a diagnosis of questionable validity. Furthermore, owing to the inconsistent use of the applicable diagnostic criteria by clinicians, the reliability of diagnosis became a doubly specious proposition.  

Conclusions

The 4-Digit Code approach was introduced to address issues of reliability and validity in diagnosis. The founders of this approach took a number of measures to improve its diagnostic reliability. First, ordinal scales were developed to quantify certain features of diagnostic significance (eg, philtrum). Second, each of the key diagnostic criteria (ie, growth deficiency, FAS facial phenotype, central nervous system dysfunction, and alcohol exposure) was scored on a 4-point scale. Third, each point on these scales was explicitly defined. The full gamut of combinations of the 4-digits generates a total of 256 codes, which may be collapsed into 22 diagnostic categories.

The main achievement of the 4-Digit Code approach has been the improvement of diagnostic reliability through the quantification of key diagnostic features. However, improved reliability does not ensure greater validity. The subtypes of a disorder typically represent distinct categories that must be validated by using multiple criteria (eg, behaviour, physiology, and response to treatment). However, too often these subtypes are treated as arbitrary categories. Meehl calls for a more rigorous application of specific diagnostic criteria: “I see classification as an enterprise that aims to carve nature at its joints (Plato), identifying categories of entities that are non-arbitrary, non-man made.”

Implications for Public Policy and Treatment Planning

I would concur with Astley. An accurate diagnosis of the full spectrum of disabilities caused by alcohol exposure is essential in both primary and secondary prevention efforts. To this end, Astley and Clarren have significantly improved the reliability of diagnosis through the development of quantifiable scales. However, the most challenging problem in the field of fetal alcohol syndrome remains the identification of alcohol-exposed children with no dysmorphia (the majority of alcohol-exposed children). The identification of this group requires the use of both sensitive neuropsychological instruments and controls for confounding variables. There is a growing body of literature supporting the use of narrow band tests (rather than global tests, such as IQ tests) and process data, in addition to scores reflecting end results, when delineating cognitive profiles. Now it remains to be seen whether subtype diagnoses based on data obtained through these kind of sensitive measures will prove more helpful in developing treatment methods than those based on the 4-Digit Codes.

References

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