FAS/FAE: Their Impact on Psychosocial Child Development with a View to Diagnosis

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February 2003

Introduction

Fetal alcohol syndrome (FAS) is a permanent birth defect caused by maternal consumption of alcohol during pregnancy. FAS is characterized by growth deficiency, central nervous system (CNS) dysfunction, and a unique cluster of minor facial anomalies.[1,2] FAS is the leading known cause of mental retardation in the Western World[3] and is entirely preventable. Not all individuals damaged by prenatal alcohol exposure have FAS. Many present with cognitive/behavioral problems, but do not have any growth deficiency or the FAS facial phenotype. These individuals have often been referred to as having fetal alcohol effects (FAE) or alcohol-related neurodevelopmental disorder (ARND).[4]

Subject

Current diagnoses of individuals with prenatal alcohol damage vary widely from physician to physician. While there are diagnostic guidelines[1,2,4,5,6] that physicians are encouraged to follow, these guidelines are not sufficiently specific to ensure diagnostic accuracy (the ability to make the correct diagnosis) or precision (the ability to consistently arrive at the same diagnosis for patients presenting with the same symptoms each time). For example, the guidelines for CNS dysfunction do not address how many areas of deficit must be present, how severe the deficits must be or how much documentation is required to substantiate the presence of the deficit. The guidelines for the facial phenotype do not address how many facial features must be present, how severe each feature must be or what scale of measurement should be used to judge their severity. The use of terms like FAE and ARND fail to address the fact that growth deficiency and CNS dysfunction are not specific to prenatal alcohol exposure.[7] These guidelines reflect a *gestalt* approach to diagnosis that relies more on an overall clinical impression than on data on exposures and outcomes that has been methodically gathered and

interpreted. The key diagnostic features (growth deficiency, facial anomalies, CNS dysfunction, and prenatal alcohol exposure) are not simply present or absent, but rather range along separate continua from normal to severe and present in every possible combination.[8,9] A diagnostic method that better addresses this complexity is therefore needed to improve diagnostic accuracy.

Problems

In the absence of an accurate and reproducible method of diagnosis, diagnoses will continue to vary widely from clinic to clinic.[4,10,11] From a clinical perspective, diagnostic misclassification leads to inappropriate patient care, increased risk for secondary disabilities[12] and missed opportunities for primary prevention.[8] From a public health perspective, diagnostic misclassification leads to inaccurate estimates of incidence and prevalence.[4,8] Inaccurate estimates thwart efforts to allocate sufficient social, educational and health care services to this high-risk population and preclude the accurate assessment of primary prevention intervention efforts. From a clinical research perspective, diagnostic misclassification also reduces our ability to identify clinically meaningful contrasts between groups. Moreover, non-standardized diagnostic methods prevent valid comparisons between studies.

Research Context

To overcome the limitations of the gestalt method of diagnosis, a new, comprehensive method for diagnosing the full continuum of outcomes associated with prenatal alcohol exposure called the 4-Digit Diagnostic Code was developed.[8,9,13,14,15,16] The four digits of the Diagnostic Code reflect the magnitude of expression among the four key diagnostic features of FAS in the following order:

1. growth deficiency
2. FAS facial phenotype
3. brain damage/dysfunction
4. prenatal alcohol exposure.

The 4-Digit Diagnostic Code is generated by first recording key clinical data on the standardized FAS Diagnostic Evaluation Form and following specific case-definitions to generate each digit. The magnitude of expression of each feature is ranked independently on a 4-point Likert scale with “1” reflecting complete absence of the FAS features and “4” reflecting a strong "classic" presence of the FAS features. Each Likert rank is specifically case-defined. The 4-Digit Diagnostic Code may be used to diagnose individuals of all ages.

Key Research Questions

The medical/research records of 1014 patients diagnosed at the Washington State FAS Diagnostic and Prevention Network of clinics[8,15] were used to develop the 4-Digit Diagnostic Code. The performance (ie, the accuracy, reproducibility, and power) of the 4-Digit Code was compared to the gestalt method of diagnosis using the records of the first 454 patients who were diagnosed using both methods.[8]

Recent Research Results

Diagnostic accuracy, reproducibility, and power were found to be substantially greater with the 4?Digit Code than with the gestalt method of diagnosis.[8] Of the 69 patients who received a gestalt FAS diagnosis, only nine met the 4-Digit criteria for FAS. In the absence of specific case-definitions and quantitative measurement scales, the gestalt method of diagnosis produced a very heterogeneous group of individuals with FAS — more heterogeneous than would have been supported by the gestalt guidelines.[6] For example, 37 of the 69 patients had no evidence of growth deficiency, 27 had only one of the three facial features, 29 had no psychometric or structural evidence of brain damage and five had unknown exposure to alcohol. Of the 344 patients who received a gestalt diagnosis of FAE, the outcomes of these patients were even more variable. When reclassified according to the 4-Digit Code, these patients presented with outcomes that spanned 13 different 4-Digit Diagnostic Categories ranging from simply alcohol-exposed to an almost, but not quite full FAS diagnosis. Research studies that treat this diverse group of patients with FAE as one “homogeneous” group are at great risk of failing to identify clinically meaningful contrasts and associations. For example, an important, statistically significant linear association between decreasing IQ with increasing magnitude of expression of the FAS facial phenotype was identified among 216 patients diagnosed using the 4-Digit Code. This association failed to be detected when the same 216 patients were diagnosed using the gestalt method. By contrast, in a preliminary assessment of diagnostic reproducibility, the inter-rater and intra-rater reliability of the 4-Digit Code ranged from 94% to 100%.

Conclusions

The 4-Digit Diagnostic Code has many strengths. It offers an intuitively logical digital approach to reporting outcomes and exposure that reflects the true diversity and continuum of disability associated with prenatal alcohol exposure. It also offers substantially greater precision, accuracy, and power than the gestalt method of diagnosis, through the use of quantitative measurement scales, specific case definitions and a multidisciplinary clinical team approach. One of the cardinal features of the 4-Digit Code is the introduction of a diagnostic nomenclature that replaces terms like FAE and ARND. This new nomenclature clearly documents a patient’s outcomes and exposures without implying that alcohol is the sole causal agent. The facial component of the Code provides an extremely sensitive and specific screening tool for FAS.[16] In addition, the 4-Digit Code establishes a common, descriptive language for more clearly communicating outcomes in medical records and medical literature.

Implications for Policy and Services

Two of the most important goals in FAS studies are primary prevention (preventing the birth of children damaged by alcohol) and secondary prevention (reducing secondary disabilities in children already damaged by prenatal alcohol exposure). These efforts are inextricably linked to our ability to accurately diagnose the full spectrum of fetal alcohol disorders. To measurably prevent FAS, we must first identify women at high risk of giving birth to children damaged by prenatal alcohol exposure. Subsequently, an accurate identification of the incidence of FAS in their children will require accurate diagnostic methods. Likewise, to measure the success of our prevention efforts, we must be able to accurately track changes in the prevalence of FAS over time. This undertaking also requires screening[16] and diagnostic methods that are precise and reproducible over time. Lastly, to measure the effectiveness of interventions targeting children with prenatal alcohol damage, we need to conduct scientifically rigorous studies of children with prenatal alcohol disorders. Once again, the accurate identification of these study populations requires accurate diagnostic tools.

Over 50 multidisciplinary clinical teams across the United States and Canada are now using the 4-Digit Diagnostic Code in a wide array of clinical/social service settings. Much of this expansion is driven by legislative mandates to establish coordinated FAS Diagnostic and Prevention Networks.

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