Perspective

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Fragile X Syndrome and Subjects with Intellectual Disability without Defined Etiology

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Description

Researchers and clinicians have questioned the possibility of a connection between fragile X syndrome (fraX) and autism based on the behavior of people with the condition. Research and discussion have been guided by two issues. To begin, how much of an impact does fraX have on people with autism? Although this connection has been questioned by numerous studies since, a significant proportion of autism cases can be attributed to fraX. There were 19 studies that looked at the prevalence of fraX in boys with autism and 21 studies that looked at the prevalence of fraX in people with mental retardation. Of the more than 1,000 boys in the autism studies, 5.4% were positive for fraX, which is remarkably similar to the 5.5% prevalence of fraX in the mental retardation studies. These findings do not necessarily imply that fraX is not a cause of some cases of autism; however, it is evident that it is not the primary cause.

An X-linked disorder that is inherited is fragile X syndrome. FMRP, a protein thought to be necessary for normal brain function, is methylated or lost when the CGG nucleotide sequence (cytosine-guanine-guanine) on the X chromosome is expanded. Premutation (50-200 CGG repeats, resulting in carrier status but typically not affected), normal (five to fifty CGG repeats), or full mutation (more than 200 CGG repeats, resulting in loss of FMRP and typically mental retardation and associated language and behavioral issues) are the most common classifications for individuals. Due to the fact that boys only have one X chromosome, they typically experience mental retardation ranging from moderate to severe. Females with the full mutation may have learning disabilities or normal intelligence. About a third of people have mild cognitive delays, and some may be affected more severely.

Behavioral Features of Fragile X Syndrome

When young boys with fragile X were evaluated in a child development unit, we developed a checklist to systematically examine certain physical and behavioral characteristics. The checklist was designed to be moderately comprehensive while also being useful in the multidisciplinary setting where several other evaluations would be occurring on the same day. It included items that had been observed in our own patient population as well as items that had been described in previous studies of individuals with fragile X. Because children with fragile X frequently have a fear of doctors, this procedure required that the physical examination be somewhat limited. We didn't want the child's subsequent evaluations to be affected by the physical exam. A behavioral questionnaire, a medical history section, a family history section, and a physical examination section were all included in the checklist. IQs were kept track of. A previous study has described the results of the behavior questionnaire, which found abnormal behaviors in five main areas: The fragile X boys exhibit tactile defensiveness, abnormal language, hand flapping, poor eye contact, and poor self-control (impulsivity). The findings from the physical examination section of the checklist are discussed in this report, as are some aspects of the patient's medical history and family history.

The boy's medical history included information about his history of hernias, allergies, eye problems, ear infections, seizures, spine curvature, and cleft lip or cleft palate. 57/73 parents reported their child's length and weight at birth. Since few parents were aware of the head circumference at birth, this feature was removed. To determine whether the family had a history of "autism," mental retardation, fragile X, or learning disabilities, a family history was gathered. Although the family histories were not always conclusive for X-linked disorders, several families came to our clinic to have their developmentally delayed child evaluated specifically due to a family history of developmental disabilities. Positive family histories of mental retardation, fragile X, or "autism" were all taken into account; however, vague concerns about relatives with developmental disorders were not taken into account.

In general, the results are fairly consistent with those of previous studies that looked at the physical characteristics of children and adults. Knowing these characteristics should help doctors identify fragile X syndrome. Testicular enlargement does not appear to be a very useful clinical characteristic for pediatricians until the child is 8 to 8 years old, which is a clear distinction between the adults and children. It may also be difficult to appreciate verbal perseverance until a child has relatively good language skills. The simian crease did not occur more frequently in this sample.

Correlations with Chronological Age

Males with fragile X frequently exhibit symptoms that can aid in diagnosis, as many adult studies have suggested. However, this study found that only 4 of 42 characteristics were significantly different from controls after multiple comparisons were adjusted for. These included a negative reaction to skin contact (tactile defensiveness), difficulty connecting the tongue to the lips (oral-motor incoordination), soft skin on the hands' dorsum, and a hallucal crease. Three of these items may be helpful to clinicians, despite being somewhat subjective. Ten items had a tendency toward significance, and it is suggested that one of them might play a role in discrimination. Over 80% of males with fragile X also had four additional symptoms, which were also common in the control group. In general, the results are fairly consistent with those of previous studies that looked at the physical characteristics of children and adults. Knowing these characteristics should help doctors identify fragile X syndrome. Testicular enlargement does not appear to be a very useful clinical characteristic for pediatricians until the child is 8 to 8 years old, which is a clear distinction between the adults and children. It may also be difficult to appreciate verbal perseverance until a child has relatively good language skills. The simian crease did not occur more frequently in this sample.

The data were analyzed using several statistical methods. Due to the use of multiple tests and the number of variables, the criterion for statistical significance was a level of alpha of 0.01.