Discovering Fragile X Syndrome: Family experiences, perceptions

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Fragile X syndrome (FXS) is a genetic disorder and the most common known inherited cause of mental retardation and developmental disabilities with an incidence of between 1:2,500 and 1:4,000 births. The disorder is on the X chromosome in both males and females, but males are typically affected more severely. The disorder can be passed for generations in a carrier status with increasing chances of the gene expanding into the full disorder

Unlike Down syndrome, however, FXS is not detectable at birth through physical examination or behavioral observation. It is only through the gradual emergence of delays and behavioral challenges that it becomes apparent to parents and professionals that something is wrong.

We wanted to determine the experiences of families in discovering FXS, factors associated with the timeliness of discovery, and the perceived consequences of obtaining this information.

Parents surveyed reported that males with FXS and born in 1990 or later were identified with FXS at an average age of 31.4 months.

Diagnosis was typically more than 18 months after someone first became concerned about the child's development. Eligibility for special education services occurred earlier than the FXS diagnosis, typically at about age 2 after diagnosis of a developmental delay.

Identification of FXS

The family physician or pediatrician was usually the first person turned to by the family. Nearly half of the respondents said that when they first expressed concern, the physician either affirmed that the child was indeed normal or suggested that the parents "wait and see." Nearly 60 percent said it was somewhat or very difficult to get health care professionals to agree that something was wrong with their child.

Eleven months typically elapsed between professional confirmation of a delay and FXS diagnosis, and more than a third of the respondents said it took more than 10 visits before FXS diagnosis. In most cases, the recommendation for FXS testing did not come from the pediatrician or family physician, which added to the delay.

Family experiences, perceptions

Families generally expressed a combination of relief and distress at the diagnosis. Most said the diagnosis ended their search for a reason for their child's problems and gave important information about reproductive risk that they wished they had known earlier.

Parents experienced challenges, including distress, guilt, worry about the future, and difficulty sharing the information with extended family members. However, most families were glad to find resolution and, despite the challenges, felt the information was essential.

(See "Implications" on reverse)

Summary of article

Researchers used surveys from 274 families who had at least 1 child with fragile X syndrome (FXS).

For families of male children who were born in the last decade, someone first became concerned about the child's development at average age of 13 months.

Professional confirmation of a developmental delay did not occur until an average age of 21 months, and a FXS diagnosis occurred at an average age of nearly 32 months.

Families reported several barriers to discovering FXS and frustration with the process.

Many families had additional children with FXS without knowing reproductive risk.

This *Snapshot* is based on "Discovering Fragile X Syndrome: Family Experiences and Perceptions" by Donald B. Bailey Jr. and Debra Skinner, both of the FPG Child Development Institute, University of North Carolina at Chapel Hill, and Karen L. Sparkman of UNC-Chapel Hill. The article was in *Pediatrics,* Vol. III No. 2 Feb., 2003, pages 407-416.

Implications

(continued from front)

The delays in determining developmental problems and the diagnosis of FXS had real consequences that could have been averted had children been identified at birth.

For example, under Part C of the Individuals With Disabilities Education Act, if these children been identified through newborn screening, they would have been eligible immediately for early intervention services in every state under the "established conditions" mandate, even if they did not at the time show any developmental delay.

Research consistently shows that early intervention can be beneficial for children with a variety of developmental disorders, that families can benefit as well from early intervention and family support services, and that families are generally highly satisfied with existing early intervention programs.

Sufficient evidence exists to make the argument that earlier intervention has the possibility of a direct benefit for child development and family adaptation.

A major consequence is that more than half of the families had additional children without knowing the reproductive risk. Of the 191 children born to these families after the birth of their first child with FXS but before it was diagnosed, 109 (57%) had the full mutation FXS.

A substantial proportion of families, therefore, ended up with 2 children with the disorder, imposing additional care-giving demands and potential stress. Once the diagnosis was made, most families decided against additional children.

If you want to know more

American Academy of Pediatrics, Committee on Children with Disabilities. (2001). Developmental surveillance and screening of infants and young children. *Pediatrics,* 108: 192.

Hagerman R. & Cronister A., (Eds.), (2002). *Fragile X Syndrome: Diagnosis, Treatment, and Research.* 3rd ed. Baltimore, MD: Johns Hopkins University Press.

Bailey, D., Skinner, D., Hatton D., & Roberts J. (2000). Family experiences and factors associated with the diagnosis of fragile X syndrome. *Journal of Developmental & Behavioral Pediatrics, 21*: 315-321.

Roberts J., Hatton D., & Bailey D. (2001). Development and behavior of male toddlers with fragile X syndrome. *Journal of Early Intervention, 24*: 207-223.

FPG's Carolina Fragile X Project: <www.fpg.unc.edu/~FX/>

Conclusions and discussion

We found that parents perceive the discovery of FXS as a process that takes too long, primarily because of their perception that the pediatrician or family physician is reluctant to acknowledge a developmental problem or does not know when to refer children for specific genetic tests.

One solution, of course, involves training of pediatricians and systemic changes in pediatric practice to encourage and allow the kinds of parent-pediatrician interactions that would help earlier identification. Two key components would be to place more credence on the validity of parents' concerns and using regular development screening in pediatric practice instead of developmental surveillance.

This study shows, though, that even if pediatricians responded to every parental concern and used development screening for every infant, the average child's age at first concern would still more than 13 months. If the child were given a developmental assessment immediately without a genetic test but developmental testing did not show a delay substantial enough to meet state criteria for early intervention, then the child and family still would not be eligible for services.

If a delay were detected and a fragile X test ordered immediately, it still likely that a diagnosis would not be made until an average of 18 months of age. Although much better than at present, children would still be missing more than a year of early intervention services and many families would have conceived additional children.

One alternative would be a program of newborn screening for FXS and other disorders as they are discovered. However, this proposal would challenge the public health system and raises a host of ethical questions.

Perhaps our society needs to accept this level of uncertainty and risk.

In the meantime, more research is needed to determine the earliest developmental patterns in FXS and test various models of early intervention (a challenging task for researchers because very few children are identified early enough to conduct these studies).

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