Screening for FX Syndrome

Parent attitudes, perspectives

FPG Snapshot
#11 November 2003

Significance, structure of study

Research on the human genome has spawned a rapid increase in discovery of genes associated with specific disorders. This new genetic knowledge and its associated technology have generated increased consumer interest in and demand for testing, and debate about who should be tested for what and when.

In these debates, relatively little attention has been paid to the desires of parents and other consumers who might be directly affected by screening programs. Fragile X syndrome (FXS), the most common inherited form of mental retardation, exemplifies many issues raised in these debates. FXS results in significant impairments in development and adaptive function. A review of the literature suggests that the number of Caucasian males with the full mutation is about 1 in 4000.

Unlike Down syndrome or spina bifida, the defining features of FXS are not so distinctive as to allow identification at birth. It is only when developmental delays become obvious that parents or physicians become concerned.

FXS is likely to be a prime candidate for inclusion in expanded newborn screening programs and could be a test case for the inclusion of other disorders for which no medical cure or treatment exists.

This study, "Screening for Fragile X Syndrome: Parent Attitudes and Perspectives," is based on 442 surveys from parents of children with FXS regarding different screening options for FXS.

Discussion and implications

On the whole, respondents in this study were optimistic about the benefits of screening and favored voluntary, widespread use of available screening tools. These perspectives place parents somewhat at odds with more conservative approaches to screening endorsed in human genetics and public health. Current screening principles do not promote testing for carrier status of children, for genetic disorders for which no medical cure and proven intervention exists, or for carrier screening for the purpose of informing reproductive decisions.

Traditional principals of newborn screening are currently being challenged due to the availability of molecular genetic analysis and tandem mass spectrometry that allow screening for numerous disorders. As these tests become commercialized and publicized, consumer demands increase for expanded state screening programs.

Proponents argue that early detection provides information and interventions that

Parents' beliefs about possible outcomes of widespread testing and early diagnosis of FSX (442 participants)

Item	Not likely	Somewhat likely	Very likely
Would disrupt bonding wtih the child	66.97%	29.19%	2.26%
Prenatal testing would endanger the baby's health	50.00	40.72	6.56
Would strain relations with family members because of the need to inform them that they might be carriers of FSX	32.13	54.52	12.90
Would result in discrimination by insurance companies	22.17	57.47	16.52
Would increase worries about how others might treat the child or parent	13.80	46.15	38.91
Would increase parents' stress because of decisions they might have to make about having children	9.73	44.12	45.48
Would increase parents' worry about child's future health and development	6.56	37.78	54.98
Would help get services not available without a diagnosis	2.71	18.78	77.38
Would inform planning for additional children	1.36	14.03	84.16
Would allow parents to inform family members about the possibility that they are carriers	0.90	11.09	87.56
Would increase parents' and others' understanding of the child's special needs	0.45	10.86	88.24
Would help parents obtain services earlier for the child	0.23	5.66	93.44
Would help parents gather information to better understand FXS	0.23	5.66	93.67

can improve health and prevent disease, and inform decisions about health and reproduction. Consumers invoke parental rights to have access to any information related to their children's health. This desire to know puts parents at center stage of a debate about who has the right to know what and when.

For many critics, however, the "new genetics" brings the threat of a "new eugenics" where an expansion of prenatal or newborn screening could lead to the elimination of individuals who carry certain genetic disorders or even propensities for disease or disability.

Another concern is that having knowledge of one's own or others' genetic makeup could lead to genetic determinism. Thus, an affected individual's behavior and personality would be seen as the immutable products of biology, eliminating the influence of social and environmental factors in that individual's development.

Other risks include discrimination by insurance companies, workplaces, and other institutions; stigmatization of carriers, affected individuals, and families; and undue worry for parents, among other social and psychological ills.

Even if one has a genetic diagnosis, providing risk assessment for individuals with known genetic disorders, even when caused by mutations within a single gene, is rarely straightforward. Risk assessment involves the multiple components of determining the chances that someone has inherited an altered gene, the chance that this alteration will produce a disorder, and the predicted severity of the symptoms.

Some critics argue that knowing about the genetic condition may endanger the parent-child bond or the child's self-concept. Thus not knowing may provide a protective barrier for the child and family. Furthermore, it is argued that genetic disorders are a family affair that affects extended, past, and future kin. Some relatives would rather not be informed of possible conditions.

These critiques raise important issues that parents may or many not be aware of, or may or may not agree with , but these issues cannot be adequately resolved without the representation and perspectives of families like those in this study who have experienced firsthand the psychological and social ramifications of an inheritable disorder.

If you want to know more

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Results

Timing for genetic testing

Overwhelmingly, respondents agreed or strongly agreed that genetic testing for fragile X syndrome (FXS) should be offered to women before conception (93%), or when behavioral or developmental problems are noticed in the child (95.9%).

Parents were also asked to choose the one best time to offer testing for FXS. The majority chose "before a woman gets pregnant" (79.9%).

Effects on bonding

Parents were asked if they thought a diagnosis of FXS during pregnancy would make bonding more difficult, easier, or have no effect if they had not previously known about the possibility of FXS. Slightly over half (52.9%) believed that the diagnosis would have no effect on bonding, explaining that the child is their child no matter what and would be loved the same as any other child.

Carrier testing

Parents were asked if they would want to be informed, either during pregnancy or after birth, if their child was a carrier of FXS (as opposed to an affected child with the full mutation). Over-whelmingly, parents said "yes," they would like to be informed during pregnancy (86.9%) and immediately after birth (94.3%).

Perceived outcomes of widespread genetic testing (see chart on reverse side)

For the most part, parents thought screening would result in more positive outcomes than negative outcomes. The most likely positive results would be that an early diagnosis would help them locate information about FXS, obtain services earlier, increase understanding of the child's special needs, inform relatives of their possible carrier status, and make informed reproductive decisions.

Parents indicated that the most likely negative outcomes would be increased worry about the child's future health and development, increased parental stress, and worry about how others might treat them or their children.