Fragile X Syndrome

Fragile X is an inherited genetic disorder affecting the X chromosome in both males and females. What is called the *full* mutation leads to fragile X syndrome (FXS), resulting in developmental delays and intellectual disability in almost all males and many females. Carriers of what is referred to as the *pre*mutation are at risk for developing other disorders, primarily fragile X-associated tremor-ataxia syndrome (FXTAS), an adult-onset degenerative neurological condition, and fragile X-associated primary ovarian insufficiency (FXPOI), resulting in reproductive issues including early menopause.

One-third to one-half of all children with FXS have some degree of autistic behavior, and for 2% to 6% of children with *classic* autism (a behavioral diagnosis), the cause of their autism is FXS (a medical diagnosis). It is not uncommon for a child to initially be diagnosed with autism spectrum disorder and later receive an additional diagnosis of FXS, or vice versa.

How Fragile X Is Diagnosed

Fragile X is diagnosed using the FMR1 DNA test (sometimes called the fragile X DNA test). This test detects more than 99% of individuals (both males and females) with FXS (the full-mutation), as well as fragile X carriers (the premutation). The test requires a blood draw and is often covered by insurance, including Medicare and Medicaid. Many clinics offering the test have sliding fee scales.

When Is Fragile X Diagnosed?

In families with a known fragile X history, testing can be done preconception (i.e., of an adult who wants to know if he or she is a carrier), prenatally, or in infancy. However, it is more typically diagnosed in the preschool years or later as delays in development become more pronounced.

Why Diagnosis Is Important

With the help of a genetic counselor, the diagnosis of fragile X can lead to multiple immediate and extended family members learning they may have or are at-risk for developing a fragile X disorder. Diagnosis will lead to specific fragile X evidence and consensus-based treatment including therapies, special education, counseling, and medication. It will also allow families to make the best reproductive decisions for their family; many families have two, three, or more children with FXS. In addition, diagnosis will allow for families to be a part of the National Fragile X Foundation (NFXF) Community Support Network. This foundation has parent-led groups in almost all 50 states and links families to research projects that could potentially lead to better prevention and treatment of fragile X disorders.

Fragile X and Inclusiveness

In an effort to increase the participation of historically underserved populations, NFXF is working to increase awareness about FXS, including the importance of diagnosis. More than 30 fragile X specialty clinics exist in the U.S. to provide expert care and treatment recommendations to individuals and families living with fragile X. In addition, families that attend one of these clinics are invited to participate in a longitudinal research project known as FORWARD and funded by the Centers for Disease Control and Prevention. This project is expanding understanding of FXS while also helping to generate targeted treatments and therapies.

Treatment Recommendations

The professionals who are part of NFXF's Fragile X Clinical & Research Consortium (FXCRC) regularly review and update, as needed, the Fragile X treatment recommendations.

HEAD START CENTER FOR INCLUSION FUNDED BY THE OFFICE OF HEAD START DEPARTMENT OF HEALTH AND HUMAN SERVICES

This resource was supported by the Administration for Children and Families (ACF) of the United States (U.S.) Department of Health and Human Services (HHS) as part of a financial assistance award totaling $10,200,000 with 100% funded by ACF. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by ACF/HHS, or the U.S. Government. This resource may be duplicated for noncommercial uses without permission.