



NATIONAL **FRAGILE X** FOUNDATION
FRAGILE X CLINICAL & RESEARCH CONSORTIUM

Consensus of the Fragile X Clinical & Research Consortium on Clinical Practices

GENETIC COUNSELING AND FAMILY SUPPORT

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fragilex.org/consensus



Genetic Counseling

Genetic counseling is defined by the National Society of Genetic Counselors (NSGC) as the process of “helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.” This process includes interpretation of family and medical histories to assess the chance of occurrence or recurrence of a disorder, education about inheritance, and providing information on testing, management, prevention, resources, research, and counseling to promote informed choices and adaptation to the risk or condition (NSGC 2005).

In the U.S., genetic counselors are usually master’s level medical professionals who have a background in both clinical genetics and psychosocial counseling. Genetic counselors are able to translate complex medical and genetic information and address the emotional and psychosocial issues facing newly diagnosed families. Genetic counselors are trained to assess and assist families coping with a genetic disorder and sharing that information with extended family members. In addition, genetic counselors make referrals to medical and mental health specialists to ensure comprehensive care for individuals and families living with genetic conditions.

Genetic counselors typically work as part of health care teams in a variety of settings. Many genetic counselors partner with medical geneticists, perinatologists, obstetricians, oncologists, and other clinical specialists. Others work in clinical laboratories and research settings, or they may work independently in private practice.

Genetic counseling for fragile X disorders (FXD) is particularly complex due to the multigenerational nature of FMR1 mutations and the variable implications for extended family members, including those who carry a pre- or full mutation. Genetic counseling for FXD is also complicated by the variable clinical presentations associated with pre- and full mutations, and the resulting implications for family members. For these reasons, the genetic counseling process is a complex one and



should be facilitated by a board certified genetic counselor.

Genetic counseling sessions typically include obtaining and analyzing a detailed multigenerational family history, called a pedigree, in order to determine how a gene has been passed down in a family. The pedigree allows the counselor to assess the risk for FXD in individuals as well as extended family members.

Often the first identified family member with an FMR1 mutation is an individual with fragile X syndrome (FXS). Subsequently, additional family members may be identified who could also be FMR1 mutation carriers and possibly at risk for fragile X-associated tremor ataxia syndrome (FXTAS) or fragile X-associated primary ovarian insufficiency (FXPOI), as well as other health implications associated with being an FMR1 carrier. While less common, an individual with FXTAS or FXPOI may be the first identified family member with an FMR1 mutation and may have no known history of intellectual disabilities, autism, or other learning disabilities in the family. Increasingly, FMR1 testing is being offered as a “routine” carrier screening option to pregnant couples and those planning a pregnancy, even when there are no obvious indications in the family history.

In taking a detailed family history, the genetic counselor should inquire about:

- Any individual, male or female, with intellectual, behavioral, and/or learning disabilities, autism spectrum disorder, attention deficit/hyperactivity disorder (ADHD), anxiety, or mental health issues.
- Any female with primary ovarian insufficiency, infertility or irregular menses.
- Any adult with Parkinson’s disease, tremors or other movement disorders, ataxia, cognitive or psychiatric changes, or dementia.

The genetic counseling session should include a detailed discussion of the inheritance pattern of FXD, the clinical presentations of all three conditions (FXS, FXPOI, FXTAS), reproductive options when appropriate, guidance regarding talking to children and extended at-risk family members, considerations for testing asymptomatic children,



research opportunities, family support, and referrals for medical, developmental and psychological providers as indicated.

To assist with the genetic counseling session, the National Society of Genetic Counselors, the National Society of Genetic Counselors (NSGC) has published practice guidelines for counseling individuals and families with FMR1 mutations (<https://link.springer.com/article/10.1007%2Fs10897-012-9524-8>) The NSGC also hosts a consumer website (<http://www.AboutGeneticCounselors.com>) with practical information and a search function that allows users to find certified genetic counselors by location.

Family Support

It can be very difficult for families when a diagnosis of FXS, FXPOI, or FXTAS is first made. Questions often arise, typically “Why me/my family?”, “What do I do now?”, and “Who can I talk to about this?” It is important that families have resources to help answer these questions.

Genetic counselors and other qualified health professionals can be a good source of information regarding the diagnosis, prognosis, and treatment options. They can also help provide psychosocial support to families during this time. Families should not hesitate to get in touch with the clinician or counselor who first gave them information about the diagnosis of fragile X.

Families often find it helpful to talk to providers who are very familiar with the Fragile X disorders. The National Fragile X Foundation (NFXF) has a list of clinics across the country that comprise the Fragile X Clinical and Research Consortium (FXCRC, see <https://fragilex.org/clinics>). These multidisciplinary clinics specialize in fragile X syndrome and associated disorders. The medical professionals at these clinics can provide additional information about the disorders, treatments, and research opportunities.



Given the inherited nature of fragile X-associated disorders, families often require guidance and assistance in providing accurate information to relatives. A genetic counselor or other qualified health professional can help families identify relatives at risk for having FMR1 gene expansions, provide letters and understandable information for family dissemination, and facilitate genetic testing for those interested.

Parents of children with FXS often find it helpful to talk to other parents of children with the condition. Parents may be linked together through their clinicians, or through support groups and organizations. The NFXF's Community Support Network (<https://fragilex.org/community/>) is a network of parent/family group chapters, sponsored by the NFXF, that provide support to families across the country. They also help raise awareness of FXD through education efforts, fundraising, and advocacy. The FRAXA Research Foundation (www.fraxa.org) is another organization with a primary goal of funding research to find a cure for FXS. Families may also be able to connect with others through social networking sites such as Facebook (<http://www.facebook.com/natlfragilex>) and Twitter (<https://twitter.com/nfxfoundation>).

Some families or individuals may have a very difficult time accepting the diagnosis and its implications. Receiving the diagnosis of FXS or a fragile X disorder often leads to feelings of grief and guilt. It is important to remember that everyone grieves differently. However, it is also important to monitor for signs of depression, anxiety, or severe grief reactions. Individual or family counseling may be very helpful for some families coping with the diagnosis of a fragile X disorder.

Additional Resources

The developmental and behavioral issues seen in FXS are often a major concern for parents. Early intervention programs can be helpful resources for parents as they learn how to help their child(ren) achieve early developmental milestones. Once children are in school, therapists can help parents understand their child's language,



sensory, or other developmental difficulties.

The National Fragile X Foundation (NFXF) has a list of available guidelines addressing many of (NFXF) has a list of available guidelines addressing many of these topics, produced by the **Fragile X Clinical & Research Consortium (FXCRC)**, providing expert consensus for the evaluation and treatment of issues common to fragile X (<https://fragilex.org/consensus>).

Another community resource that may be helpful for families is the local ARC (www.thearc.org). The ARC's mission is "to promote and protect the human rights of individuals with intellectual and developmental disabilities and to actively support their full inclusion and participation in the community throughout their lifetimes." It is also important for families to contact their state's department of developmental disabilities, which may be able to provide additional services and support.

The Wrightslaw website (www.wrightslaw.com) provides information regarding special education law and advocacy for children with special needs. Some of the topics include advocacy, ADHD, behavior and discipline, evaluations and more. The NFXF also has educational resources on their website divided into age ranges: birth to age 3, preschool, elementary school, etc.

(<https://fragilex.org/fragile-x-syndrome-resources-age-group/>).

Many families are interested in knowing what research opportunities are available or what is being done to help families affected by FXD. Research is an important part of the Fragile X world, and information about current research studies can be found on the NFXF (<https://fragilex.org/research>) and FRAXA websites. Information about clinical trials for FXD can also be found at ClinicalTrials.gov (www.clinicaltrials.gov).



Author note: This guideline was authored by Liane Abrams, MS, CGC, Sharyn Lincoln, MS, CGC, Brenda Finucane, MS, CGC, and Susan Howell, MS. CGC and reviewed and edited by consortium members both within and external to its Clinical Practices Committee. It has been approved by and represents the current consensus of the members of the Fragile X Clinical & Research Consortium.

The Fragile X Clinical & Research Consortium was founded in 2006 and exists to improve the delivery of clinical services to families impacted by Fragile X and to develop a research infrastructure for advancing the development and implementation of new and improved treatments. Please contact the **National Fragile X Foundation** for more information.

(800-688-8765 or **fragilex.org**)