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 disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources and 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 trained professionals who have a background in both clinical genetics and psychological counseling. Genetic counselors often provide the link between medical information and the emotional and psychosocial issues facing newly diagnosed families. Genetic counselors have training in assessing, intervening and assisting families with the complexities of coping with a genetic disorder and sharing that information with extended family members. In addition, genetic counselors can make referrals to other mental health providers in the community if the family’s psychosocial needs extend beyond the scope of the genetic counselor’s time, skills and breadth.

Genetic counselors work in a variety of settings, including partnering with medical geneticists, perinatologists and obstetricians, oncologists, and other clinical specialists, clinical laboratories, or working independently.

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, the genetic counseling process is a complex one and should be facilitated by a genetic counselor. Genetic counseling for Fragile X-associated Disorders (FXD) is complicated by the variable phenotypes of the pre and full mutations and the implications for family members.

Genetic counseling sessions typically include obtaining and analyzing a detailed multigenerational family history. It is often through the analysis of the family history that a counselor is able to gain more information about the patient and to assess which family members may also be at risk for fragile X syndrome or its associated disorders.

The proband, or first identified family member with the FMR1 mutation, is often the individual with fragile X syndrome (FXS) and can sometimes direct attention to family members who may be at risk for fragile X-associated tremor ataxia syndrome (FXTAS) or fragile X-associated primary ovarian insufficiency (FXPOI). However, the proband may be an individual with FXTAS
or FXPOI. The number of probands who present with FXPOI or FXTAS is on the rise, and very often these individuals have no known history of intellectual disabilities, autism, or other learning disabilities in the family.

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 disorders, anxiety or mental health issues
- Any female with primary ovarian insufficiency, infertility, irregular menses
- Any adult with Parkinson’s disease or other movement disorders, tremors, ataxia, cognitive or psychiatric changes, or dementia

The genetic counseling session will 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 and testing asymptomatic children, research opportunities, family support and referrals.

To assist with the genetic counseling session, the National Society of Genetic Counselors has recently published practice guidelines for counseling individuals and families with FMR1 mutations (http://www.fragilex.org/wp-content/uploads/2012/10/FMR1-2012.pdf) The website also has a “Find a Counselor” feature that allows the user to identify genetic counselors close to a specified (i.e. patient’s) 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.

Physicians and genetic counselors 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 crisis 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-associated Disorders. The National Fragile X Foundation (NFXF) has a list of clinics across the country that make up the Fragile X Clinical and Research Consortium (FXCRC, see http://www.fragilex.org/treatment-intervention/fragile-x-clinics/). These multidisciplinary clinics specialize in fragile X syndrome and its associated disorders. The medical professionals at
Genetic Counseling and Family Support

these clinics can provide additional information about the disorders, treatments, and research opportunities.

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. Linking Individuals Nationally in Knowledge and Support (LINKS) (http://www.fragilex.org/community/links-support-network/) is a network of parent/family groups, 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 that provides information to families and helps to raise awareness of FXS. Families may also be able to connect with others through social networking sites such as Facebook (http://www.facebook.com/natlfragilex) and Twitter (http://twitter.com/fragilexnews). 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-related 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.

The developmental and behavioral issues seen in FXS are often a major concern for parents. Early intervention can be a helpful resource 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. 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. (currently under development).

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
Genetic Counseling and Family Support

world, and information about current research studies can be found on the NFXF (http://research.fragilex.org) 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 and Sharyn Lincoln, 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.

Funding: This project was made possible by Cooperative Agreement U01DD000231 from the Centers for Disease Control and Prevention to the Association of University Centers on Disabilities (AUCD) and RTOI 2008-999-03 from AUCD to W.T. Brown in support of the National Fragile X Clinical and Research Consortium. The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

*The Fragile X Clinical & Research Consortium was founded in 2006 and exists to improve the delivery of clinical services to families impacted by any Fragile X-associated Disorder 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 www.fragilex.org)*