NATIONAL FRAGILE X FOUNDATION

GENETIC COUNSELORS

WHO ARE GENETIC COUNSELORS?

A **genetic counselor** is a master's level medical professional who is specially trained in genetics. They coordinate genetic testing and provide counseling and support to patients and their families through a diagnosis of a genetic condition like Fragile X.

Fragile X and other genetic conditions can be complex. The role of a genetic counselor is to help individuals and families understand the risks and implications.

Genetic counselors are trained to do the following as needed:

- Explain the benefits and limitations of genetic testing to help families make informed decisions about whether to proceed with genetic testing.
- Facilitate ordering genetic testing for the *FMR1* gene.
- Review genetic testing results and explain the diagnosis, if one is found.
- Explain how genetic conditions can be passed down in families.
- Help identify family members who may be at risk, referred to as cascade testing.
- Discuss and explain the potential symptoms and characteristics of Fragile X-associated conditions.
- Explain potential reproductive challenges and options.
- Recommend additional medical specialists for patients, when needed.
- Provide resources, educational materials, and connections to support options such as groups, therapists, and patient advocacy organizations.

Genetic counselors often work alongside geneticists and healthcare providers you may already be seeing. Genetic counselors are also available at Fragile X clinics. If you are not already connected to a genetic counselor, please visit findageneticcounselor.nsgc.org to find one near you.

WHICH TYPE OF GENETIC COUNSELOR SHOULD I SEE?

A **prenatal** genetic counselor can help discuss the benefits and limitations of carrier screening (for parents) and prenatal testing (for a current pregnancy). If needed or desired, they can help arrange further testing, such as a CVS or amniocentesis.

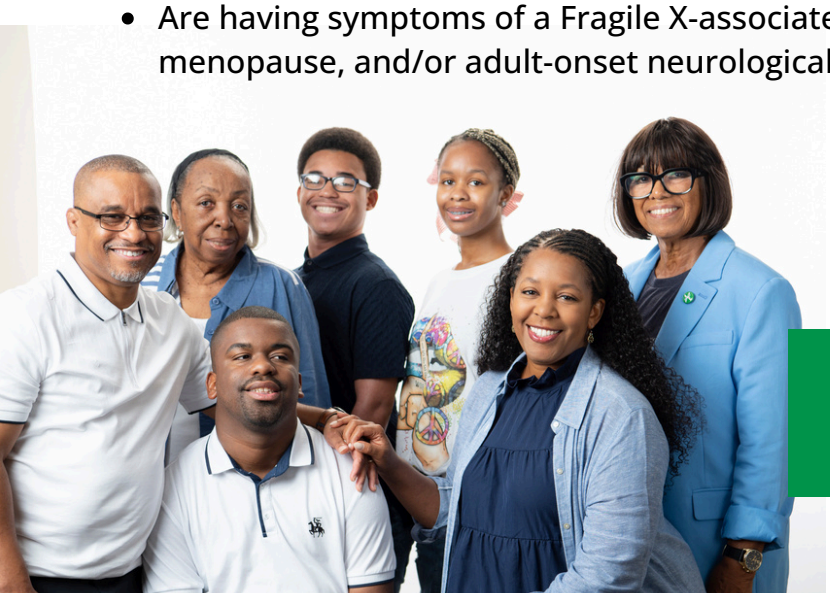
A **pediatric** genetic counselor can help diagnose Fragile X in a child and provide support to the child and their family once a diagnosis has been confirmed.

An **adult** genetics or medical genetics counselor can help an adult experiencing symptoms related to Fragile X-associated conditions, such as FXTAS or FXPOI. They can provide counseling to evaluate the risk and potential causes of these symptoms, including the Fragile X premutation.

WHEN SHOULD I CONSULT WITH A GENETIC COUNSELOR?

Consider speaking to a genetic counselor if you:

- Are planning a pregnancy and are interested in knowing options for carrier screening.
- Have already been identified as having positive, inconclusive, or unusual Fragile X testing results.
- Are currently pregnant and would like to learn more about prenatal diagnostic testing options.
- Have a family member who has been diagnosed with Fragile X syndrome, a Fragile X-associated condition, or has been identified as having a Fragile X premutation, commonly referred to as a “Fragile X carrier”.
- Have a personal or family history of autism, any undiagnosed intellectual disabilities, or learning challenges.
- Are having symptoms of a Fragile X-associated condition such as infertility, early menopause, and/or adult-onset neurological or movement disorders.



Want to know more?

- fragilex.org
- contact@fragilex.org