



# Genetic Counseling: Who, What & How

## Who Should Seek Out a Genetic Counselor?

Anyone who has:

- Been identified as being a “Fragile X carrier” or has positive, inconclusive, or in any way unusual [Fragile X testing](#) results.
- A family member who has been identified as having the Fragile X [premutation](#). (sometimes called a “carrier”).
- A child or family member diagnosed with [Fragile X syndrome](#) or any of the [Fragile X-associated disorders](#).
- A family or personal history of undiagnosed intellectual disabilities, autism, or learning disorders.
- [Symptoms](#) of any of the Fragile X-associated disorders, including infertility, early menopause, or adult onset neurological or movement disorders.

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## What Does a Genetic Counselor Do?

A genetic counselor is a trained, master's level medical professional who works with individuals and families to:

- Review genetic testing and diagnoses.
- Review inheritance patterns.
- Help identify possible carriers in the family.
- Discuss the symptoms and features of Fragile X-associated disorders.
- When appropriate, review [reproductive issues and options](#).
- Make referrals and provide professional and emotional support.

Due to the complexity of Fragile X testing, the counselor is essential in the interpretation of results and for providing accurate information regarding Fragile X.

## What to Expect

The counselor:

- Will ask questions regarding the medical or developmental/learning history of your family members (including children, parents, brothers and sisters, aunts and uncles, cousins, nieces and nephews, grandparents).
- Will review the genetic implications of the diagnosis or test results.
- May identify other family members who may have inherited the gene mutation.
- May make suggestions or assist with informing or contacting family members regarding testing.
- May meet with you either independently or in conjunction with a medical geneticist or other provider, who will likely examine your child or affected family member. If the counseling will be focused on carrier testing, an exam is often not indicated.
- Can work with you and other family members to coordinate testing throughout the family.
- Can provide information and help with decision-making regarding reproductive issues and options and coordinate any further testing.
- Can make referrals to other providers for medical evaluations and care, prenatal care and testing, therapies for your child, family counseling, or whatever needs you have at the time.

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## How to Find a Genetic Counselor

We recommend making an appointment at your nearest [Fragile X clinic](#). Fragile X clinics provide genetic counseling and medical services (including medication evaluations and consultations) supervised by a physician and supported by the latest medical, educational, and research knowledge available. Multidisciplinary services, such as occupational, speech, language, and behavioral therapies, are also available either at the clinic or by referral.

Alternatively, ask your doctor or provider for a referral to a local genetic counselor.

You can also use the “[find a counselor](#)” feature on the [National Society of Genetic Counselors](#) website, or contact us at [contact@fragilex.org](mailto:contact@fragilex.org) or by phone at (800) 688-8765.

Genetic counselors can work either independently or as part of a medical team. Some common settings where genetic counselors work are:

- Medical genetics departments in a hospital or other medical center setting.
- Prenatal, perinatal, or infertility care providers.
- Specialist medical groups such as oncology, hematology, neurology, cardiology.
- Multidisciplinary clinics such as craniofacial, skeletal dysplasia,
- [Fragile X clinics](#).
- Private practice.
- Public and community health settings.
- Genetics laboratories.
- Support organizations (such as us, the [NFXF](#)).

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## Genetic Testing & Autism

According to the [latest CDC findings](#), 1 in 59 children in the U.S. were diagnosed with autism by age 8 in 2014, up 15% from the previous year. Of those, Fragile X

syndrome accounts for an [estimated 1% to 6% of all cases of autism](#) — and among the genetic causes is the most common known inherited single-gene disorder. However, some children are diagnosed with autism before they are diagnosed with Fragile X.

Given the possibility of a link, Fragile X expert Dr. Randi Hagerman recommends that all children with autism, both male and female, be referred for genetic evaluation and testing for FXS and any other genetic cause of autism.

[Also see Understanding Fragile X Syndrome & Autism](#)

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## Related Treatment Recommendations

### Genetic Counseling and Family Support

Genetic counseling includes family and medical histories to assess the chance of carriers (premutations) throughout a family's lineage.

[Go to the treatment recommendation](#)

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