



INFORMATION ABOUT FRAGILE X SYNDROME

Fragile X syndrome is the most common form of inherited mental retardation. A problem with a specific gene (called the FMR1 gene) causes the disease. Normally, the gene makes a protein needed for brain development, but the mutation causes a person to make little or none of the protein, which results in the symptoms of fragile X.

People with only a small change in the gene might not show any signs of fragile X. People with bigger changes can have severe symptoms. These might include:

- Intelligence problems, ranging from learning disabilities to severe mental retardation
- Social and emotional problems, such as aggression in boys or shyness in girls
- Speech and language problems, especially in boys

Fragile X syndrome has no cure. You can treat some symptoms with educational, behavioral or physical therapy, and with medicines. Early treatment for fragile X can help.

Source: The National Institutes of Health

FRAGILE X SYNDROME CARRIER SCREENING

What is carrier screening?

The American College of Obstetricians and Gynecologists recommends that couples planning a pregnancy, or those already pregnant, be informed about genetic carrier screening. Specific ethnic groups are known to have an increased risk for certain genetic conditions, and carrier screening is available to find out if a couple is at risk for having a child with one of these conditions. Carrier screening for other conditions, including fragile X syndrome, is available for couples with a concerning family history.

How is fragile X syndrome inherited?

Fragile X syndrome is found among all ethnic backgrounds and racial groups. Approximately 1 in 260 women in the general population are carriers of the abnormal gene that causes fragile X syndrome.

Fragile X syndrome follows an X-linked inheritance pattern. The gene mutation involves the triplet repeat, CGG, of variable length within the FMR1 gene. A "fragile X carrier" is someone who has an abnormal FMR1 gene repeat number (premutation), but does not show any obvious symptoms of fragile X syndrome. The premutation can expand considerably when passed from the mother to a child. If the premutation expands to a full mutation, it could result in fragile X syndrome. Almost all males and approximately half of females with the full mutation have significant mental retardation. Children of women with either the premutation or the full mutation have a 50% chance of inheriting the expanded gene. It is estimated that 1/2000 males and 1/4000 females have the full mutation.

The inheritance of fragile X syndrome is very complicated. We would be available to meet with you for a genetics consultation to discuss your family history as well as the benefits and limitations of carrier screening for fragile X syndrome.

Who should be screened?

- Individuals with a family history of fragile X syndrome
- Individuals with a family history of mental retardation, developmental delay, or autism of unknown cause
- Women with developmental disabilities who are pregnant or planning to become pregnant
- Women with infertility problems associated with elevated follicle stimulating hormone (FSH) levels, premature ovarian failure (POF) or unexplained early menopause

When should carrier screening be performed?

It is strongly recommended that you undergo fragile X carrier screening prior to pregnancy, or as early in pregnancy as possible.

What is the carrier screening process?

- Genetic counseling is necessary prior to fragile X carrier screening, as the inheritance of fragile X syndrome is complicated.
- Carrier screening is performed through a simple blood test; no preparation is necessary.
- Test results are provided to you and your physician within 2-3 weeks.
- If you are found to be a carrier of fragile X syndrome, additional genetic counseling will be available to discuss the nature of the specific disorder as well as prenatal testing options.

UNDERSTANDING YOUR RESULTS

Test result	What the test result means for the mother	What the test result means for the pregnancy
“Negative”	You are not a carrier for the most common alteration in the FMR1 gene.	The pregnancy is not at increased risk for fragile X syndrome.
“Intermediate”	Your results fall in the range between negative and premutation.	The pregnancy is not at increased risk for fragile X syndrome. Future generations may be at risk for fragile X syndrome.
“Premutation”	You are a carrier for the altered FMR1 gene. You may be at risk for early menopause.	The pregnancy is at risk for fragile X syndrome. Prenatal diagnostic testing should be considered.
“Full mutation”	You are a carrier for the altered FMR1 gene. You may be at risk for early menopause.	The pregnancy is at risk for fragile X syndrome. Prenatal diagnostic testing should be considered.

QUESTIONS?

If you have any questions concerning the above information, please do not hesitate to contact us at 312-981-4400.