NxGen MDx

Fragile X and *FMR1*-Related Disorders

Fragile X Intermediate Carrier

What Your Results Mean

Test results indicate that you are an intermediate allele carrier of fragile X syndrome. This means that you are **not** at an increased risk to have a child with fragile X syndrome; however, your family members (siblings or offspring) may be at an increased risk to be premutation allele carriers of fragile X syndrome. Intermediate allele carriers do not have an increased risk for developing *FMR1*-related conditions.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

As a female carrier, there is a 50% chance that you will pass your intermediate allele to your offspring. Your offspring are not at risk to have fragile X syndrome but may be an intermediate or premutation carrier. Carrier testing of your male partner or donor is usually not necessary for conditions that are carried on the X chromosome, such as fragile X syndrome, because the result would not change the testing strategy and/or medical management of future pregnancies and children. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

Fragile X Premutation Carrier

What Your Results Mean

Test results indicate that you are a premutation allele carrier of fragile X syndrome. This means that you are at an increased risk to have a child with fragile X syndrome. As a female carrier, there is a 50% chance that you will pass your premutation allele to your offspring. Your offspring that inherit the allele will either be premutation carriers, like yourself, or the allele will expand, and they will have a diagnosis of fragile X syndrome. Premutation allele carriers are at an increased risk for *FMR1*-related conditions.

We recommend that you share and discuss this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

For individuals who are carriers of a premutation with 55-90 CGG repeats, additional testing known as AGG interruption analysis can further refine the risk to have a child with fragile X syndrome. Testing embryos for fragile X syndrome, or diagnostic prenatal testing via chorionic villus sampling (CVS) or amniocentesis may be considered.

Carrier testing of your male partner or donor is usually not necessary for conditions that are carried on the X chromosome, such as fragile X syndrome, because the result would not change the testing strategy and/or medical management of future pregnancies and children. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

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Fragile X Syndrome Explained

What is Fragile X Syndrome?

Fragile X syndrome is a condition associated with intellectual disabilities and cognitive impairment. Typically, males are more severely affected than females. Affected individuals have developmental delays, mild-to-moderate intellectual disabilities, and behavioral differences such as hyperactivity, anxiety, and autism spectrum disorders. Most males, and approximately half of affected females, have characteristic physical features including a long and narrow face, large ears, a prominent jaw and forehead, flat feet, and unusually flexible fingers.

What are *FMR1*-Related Conditions?

Individuals who are carriers of a premutation allele are also at risk to develop *FMR1*-related conditions such as fragile X tremor ataxia syndrome (FXTAS) and/or primary ovarian insufficiency (POI).

Fragile X Tremor Ataxia Syndrome (FXTAS)

Both male and female premutation carriers (those with 55-200 CGG repeats) are at risk to develop a late onset (after age 50) neurological condition known as FXTAS. Individuals with FXTAS can develop trouble walking, leading to increased tendency to fall and subsequent use of walking aids. On average, male premutation carriers over age 50 have a 40% chance of developing FXTAS. The risk for female premutation carriers to develop FXTAS is not well-defined; however, females seem to be at a lower risk of developing FXTAS than males.

Fragile X-Associated Primary Ovarian Insufficiency (POI)

Females who are premutation carriers are at increased risk of POI (early menopause and reduced fertility). Approximately 1% of women in the general population develop POI, while premutation carriers have an up to 21% chance of developing POI. It is important to note that 5% to 10% of women with POI are able to conceive after receiving a diagnosis of POI.

Prognosis

Males who have fragile X syndrome typically have developmental delays, intellectual disabilities, and may have autism spectrum disorders. Affected females may have a wider spectrum of features that vary in severity, but females are still at risk for significant intellectual involvement and autism spectrum disorders. Fragile X syndrome does not typically affect lifespan.

Treatment

Treatment for fragile X syndrome is supportive as there is no cure. Early intervention, special education classes, and medication to treat behavioral issues may be indicated.

Resources

Centers for Disease Control and Prevention
https://www.cdc.gov/ncbddd/fxs/index.html
The National Fragile X Foundation
https://fragilex.org
National Society of Genetic Counselors
https://www.nsgc.org/

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