Mucolipidosis IV

What Your Results Mean

Test results indicate that you are a carrier of mucolipidosis IV. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of mucolipidosis IV for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

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

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for mucolipidosis IV, each of your children has a 1 in 4 (25%) chance to have the condition.

Mucolipidosis IV Explained

What is Mucolipidosis IV?

Mucolipidosis IV is an inherited metabolic disorder in which harmful amounts of fats and proteins accumulate within the recycling compartments (lysosomes) of cells in the body. Over time, this excessive build-up of fats and lipids within the lysosomes causes developmental delay, visual impairment, and other signs of the disease.

There are two forms of mucolipidosis IV. In the most common form, typical mucolipidosis IV, developmental delays are noted in the first year of life. Maximum development achieved is typically 18 months of age. Neurodegeneration is seen in approximately 15% of individuals with typical mucolipidosis IV. By the teenage years, severe vision loss or blindness caused by corneal clouding or progressive retinal degeneration is present in most individuals. Additional characteristics of the disease include impaired stomach acid production causing elevated gastrin in the blood, as well as iron deficiency that can lead to anemia. Individuals with the milder form, atypical mucolipidosis IV, have milder delays and eye abnormalities than those with typical mucolipidosis IV.

Prognosis

Treatment of individuals with mucolipidosis IV typically includes supportive therapies (speech therapy, physical therapy, and ankle-foot orthotics) and iron supplementation for iron deficiency anemia.

Treatment

Prognosis is generally poor. Most affected individuals are never able to walk and nearly all individuals will develop severe vision loss by their teens. Life expectancy may be shortened, but individuals with mucolipidosis IV typically live into adulthood.

Resources

Mucolipidosis type IV Foundation
http://ml4.org/

Genetic and Rare Diseases
https://rarediseases.info.nih.gov/diseases/94/mucolipidosis-type-4

National Society of Genetic Counselors
https://www.nsgc.org/

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