

Mucopolipidosis IV

What Your Results Mean

Test results indicate that you are a carrier of mucopolipidosis IV. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of mucopolipidosis 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 mucopolipidosis IV, each of your children has a 1 in 4 (25%) chance to have the condition.

Mucopolipidosis IV Explained

What is Mucopolipidosis IV?

Mucopolipidosis 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 mucopolipidosis IV. In the most common form, typical mucopolipidosis 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 mucopolipidosis 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 mucopolipidosis IV, have milder delays and eye abnormalities than those with typical mucopolipidosis IV.

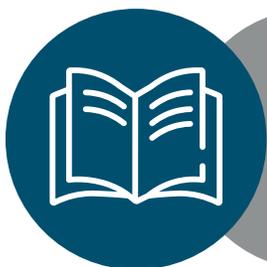


Prognosis

Treatment of individuals with mucopolipidosis 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 mucopolipidosis IV typically live into adulthood.



Resources

Mucopolipidosis type IV Foundation

<http://ml4.org/>

Genetic and Rare Diseases

<https://rarediseases.info.nih.gov/diseases/94/mucopolipidosis-type-4>

National Society of Genetic Counselors

<https://www.nsgc.org/>