NxGen MDx

Mucopolysaccharidosis Type I

What Your Results Mean

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

Mucopolysaccharidosis Type I Explained

What is Mucopolysaccharidosis Type I?

Mucopolysaccharidosis type I, also known as Hurler syndrome, is an inherited lysosomal storage disorder in which sugar molecules accumulate in the body and can lead to organ damage. Individuals with mucopolysaccharidosis type I may appear normal at birth and typically develop symptoms within the first two years of life. The symptoms can include vision issues and possible blindness due to cloudy corneas, heart valve issues, narrowed arteries, lung disease with frequent infections, and cognitive impairment.

Prognosis

Prognosis is typically poor. The average age of the onset of symptoms is 10 months, and individuals usually die approximately eight years after symptom onset.

Treatment

There is no cure for mucopolysaccharidosis type I, but enzyme replacement therapy is available. Enzyme replacement therapy can lead to functional improvements; however, the neurological complications of the disorder do not typically improve with enzyme replacement therapy. Bone marrow transplant is possible and can help prolong lifespan. Treatment is otherwise symptomatic.

Resources

National MPS Society
https://mpssociety.org/

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/mucopolysaccharidosis-type-i/

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

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