Alpha-Mannosidosis

What Your Results Mean

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

Alpha-Mannosidosis Explained

What is Alpha-Mannosidosis?

Alpha-mannosidosis is an autosomal recessive lysosomal storage disorder caused by a deficiency of the enzyme alpha-mannosidase. Without this enzyme, carbohydrates accumulate and cause cell death and tissue damage in the body. This disorder is characterized by intellectual disability, coarse facial features, skeletal abnormalities, hearing impairment, neurologic motor problems, and immune deficiency. It is important to note that the severity of signs and symptoms varies amongst individuals with a diagnosis of alpha-mannosidosis.

Type 1 alpha-mannosidosis is the mildest form, often diagnosed after the age of 10 in an individual. These individuals typically have mild or barely detectable symptoms that progress slowly and typically do not have skeletal abnormalities.

Type 2 alpha-mannosidosis is a moderate form, often diagnosed before the age of 10. Individuals with this type can have both skeletal abnormalities and muscle weakness.

Type 3 alpha-mannosidosis is the most severe form and is typically fatal in childhood. The disease rapidly progresses in these individuals, and the central nervous system is involved. Symptoms show up in infancy and some affected pregnancies may not survive to term.

Prognosis

Prognosis is dependent on the severity and onset of signs and symptoms. Individuals with type 1 alpha-mannosidosis typically do not survive past childhood. If a case is very severe, pregnancies affected with alpha-mannosidosis may not survive before birth. Individuals with type 2 or type 3 can survive up to their fifth decade of life. Sometimes a diagnosis of alpha-mannosidosis can only be detected through testing and those individuals may have little to no symptoms at all.

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Treatment

Treatment for alpha-mannosidosis is symptomatic. For example, antibiotics may be needed for bacterial infections or the individual may need supportive devices such as a hearing aid. Early intervention can be beneficial for children with alpha-mannosidosis to reach their highest potential. Bone marrow or stem cell transplantation has been noted to increase the chance of preventing cognitive decline and improving the overall symptoms of an individual.

Resources

National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/alpha-mannosidosis/
National Society of Genetic Counselors
https://www.nsgc.org/

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