

Tay-Sachs Disease

What Your Results Mean

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

Tay-Sachs Disease Explained

What is Tay-Sachs Disease?

Tay-Sachs disease is an inherited metabolic disorder that progressively destroys nerve cells in the brain and spinal cord. Over time, this can cause permanent cellular and tissue damage, particularly in the spleen, liver, bone marrow, and rarely, the brain. Individuals with Tay-Sachs disease typically have normal development in the first few months of life followed by regression of developmental milestones within the first year. With time, decreasing visual attentiveness, unusual eye movements, seizures, and progressive enlargement of the head become apparent. An eye abnormality called a cherry-red spot, which can be identified with an eye examination, is characteristic of this disorder.



Prognosis

Prognosis for Tay-Sachs disease is poor, as affected individuals typically die by the age of four.

Treatment

There is no cure for Tay-Sachs disease. Treatment is mostly supportive and directed to providing adequate nutrition and hydration, managing infectious disease, protecting the airway, and controlling seizures.



Resources

National Tay-Sachs and Allied Diseases Association

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

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/tay-sachs-disease>

National Society of Genetic Counselors

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