Genetics Uncoded:

Facts about Tay-Sachs Disease

What Your Test Results Mean

Carriers typically show no symptoms of Tay-Sachs disease; however, carriers are at an increased risk of having a child with Tay-Sachs disease. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Tay-Sachs Disease Explained

Tay-Sachs disease is an inherited metabolic disorder in which harmful amounts of a glycosphingolipid called GM2 ganglioside accumulate within lysosomes of cells. Individuals with Tay-sachs disease do not produce enough of the enzyme -hexosaminidase A, which is needed to break down GM2 ganglioside. Over time, this excessive storage in the lysosomes can cause permanent cellular and tissue damage, particularly in the spleen, liver, bone marrow, and rarely, the brain.

While juvenile, chronic, and adult-onset forms of hexosaminidase A deficiency have been described, Tay-Sachs disease is the most severe form of hexosaminidase A deficiency. Individuals with Tay-Sachs 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. Life expectancy is typically between two and four years.

Treatment is mostly supportive and directed to providing adequate nutrition and hydration, managing infectious disease, protecting the airway, and controlling seizures.

How the Genetics Work

Tay-Sachs disease is an autosomal recessive disorder caused by pathogenic variants in the *HEXA* gene. In general, individuals have two copies of the *HEXA* gene. Carriers of Tay-Sachs disease have a single variant in one copy of the *HEXA* gene while individuals with Tay-Sachs disease have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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