



One-Day Tay-Sachs DNA Test

Tay-Sachs disease (also known as GM2-gangliosidosis type I) is a fatal, autosomal recessive, neurodegenerative disorder. It is characterized by normal motor development in the first few months of life followed by progressive weakness and loss of motor skills at 2-6 months, decreased social interaction, seizures, blindness, and total debilitation. Death typically occurs between 2 and 5 years of age. Tay-Sachs disease is caused by excess storage of GM2 ganglioside in lysosomes due to a deficiency of the isoenzyme β -hexosaminidase (hex A).

Mutations in the HEXA gene are responsible for Tay-Sachs disease. The carrier frequency for these mutations varies with ethnicity. The carrier frequency in Ashkenazi Jewish individuals is approximately 1 in 30. Three mutations in the HEXA gene account for approximately 98% of all Tay-Sachs mutations found in the Ashkenazi Jewish population: 1278insTATC (81%), 1421+1G→C (15%), and G805A (2%). The overall carrier frequency for Tay-Sachs disease in non-Jewish individuals is 1 in 300. However, in select populations such as French Canadian and Cajun, the carrier frequency is reported to be increased.

The American College of Obstetrics and Gynecology (ACOG) has recommended that individuals of Ashkenazi Jewish, French-Canadian, or Cajun descent be screened for Tay-Sachs disease. In addition, Tay-Sachs DNA testing is recommended for individuals with ambiguous or positive hex A enzyme test results.

Indications for Testing

- Individuals of Ashkenazi Jewish, French Canadian, or Cajun descent who are planning a pregnancy or who are currently pregnant
- Reproductive partners of known Tay-Sachs carriers
- Family history of Tay-Sachs disease

Special Aspects of our Service

- Rapid turnaround time (1 day)
- Detailed reports with genetic interpretation, recommendations, and education
- Genetic consultation by board-certified genetic counselors and geneticists

Please call Kimball Genetics for more information

Tay-Sachs DNA Testing Service

- Specimen requirements:
5 ml whole blood in a lavender top tube (EDTA)
or
Cheek cell sample (free collection kits provided)
- Turnaround time: 1 day

Additional Tay-Sachs Testing Services (available upon request)

- Hex A enzyme testing
- DNA testing for additional mutations and pseudodeficiency alleles
- Prenatal diagnosis

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