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CARRIER SCREEN

Tay-Sachs disease

What is Tay-Sachs disease?

Tay-Sachs disease is an inherited disease characterized by progressive degeneration of the central nervous system and death, occurring typically in early childhood. The symptoms of Tay-Sachs disease are due to a defect in the enzyme beta-hexosaminidase A, which is responsible for breaking down a fatty substance in the body called GM2 ganglioside. Without this enzyme, GM2 accumulates primarily in the brain and nerve cells, causing severe damage. Tay-Sachs disease is also known as hexosaminidase A deficiency.[1]

What are the symptoms of Tay-Sachs disease and what treatment is available?

Tay-Sachs disease is a progressive condition that is typically evident in infancy between three and six months of age and results in early childhood death.[1] Although less common, juvenile and adult forms of Tay-Sachs disease occur and are associated with later onset, slower disease progression, and variable neurological findings.[2]

Symptoms of infantile Tay-Sachs disease may include: [1,2]

- Progressive muscle weakness
- Loss of motor skills (rolling, sitting, crawling)
- Exaggerated startle response to loud noise
- Reduced attentiveness
- Spasticity (tight muscles)
- Cherry-red spot seen on eye examination
- Vision and hearing loss
- Seizures
- Severe intellectual disability

Symptoms of adult-onset TSD may include: [2]

- Loss of muscle mass
- Muscle weakness
- Loss of motor skills (walking)
- Speech problems
- Dementia
- Mental illness

There is no cure for Tay-Sachs disease. Treatment is supportive.[2]

How is Tay-Sachs disease inherited?

Tay-Sachs disease is an autosomal recessive disease caused by mutations in the *HEXA* gene.[1] An individual who inherits one copy of a *HEXA* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *HEXA* mutations, one from each parent, is expected to be affected with Tay-Sachs disease .

If both members of a couple are carriers of a mutation in the same gene, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Tay-Sachs disease?

Tay-Sachs disease can occur in individuals of any race or ethnicity; however, it is most common in individuals of Ashkenazi Jewish and French-Canadian ancestry.

Carrier frequency in select populations

Population	Carrier frequency
Ashkenazi Jewish	1/27[3]
French Canadian	1/73[4]
General	1/300[5]

Having a relative who is a carrier or who is affected can also increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

National Tay-Sachs and Allied Diseases: http://www.ntsad.org/

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/tay-sachs-disease

References

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3. Scott S, *et al*. Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. *Hum Mut*. 2010; 31: 1-11.
4. Martin DC, *et al*. Evaluation of the risk for Tay-Sachs disease in individuals of French Canadian ancestry living in New England. *Clin Chem*. 2007; 53(3): 392-398.
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