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My46 Trait Profile

Tay-Sachs disease

Other Names: Acute infantile hexosaminidase A deficiency, Type I GM2 gangliosidosis

Tay-Sachs disease is a genetic condition characterized by the body's inability to break down a fatty substance called GM2 ganglioside. As a result, the GM2 ganglioside builds up in the brain and spinal cord. It is caused by mutations in the HEXA gene which makes the hexosaminidase subunit A protein.

Characteristics of Tay-Sachs disease

Tay-Sachs disease is a neurodegenerative condition characterized by progressive deterioration, weakness, loss of movement and cognitive decline. Children with Tay-Sachs disease typically begin showing symptoms of the condition around three to six months of age. Tay-Sachs disease is caused by an inability to break down a fatty substance called GM2 ganglioside, allowing it to build up in the brain, and cause the physical and intellectual disabilities seen in the condition. With continual damage to the brain and spinal cord, blindness, seizures, loss of muscle tone, and eventual death result. A child with Tay-Sachs disease usually does not live past the age of five or six. Tay-Sachs disease is more common in the Ashkenazi Jewish, French Canadian, Louisiana Cajun, and Irish American populations. Approximately 1 in 30 Ashkenazi Jews is a carrier of a Tay-Sachs disease, while in the non-Jewish population, about 1 in 300 individuals are carriers.

Diagnosis/Testing

Tay-Sachs disease can be diagnosed either by enzyme testing or by genetic testing for a change or mutation in a gene called HEXA. This gene encodes a protein that is part of the beta-hexosaminidase A enzyme. This enzyme plays an important role in the brain and spinal cord, and helps to break down a fatty substance called GM2 ganglioside. Mutations in the HEXA gene result in deficiency of the beta-hexosaminidase A enzyme, thus allowing GM2 ganglioside to build up in the brain and spinal cord, and cause the features seen in the condition.

Not all mutations in the HEXA gene cause Tay-Sachs disease, also referred to as acute infantile hexosamindase A deficiency. Certain mutations in this gene allow some beta-hexosaminidase A enzyme to be made, and thus are associated with juvenile, chronic, and adult-onset variations of hexosaminidase A deficiency. Compared to classic Tay-Sachs disease, these forms are usually milder, symptoms usually appear in later childhood, adolescence or adulthood and include weakness, ataxia (i.e., loss of muscle coordination), mental illness, and psychiatric disturbances.

Management/Surveillance

Management of children with Tay-Sachs disease typically includes providing supportive care, adequate nutrition and hydration, and medication to control seizures.

Mode of inheritance

Tay-Sachs is inherited in an autosomal recessive pattern. This means that an individual has to inherit two HEXA mutations (i.e., one from each parent) to be affected with Tay-Sachs disease. If both parents are carriers of a HEXA mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with Tay-Sachs disease.

Risk to family members

Parents of a child with Tay-Sachs disease are carriers of Tay-Sachs disease. If a sibling of a child with Tay-Sachs disease is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of Tay-Sachs disease.

Special considerations

None

Resources

National Tay-Sachs and Allied Diseases Association, Inc.
http://www.ntsad.org
Genetics Home Reference: Tay-Sachs disease
http://ghr.nlm.nih.gov/condition/tay-sachs-disease

References

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