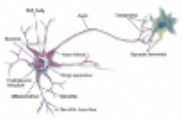
TAY-SACHS DISEASE

By: Vincent

Overview



A rare, inherited disorder that destroys nerve cells in the brain and spinal cord. They are typically found in a certain ancestry which are the Easten European Jews.

Causes

It occurs when a child inherits a mutation in the HEXA gene from both parents.

Symptoms

- "Cherry-red" spots in the eyes.
- Loss of motor skills
- Vision loss and blindness.
- Seizures

Current Treatment

- **There is no specific treatment for the Tay-Sachs Disease**
- **Anticonvulsant medicine may control seizures.**
- **Proper nutrition and hydration and techniques to keep the airway open.**
- **Children may eventually need a feeding tube.**

Prognosis

- Rapidly progressive
- Affected children typically do not survive past early childhood.

Prevention

- There is nothing you can do to prevent this disease
- However, genetic counselling and screening can be crucial

Tay-Sachs disease is a disorder that damages nerve cells in the brain and the spinal cord. It is passed through from parents to children and it is caused by the absence of an enzyme. This enzyme helps break down fatty substances, or gangliosides, that build up toxic levels in the brain and spinal cord. The most common form of Tay-Sachs disease is infantile Tay-Sachs disease, where infants with this disorder develop normally up to 3 to 6 months of age. Over time, their development decelerates, and muscles used for movement deteriorate. Affected infants cease to meet normal development stages and begin to lose skills previously attained such as rolling over, sitting up, and crawling. Many infants with this disorder develop an exaggerated startle reaction to loud noises. Children with Tay-Sachs disease become cognitively impaired as their disease advances and develop other involuntary spasms of the muscles, seizures, swallowing problems, deafness and blindness, and intellectual disability. Most children with infantile Tay-Sachs disease die in early childhood.

Moving on, there are also two unusual forms of Tay-Sachs disease called juvenile and late-onset. The juvenile form shows symptoms in late childhood. Signs of late-onset Tay-Sachs generally start in adulthood. People who have either of these types of the disorder tend to have milder and more variable signs and symptoms relative to those with the infantile form. The features of the juvenile or late-onset form of Tay-Sachs disease are muscle weakness, loss of muscle coordination, speech problems, and psychiatric symptoms. For the majority, the signs and symptoms thus broadly vary among those with late-onset forms of Tay-Sachs disease.

Unfortunately, there is no possible cure for Tay-Sachs disease. The treatment provided is mostly focused on symptom alleviation, and supportive care, significantly creating a better quality of life for affected children. There are medications to alleviate the symptoms. Seizure management is crucial as the anticonvulsant medications used are indicated to reduce the frequency and severity of seizures. Also, physical therapy is required to maintain muscle function which can be helpful in delaying the development of muscle weakness and improvement of mobility. Furthermore, respiratory care, which consists of chest physiotherapy for keeping the lungs clear of mucus and it makes a huge difference in preventing infections. It also stops one from coughing up fumes from the lungs, ultimately improving breathing. When the illness has reached the point at which swallowing becomes difficult, feeding tubes are necessary to provide adequate nutrition. All of these methods are just ways to help lighten the symptoms and impacts.

While there is no prevention from getting the disease, there are some methods to help make someone infected with Tay-Sachs disease healthcare decisions. Genetic screening, for example, should be conducted among higher-risk groups, such as the Ashkenazi Jews. Since they are more prone to the disease, knowing their status as a carrier will help in making informed reproduction decisions. If both parents are found to be carriers, prenatal diagnosis by chorionic villus sampling or amniocentesis can be carried out early in pregnancy, which allows parents to know if an affected child is on the way and to decide whether or not to begin or continue a pregnancy. By applying these strategies, individuals and families will be in a good position to understand health decisions and manage their condition better if it arises, thereby improving the outcome and quality of life of individuals affected by Tay-Sachs disease. The prognosis for Tay-Sachs Disease is poor; most children born with this disorder do not usually survive beyond early childhood until 4 to 5 years. The

rapid rate by which it progresses unmistakably points out the need for continuous research and support to families with these gene disorders.

The video "Understanding Tay-Sachs Disease" provided a detailed overview of Tay-Sachs disease, particularly highlighting its four stages and the profound effects on patients and their families. I was particularly surprised to learn that there were actually four stages instead of three and it was interesting to learn about the progression of the disease through its distinct stages, each bringing its own set of challenges and symptoms. What makes Tay-Sachs such a terrible disease is that the symptoms are detrimental and typically leads to someone infected with a lifespan of 4 years. This rapid and fatal progression underscores the need for awareness and support in order to find research and combat this deadly disease. It was also interesting how the disease has a 50-50 for boys and girls since typically diseases are usually more likely to fall on boys and it was something new that I had learned. The video also was quite similar to my report because there is no treatment, and the best approach is the supportive care to manage symptoms, improving the quality of life of those affected. These would include multidisciplinary care by neurologists, physical therapists, and palliative care specialists who may be necessary in taking charge of the complex needs of patients.