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Short Note on Tay Sachs Disease

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Perspective

Tay-Sachs disease is a rare genetic disorder that is passed down from father to son. It is caused by the lack of an enzyme that aids in the breakdown of fatty substances. These fatty substances, known as gangliosides, accumulate to toxic levels in the brain and spinal cord, impairing nerve cell function. Signs and symptoms of the most common and severe form of Tay-Sachs disease appear between the ages of 3 and 6 months. As the disease progresses, growth slows and muscles weaken. Seizures, vision and hearing loss, paralysis, and other serious problems develop as a result of this.

Children with this form of Tay-Sachs disease have a short lifespan. Some children have the juvenile form of Tay-Sachs disease and may live into their adolescence. Some adults may have a late-onset form of Tay-Sachs disease, which is often milder than forms that begin in childhood. If you have a family history of Tay-Sachs disease or are a member of a high-risk group and want to have children, your doctor will strongly advise you to get genetic testing and genetic counselling. In the most common and severe form, called infantile form, an infant typically begins showing signs and symptoms by about 3 to 6 months of age. Survival is usually only a few years. Exaggerated startle response when the baby hears loud noises,"Cherry-red" spots in the eyes, Loss of motor skills, including turning over, crawling and sitting up, Muscle weakness, progressing to paralysis, Movement problems, Seizures

Tay-Sachs disease is a genetic disorder that is passed from parents to their children. It occurs when a child inherits a flaw (mutation) in the HEXA gene from both parents. The genetic change that causes Tay-Sachs disease results in a deficiency of the enzyme beta-hexosaminidase A. This enzyme is required to break down the fatty substance GM2 ganglioside. The buildup of fatty substances damages nerve cells in the brain and spinal cord. Severity and age of onset of the disease relates to how much enzyme is still produced. To determine whether your child has Tay-Sachs disease, your doctor will ask about symptoms and any family hereditary disorders, as well as perform a physical exam. For nervous system and eye examinations, your child may need to see a neurologist and an ophthalmologist.

A blood test for diagnosis. The blood test measures the amount of the enzyme hexosaminidase A in the blood. Tay-Sachs disease has low or non-existent levels. Genetic analysis. This test can look at the HEXA gene to see if there are any changes that could indicate Tay-Sachs disease. Examine your eyes. During an eye exam, the doctor may notice a cherry-red spot in the back of the eyes, which is an indication of the disease. Tay-Sachs disease can be tested for during pregnancy by removing a tiny piece of the placenta (chorionic villi sampling) or a small sample of the amniotic fluid surrounding the baby (amniocentesis) [1-5].

There is no cure for Tay-Sachs disease, and no treatments have been shown to slow the disease's progression. Some treatments can aid in the management of symptoms and the prevention of complications. The treatment's goal is to provide support and comfort. There are a variety of prescription medications available to alleviate symptoms and prevent complications, such as anti-seizure medications or antibiotics for infection. Care for the lungs. Accumulated mucus in the lungs is common and increases the risk of lung infections that cause breathing difficulties. Chest physiotherapy (CPT), exercise, and other methods can aid in the removal of mucus from the lungs. To reduce the risk of mucus accumulation and prevent aspiration pneumonia, medications that reduce saliva production and positioning techniques are also options.

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