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Short Note on Lysosomal Storage Disorder

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Introduction

A rare genetic illness called Tay-Sachs is passed from father to son. It is brought on by a deficiency in an enzyme that helps break down fatty compounds. These fatty compounds, known as gangliosides, build up in the brain and spinal cord to hazardous levels, affecting nerve cell function. The most prevalent and severe form of Tay-Sachs disease first exhibits symptoms between the ages of 3 and 6 months. Growth slows down and muscles become weaker as the condition worsens. This leads to seizures, eyesight and hearing loss, paralysis, and other catastrophic issues [1].

Description

The longevity of children with this variant of Tay-Sachs illness is brief. Some young patients with Tay-Sachs disease's juvenile form may survive until puberty. Tay-Sachs disease can develop late in life in some individuals, and these cases are frequently milder than childhood-onset cases. Your doctor would strongly encourage you to seek genetic testing and genetic counselling if you wish to have children and you belong to a high-risk group or have a family history of Tay-Sachs disease. Infants often start exhibiting signs and symptoms of the most prevalent and severe kind, known as the infantile form, by the time they are 3 to 6 months old. The average lifespan is only a few years. When a baby hears loud noises, they react startledly and exhibit "Cherry-red" patches [2].

A genetic ailment called Tay-Sachs is passed from one generation of parents to the next. When a child inherits a defect (mutation) in the HEXA gene from both parents, the condition takes place. The enzyme beta-hexosaminidase A is deficient as a result of the genetic mutation that causes Tay-Sachs disease. The fatty compound GM2 ganglioside must be broken down in order for this enzyme to function. The brain and spinal cord's nerve cells are harmed by the accumulation of fatty substances) [3]. How much enzyme is still produced is related to the severity and age of the disease's development. Your doctor will conduct a physical examination, inquire about your child's medical history, look for any hereditary conditions in the family, and then make a diagnosis of Tay-Sachs disease. Your youngster may need to visit a doctor for an eye exam and a nervous system checkup) [4].

A blood test to determine the cause. Hexosaminidase A levels in the blood are determined via a blood test. There is little to no Tay-Sachs disease. genetic examination. The HEXA gene can be examined with this test to check for any mutations that can point to Tay-Sachs disease. Look at your eyes. A cherry-red patch in the rear of the eyes, a sign of the condition, may be seen by the doctor during an eye checkup. During pregnancy, a small sample of the amniotic fluid around the foetus or a little portion of the placenta can be taken to test for Tay-Sachs disease (amniocentesis) [5].

Conclusion

There is currently no known treatment for Tay-Sachs disease, and neither is there a cure. Some medical procedures can help with symptom control and problems avoidance. The aim of the treatment is to offer consolation and support. To treat symptoms and avoid consequences, a number of prescription drugs are available, such as antibiotics for infections or anti-seizure drugs. look after your lungs. It is typical for the lungs to accumulate mucus, which raises the danger of lung infections that make breathing difficult. Exercise, chest physiotherapy (CPT), and other techniques can help clear mucus from the lungs. Medication that decreases saliva production and posture strategies are other options to lower the risk of mucus collection and avoid aspiration pneumonia.

Acknowledgement

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Conflict of Interest

None.

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