Tay-Sachs Disease

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Key Points
➢ What is tay-sachs disease ➢ Prognosis of tay- sach disease ➢ Causes of tay- sach disease ➢ Sign and symptoms tay-sach disease

Tay-Sachs's disease belongs to the group of autosomal-recessive lysosomal storage metabolic disorders.[1] In this disease there is a damaging of neurons, ultimately causing the damage of nervous cells in the brain and spinal cord. This resultantly causes the patient's inability to move, loss of hearing and seizures; patients with this disease usually die at the ages 3 to 5.[1]

Causes

This disease is caused by the genetic mutation in the HEXA gene on chromosome 15, encoding for a subunit of hexosaminidase enzyme called the hexosaminidase A. This causes the disintegration of the enzyme GM2 ganglioside within the cells, causing its accumulation.[1]

Signs & Symptoms

In most cases, the disease manifests itself during infancy, the "infantile form," which characterizes the most severe disorders of the nervous system.[2]The infants at this stage are shown to make sudden noises and startled responses. In this stage (infants) there is delayed response and repressed mental and physical abilities. The patient may become blind, deaf, showing paralysis or having the inability to swallow. Death occurs at around age 4 years.[2]

The Juvenile stage is quite rare, it starts in the ages 2 to 10 year old. The patient is a target of motor skills deterioration and death occurs at 15.[2]

References:

1. Solovyeva VV, Shaimardanova AA, Chulpanova DS, Kitaeva KV, Chakrabarti L, Rizvanov AA. New approaches to Tay-Sachs disease therapy. Frontiers in physiology. 2018:1663.
2. Solovyeva VV, Shaimardanova AA, Chulpanova DS, Kitaeva KV, Chakrabarti L, Rizvanov AA. New approaches to Tay-Sachs disease therapy. Frontiers in physiology. 2018:1663.