Sandhoff Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Sandhoff Disease Information Page.

Sandhoff disease is a rare, inherited lipid storage disorder that progressively destroys nerve cells in the brain and spinal cord. It is caused by a deficiency of the enzyme beta-hexosaminidase, which results in the harmful accumulation of certain fats (lipids) in the brain and other organs of the body. Sandhoff disease is a severe form of Tay-Sachs disease, the incidence of which had been particularly high in people of Eastern European and Ashkenazi Jewish descent, but Sandhoff disease is not limited to any ethnic group. Each parent must carry the defective gene and pass it on to the child. Individuals who carry only one copy of the mutated gene typically do not show signs and symptoms of the disorder. Onset of the disorder usually occurs at 6 months of age. Symptoms may include:

- progressive nervous system deterioration,
- problems initiating and controlling muscles and movement,
- increased startle reaction to sound,
- early blindness,
- seizures,
- spasticity (non-voluntary and awkward movement),
- myoclonus (shock-like contractions of a muscle,
- macrocephaly (an abnormally enlarged head),
- cherry-red spots in the eyes,
- frequent respiratory infections,
- doll-like facial appearance, and
- enlarged liver and spleen.