Tay-Sachs Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Tay-Sachs Disease Information Page.

Tay-Sachs disease is an inherited metabolic disease caused by the harmful buildup of lipids (fatty materials such as oils and acids) in various cells and tissues in the body. It is part of a group of genetic disorders called the GM_2 gangliosidoses. Tay-Sachs and its variant form are caused by a deficiency in the enzyme hexosaminidase A. Affected children appear to develop normally until about age 6 months and then begin to show symptoms, including:

- progressive loss of mental ability,
- dementia,
- blindness,
- increased startle reflex to noise,
- progressive loss of hearing leading to deafness,
- difficulty with swallowing,
- seizures that may begin in the child's second year, and
- "cherry-red" spots in their eyes.

A much rarer form of the disorder, called late-onset Tay-Sachs disease, occurs in individuals in their twenties and early thirties and is characterized by an unsteady gait and progressive neurological deterioration.

The incidence of Tay-Sachs has been particularly high among people of Eastern European and Askhenazi Jewish descent., as well as in certain French Canadians and Louisiana Cajuns. Affected individuals and carriers of Tay-Sachs disease can be identified by a blood test that measures hexosaminidase A activity. Both parents must carry the mutated gene in order to have an affected child. In these instances, there is a 25 percent chance with each pregnancy that the child will be affected with Tay-Sachs disease. Prenatal diagnosis is available if desired. A very severe form of Tay-Sachs disease is

known as Sandhoff disease, which is not limited to any ethnic group.