Sandhoff Disease

Sandhoff disease is rare. Sandhoff disease is genetic. It is caused by a lack of the enzyme *hexosaminidase*. This results in the collection of certain fats (*lipids*) in the brain and other organs of the body. It results in the progressive deterioration of the central nervous system. Sandhoff disease is not limited to any ethnic group. The disorder usually occurs at 6 months of age.

SYMPTOMS

- Motor weakness.
- Startled reaction to sound.
- Early blindness.
- Progressive mental deterioration.
- Progressive motor deterioration.
- Frequent respiratory infections.
- An abnormally enlarged head (*macrocephaly*).
- Doll-like facial appearance.
- Cherry-red spots in the back of the eyes.
- Seizures.
- Shock-like contractions of a muscle (*myoclonus*).

TREATMENT

There is no specific treatment for Sandhoff disease. Treatment is symptomatic and supportive.

WHAT WILL HAPPEN

The prognosis for individuals with Sandhoff disease is poor. Death usually occurs by age 3. Death is generally caused by respiratory infections.

RESEARCH BEING DONE

The National Institute of Neurological Disorders and Stroke (NINDS) does research on genetic disorders such as Sandhoff disease. The goals of this research are to understand, prevent, treat, and, ultimately, cure them. Scientists have had some limited success in studies using the mouse model of this disorder by inhibiting the formation of the accumulating lipid. The animals fare better but the treatment is still far from life-saving.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

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