Tay-Sachs Disease

March of Dimes Resource Center

What is Tay-Sachs disease?

Tay-Sachs disease is an inherited disease of the central nervous system. The most common form of the disease affects babies and is fatal. Affected babies appear healthy at birth and seem to develop normally for the first few months of life. After this time, development slows and symptoms begin. Sadly, there is no effective treatment for these babies.

Babies with Tay-Sachs disease lack an enzyme (protein) called hexosaminidase A (hex A) necessary for breaking down certain fatty substances in cells of the brain. These substances build up and gradually destroy brain cells' ability to function, until the entire central nervous system stops working.

Symptoms of classic (infantile) Tay-Sachs disease first appear at 4 to 6 months of age when an apparently healthy baby gradually stops smiling, crawling or turning over, loses ability to grasp or reach out and, eventually, becomes blind, paralyzed and unaware of surroundings. Death occurs by age 5, and often sooner.

Who is at risk of Tay-Sachs disease?

Tay-Sachs disease occurs most frequently in descendants of Central and Eastern European (Ashkenazi) Jews. About one out of every 30 American Jews carries the Tay-Sachs gene. Some non-Jewish individuals of French-Canadian ancestry (from the East St. Lawrence River Valley of Quebec), and members of the Cajun population in Louisiana, are at similarly increased risk. These groups have about 100 times the rate of occurrence of other ethnic groups. The juvenile form (see below) of Tay-Sachs, however, may not be increased in these groups.

Are there other forms of Tay-Sachs disease besides the classical type that affects babies?

The classical infantile type of Tay-Sachs is the most common. However, there are other rare deficiencies of the hex A enzyme that sometimes are included under the name of Tay-Sachs disease. These often are referred to as juvenile and late-onset forms of Tay-Sachs disease. Affected individuals have very low levels of the hex A enzyme that is missing entirely in the classic, infantile form.

This fact may help explain why symptoms begin later in life and, generally, are milder than in the classic, infantile Tay-Sachs disease.

Children with juvenile Tay-Sachs disease develop symptoms between 2 and 10 years of age that resemble those of the classic, infantile form. Although the course of the disease is slower, death generally occurs by age 15.

Individuals with late-onset Tay-Sachs disease (also called chronic Tay-Sachs disease) have far milder symptoms than children with the infantile or juvenile forms. Symptoms usually begin between adolescence and the mid-30s, although they can begin in childhood. Vision and hearing are unaffected in this form of the disease and, in most cases, mental abilities remain intact. However, some individuals may have some loss of mental abilities, including problems with memory and comprehension. Symptoms vary greatly in severity and can include slurred speech, muscle weakness, muscle cramps, tremors, unsteady gait and, sometimes, mental illness. Life expectancy appears to be unaffected.

How is the disease transmitted?

All forms of Tay-Sachs disease are inherited. The disease is passed on through parents who carry the Tay-Sachs genes in their cells. A Tay-Sachs carrier has one normal gene for hex A and one Tay-Sachs gene. The carrier does not have the illness. However, when two carriers become parents:

- There is a 25 percent (one-in-four) chance that any child they have will inherit a Tay-Sachs gene from each parent and have the disease.
- There is a 25 percent chance (one-in-four) that the child will inherit the normal gene from each parent, and so be completely free of the disease and the Tay-Sachs gene.
- There is a 50 percent (two-in-four) chance that the child will inherit one of each kind of gene and be a carrier like the parents.

If only one parent is a carrier, none of that person's children can inherit the disease, but each child has a 50-50 chance of inheriting the Tay-Sachs gene and being a carrier.

Is there any treatment for Tay-Sachs?

Tragically, there is no treatment that will prevent the disease from running its course. Affected children can only be made as comfortable as possible.

Can Tay-Sachs disease be diagnosed before birth?

Yes. Prenatal tests called amniocentesis and chorionic villus sampling (CVS) can diagnose Tay-Sachs before birth. In amniocentesis, which usually is done between the 15th and 18th weeks of pregnancy, a needle is inserted into the mother's abdomen to take a sample of fluid that surrounds the fetus. The fluid contains fetal cells that can be examined for the presence of hex A. In CVS, the doctor retrieves a sample of cells either through a thin tube inserted through the vagina and cervix to the placenta or by inserting a needle through the mother's abdomen. The placenta contains cells that nearly always are genetically identical to those of the fetus, and these cells are examined for the presence of hex A. CVS usually is done between the 10th and 12th weeks of pregnancy.

If prenatal testing shows that hex A is present, the baby will not have infantile Tay-Sachs. If it is missing, he or she will be affected. In unusual cases, DNA-based genetic testing can determine whether the fetus has infantile Tay-Sachs or another form of the disease and, possibly, how severely affected the child will be.

A few medical centers have begun offering carrier couples in vitro fertilization (in which eggs are removed from a woman's ovaries and fertilized in the laboratory with her partner's sperm), with genetic testing of the embryos, so that only healthy ones are implanted in the mother. However, this approach is still considered experimental.

How can people find out if they are carriers?

An individual can take a test that measures the amount of hex A in his or her blood. Tay-Sachs carriers have about half as much of the enzyme as noncarriers, but this is plenty for the carrier's own needs.

A blood sample also can be used to perform DNA-based genetic testing. These are tests that look for known mutations (changes) in the hex A gene that cause the various forms of Tay-Sachs. This kind of testing may be recommended if the results of the usual carrier screening test discussed above are uncertain.

Where is carrier screening available?

Carrier screening for Tay-Sachs is available from a genetic services center or clinic. A physician can provide referrals to local sites where testing is available, as can the National Tay-Sachs and Allied Diseases Association.

The genetic services center performs the carrier screening test that can determine whether one or both partners carry the Tay-Sachs gene. Trained genetic counselors will explain the test results, so that individuals know whether or not their children will be at risk of the disease.

What research on Tay-Sachs disease is being conducted by March of Dimes grantees?

March of Dimes-sponsored researchers have helped pinpoint mutations in the hex A gene that are responsible for rare forms of Tay-Sachs disease. Information about specific mutations leads to improved diagnosis and carrier screening for all forms of Tay-Sachs.

Currently, a grantee is attempting to develop a drug treatment that may prevent the production of certain fatty substances that build up and impair brain cells in affected individuals. This approach eventually may help prevent the early deaths and loss of central nervous system function associated with the disease.

The March of Dimes also supports research on gene therapy, by which scientists seek to repair or replace an abnormal or missing gene. The goal is to cure or alleviate the symptoms of certain genetic diseases, possibly including Tay-Sachs disease.

How can someone learn more about Tay-Sachs?

Families affected by Tay-Sachs disease can contact The National Tay-Sachs and Allied Diseases Association, Inc. The Association is involved in research and educational programs related to the prevention and treatment of Tay-Sachs, provides families with referrals to medical resources, and also coordinates a parent support network.

Contact the Association at:
The National Tay-Sachs and
Allied Diseases Association, Inc.
2001 Beacon St., Suite 204
Brookline, MA 02135
Telephone: 1-800-906-8723
E-mail: ntsad-boston@worldnet.att.net
www.ntsad.org

Additional information also is available from:
Late Onset Tay-Sachs Foundation
1303 Paper Mill Road
Erdenheim, PA 19038
1-800-672-2022
215-836-9426 (in PA)
E-mail: mpf@bellatlantic.net
www.lotsf.org

References

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