

WHAT IS TAY-SACHS DISEASE?

A baby with Tay-Sachs disease* appears healthy at birth, and seems to be developing normally for a few months. Symptoms generally appear by six months of age. While symptoms vary from one child to the next, there is always a slowing down of development. Gradually, Tay-Sachs children lose motor skills and mental functions. Over time, the child becomes blind, deaf, mentally retarded, paralyzed and nonresponsive to the environment. Tay-Sachs children usually die by age five.

Children with Tay-Sachs disease lack a vital enzyme, hexosaminidase A (Hex-A). Hex-A is needed for the body to break down a fatty waste substance found in brain cells. Without Hex-A, this substance accumulates abnormally and causes progressive damage until the nervous system can no longer sustain life.

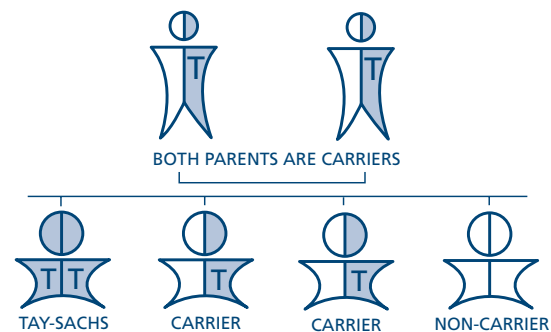
Why is a baby born with Tay-Sachs disease?

Tay-Sachs is an inherited disease that only occurs when both parents carry a Tay-Sachs gene and each parent transmits the defective gene to their child. A child who inherits two Tay-Sachs genes (one from each parent) produces no functional Hex-A enzyme and is certain to develop Tay-Sachs disease.

A person with only one Tay-Sachs gene is perfectly healthy, but is a Tay-Sachs carrier. When both parents are carriers, there is a 1 in 4 (25%) chance, with every pregnancy, of having a child with Tay-Sachs disease.

When both parents are carriers, there is a 2 in 4 (50%) chance, with every pregnancy, of having a child who is a carrier.

When only one parent is a carrier, there is no chance the child will have Tay-Sachs disease. There is a 2 in 4 (50%) chance, with every pregnancy, of having a child who is a Tay-Sachs carrier.



Every person of Jewish, Irish, French-Canadian or Cajun heritage should be tested for Tay-Sachs

non-Jewish population, the carrier rate is about one in 250. There are certain exceptions. French-Canadians and the Cajun community of Louisiana have the same carrier rate as Ashkenazi Jews, one in 27. Also, individuals with ancestry from Ireland are at increased risk for the Tay-Sachs gene. Current research indicates that among Irish Americans, the carrier rate is about one in 50.

All couples planning to have children should carefully consider their ancestry to evaluate the risk of each partner. Any person who can trace his or her lineage to a high-risk population must be tested. In addition, close relatives of carriers (children, sisters, brothers, cousins, aunts, uncles) must be tested since they may also be carriers.



How do you know if you're a Tay-Sachs carrier?

The answer is a simple blood test.

The Tay-Sachs blood test, referred to as carrier screening, identifies Tay-Sachs carriers and non-carriers. It is urgent to understand that the Tay-Sachs gene gets passed from one generation to the next. Without carrier screening,

it can remain hidden in a family for decades, surfacing unexpectedly and tragically with the birth of an affected child. The fact that there is no family history of Tay-Sachs disease does not lower an individual's risk of being a carrier. It may simply be a lucky accident that, thus far, no child inherited a pair of Tay-Sachs genes.

Very often, Tay-Sachs testing is not included in routine health care. To be safe, remember to discuss testing with your health care provider.

Who is at-risk?

Tay-Sachs carriers are found most frequently among families of Eastern European Jewish descent (Ashkenazi Jews). In the United States today, approximately one in every 27 Jews is a Tay-Sachs carrier.

Among Jews of Sephardic origin and in the general, non-Jewish population, the carrier rate is about one in 250. There are certain exceptions. French-Canadians and the Cajun community of Louisiana have the same carrier rate as Ashkenazi Jews, one in 27. Also, individuals with ancestry from Ireland are at increased risk for the Tay-Sachs gene. Current research indicates that among Irish Americans, the carrier rate is about one in 50.

All couples planning to have children should carefully consider their ancestry to evaluate the risk of each partner. Any person who can trace his or her lineage to a high-risk population must be tested. In addition, close relatives of carriers (children, sisters, brothers, cousins, aunts, uncles) must be tested since they may also be carriers.

WHAT IS CANAVAN DISEASE (CD)?

Newborns with Canavan disease appear healthy until between three and nine months of age when subtle changes are noticeable, such as visual inattentiveness or an inability to grasp objects, roll over and perform other motor tasks. The child eventually becomes blind, but hearing remains sharp, so that affected children continue to respond to the voices of their caregivers. As the child grows, the presentation and progression of Canavan symptoms vary from one child to the next. In general, difficulties that surface include weak muscles that keep children from sitting or crawling, seizures and eating problems. Children with Canavan disease usually die in infancy or early childhood, although some survive into adolescence.

Canavan disease (CD) belongs to a group of conditions known as leukodystrophies, characterized by defects in myelin, commonly known as the "white matter" in the brain. Myelin protects nerves and allows messages to be sent to and from the brain. All CD symptoms are explained by the progressive loss of myelin.

As with Tay-Sachs, children with CD have an enzyme deficiency. The enzyme, aspartoacylase, is responsible for the break-down of a particular chemical in the brain. Without this enzyme, the chemical accumulates and causes the destruction of myelin.

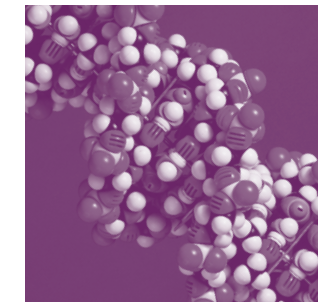
Why is a baby born with Canavan?

Canavan, like Tay-Sachs, is an inherited disease, transmitted from parent to child through genes. In the case of Canavan, the critical pair of genes controls production of the enzyme mentioned above, aspartoacylase enzyme. When both parents are Canavan carriers, each one may transmit the mutated gene to their offspring, in

which case the child is born with Canavan disease. The inheritance pattern is identical to that of Tay-Sachs, so you may want to re-read the section "Why is a baby born with Tay-Sachs?"

Who is at-risk?

Canavan disease is most prevalent among folks of Eastern European Jewish descent. The carrier rate in this population is one in 35. While Canavan does occur in other ethnic groups, it is rare and carrier frequency in the non-Jewish population has not yet been measured.



How do you know if you're a Canavan carrier?

As with Tay-Sachs, the answer is a simple blood test.

While the common Tay-Sachs screening is a serum test that measures enzyme level in the blood, DNA testing is required in

order to identify Canavan carriers.

Remember, Tay-Sachs and Canavan are caused by different alterations in different genes. The fact that you are, or are not, a Tay-Sachs carrier has no bearing on whether you are a Canavan carrier. You must be tested for both diseases to know your Tay-Sachs and Canavan carrier status.

THERE ARE MANY FAMILY PLANNING OPTIONS FOR CARRIER COUPLES

Couples who are both carriers of the same disease will want to explore their many options for a healthy family. It is best to consult a genetic counselor who can explain the various choices open to you when planning a family.

Prenatal diagnosis early in pregnancy will reveal if the fetus has Tay-Sachs or Canavan disease. At-risk couples can choose from two procedures: amniocentesis, done around the 16th week of pregnancy, and chorionic villus sampling (CVS), performed between the 10th and 13th weeks. In either case,

if the fetus is affected with Tay-Sachs or Canavan, couples may elect to have a therapeutic abortion.

In addition to adoption, there are several medical procedures available to couples for whom abortion is not an option. Your genetic counselor can explain these alternatives so that you are prepared to meet with a medical specialist, should you choose this route.

*In this pamphlet, Tay-Sachs disease refers to Classic or Infantile Tay-Sachs

FIND OUT YOUR RISK,
AND THE RISK TO YOUR LOVED ONES,
OF HAVING A BABY WITH TAY-SACHS
OR CANAVAN DISEASE.
KNOW HOW TO PREVENT THESE
DEVASTATING ILLNESSES.

YOU CAN MAKE INFORMED DECISIONS
FOR A HEALTHY FAMILY NOW,
AND FOR GENERATIONS TO COME.

National Tay-Sachs & Allied Diseases Association of Delaware Valley (NTSAD-DV) is a non-profit, volunteer health agency dedicated to the prevention and elimination of Tay-Sachs, Canavan and the allied diseases. We strive to achieve our goal through public awareness and education, carrier screening, resource and referral services, advocacy and research.

For more information about NTSAD-DV or the allied diseases, to become involved as a volunteer, or to make a donation in support of the programs that fight these deadly diseases, please contact us:



NTSAD - D V

720 Greenwood Ave., Suite 203
Jenkintown, PA 19046
215-887-0877 215-887-1931 FAX
<http://www.tay-sachs.org>
email: NTSAD@aol.com

*Funded through the Alison Ossip Fund,
these brochures were created in loving memory of Alison,
born with Canavan disease, and in the hope that no child
will again suffer from this deadly disease.*

WHEN IT COMES TO HAVING A HEALTHY BABY, SOME THINGS ARE IN YOUR HANDS.

WOULD YOU ENDANGER YOUR BABY'S LIFE
WHEN A SIMPLE TEST CAN ELIMINATE THE RISK?

TAY-SACHS AND CANAVAN CAN STRIKE

ANY FAMILY WITHOUT WARNING

AND ARE ALWAYS FATAL IN CHILDHOOD.

TAY-SACHS DISEASE
CANAVAN DISEASE

CAN BE PREVENTED

if you

KNOW THE FACTS



WHILE THERE IS NO TREATMENT FOR
EITHER DISEASE, MEDICAL SCIENCE HAS
DEVELOPED A UNIQUE STRATEGY OF
PREVENTION. COUPLES CAN PREVENT THE
BIRTH OF AN AFFECTED CHILD WITH A
SIMPLE BLOOD TEST BEFORE PREGNANCY.

TAKE THE FIRST STEP TO
PREVENTION BY BEING INFORMED.
READ THE INFORMATION IN THIS BOOKLET
TO LEARN WHAT THESE DISEASES ARE.

WHAT IS
CANAVAN
DISEASE

WHAT IS
TAY-SACHS
DISEASE