

Tay-Sachs disease

Tay-Sachs disease (TSD) is an inherited condition that tends to affect people of central and northern European Jewish (Ashkenazi) or French-Canadian ancestry. The faulty gene targets the nervous system. Symptoms first appear at around six months of age in a previously healthy baby. Over a short period of time, the baby stops moving and smiling, becomes paralysed and eventually dies. Most children with TSD die before their fifth birthday. There is no cure.

An essential enzyme is missing

The cells of the nervous system (neurons), including the brain and spinal cord, need an enzyme called B-Hexosaminidase A (HexA) to regularly break down a particular fatty substance called GN12 ganglioside that is normally present. In children with Tay-Sachs disease, the gene that tells the neurons to produce the HexA enzyme is faulty and stops the neurons from producing the enzyme. This means that the fatty substance builds up in the brain and spinal cord, causing cell damage and death. Both boys and girls are equally affected.

The symptoms in a young baby include:

- No longer smiling, reaching out or holding onto objects
- Becoming immobile and losing the ability to crawl or turn over
- Blindness.

The faulty gene

The genes in our cells are in pairs because we inherit a set from each of our parents. A person can be a carrier of the faulty gene (a genetic carrier for TSD) if they have inherited a working HexA gene copy from one parent and a faulty HexA gene copy from the other parent. Carriers of the faulty gene that causes TSD will never develop the condition. To develop TSD, the child must inherit the faulty (mutated) gene copy from **both** parents. So both these parents must be genetic carriers for TSD to have a child with the condition.

While the condition is particularly common in Jewish people from central and eastern Europe (Ashkenazi Jews) as well as French-Canadians, people of other nationalities can also carry the faulty gene. Worldwide, about one in every 30 Ashkenazi Jews and one in 40 French-Canadians are genetic carriers of TSD.

In Australia, however, about one in every 25 Ashkenazi Jews is a genetic carrier for TSD. On the other hand, about one in 280 Jewish people from the Middle East (Sephardic Jews) and non-Jewish people will be genetic carriers of TSD.

If a man and a woman each carrying a faulty HexA gene conceive a child, there is a one in four chance that the child will have TSD. If only one parent has the faulty gene, the child won't develop the condition but has a 50 per cent chance of being an unaffected genetic carrier, just like their parents.

Tests are available

There is a test to check whether a person is a TSD genetic carrier. There are two ways to do the test. One way is to check the level of HexA enzyme that is in the person's blood. Since a TSD genetic carrier has only one functioning gene copy out of the two copies, they will have half the correct amount of HexA in their bloodstream – this is enough for normal brain function. The diagnosis can then be confirmed by examining the gene that contains the information for HexA production.

The second way of doing the test is more accurate and becoming much more common. Here, the person's DNA is directly examined for the faulty HexA gene. The DNA is usually obtained by taking a sample of the cells lining the inside of their cheek. The person is asked to swill a mouthful of water and spit it into a container. This is called the mouthwash test. The DNA can also be obtained by wiping the inside of the cheek with a cotton swab.

Counselling and support

If the testing shows that both partners in a couple are genetic carriers for TSD, they can get genetic counselling to help them decide on their options in regard to having children. Genetic services in Victoria provide information and counselling and referral to community resources including support groups if needed.

TSD genetic carriers aren't harmed by the condition

Being a TSD genetic carrier doesn't pose any risk to the carrier. One functioning gene copy is all that's required to produce enough of the HexA enzyme to regularly break down the fatty substance in the brain and spinal cord.

Where to get help

- Your doctor
- Genetic counsellor – available at most large public maternity hospitals
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Genetic Support Network Victoria Tel. (03) 8341 6315

Things to remember

- Tay-Sachs disease is a genetic condition common in Ashkenazi Jews and French-Canadians.
- The faulty (mutated) gene stops the cells of the nervous system from producing an enzyme vital to the health of the brain and spinal cord.
- If a child inherits both copies of faulty (mutated) genes for TSD from their genetic carrier parents, he or she becomes paralysed and dies by the time they are three or four years old.

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