Tay-Sachs disease

Summary

- Tay-Sachs disease (TSD) is a genetic condition that affects the nervous system.
- It becomes progressively worse over time.
- Symptoms usually first appear at around six months of age in previously healthy babies.
- The life expectancy for children with TSD is around five years of age.
- There is currently no effective treatment.
- TSD is more commonly seen in people who are of Ashkenazi Jewish or French-Canadian descent.

Tay-Sachs disease (TSD) is a genetic condition that affects the nervous system. It is caused by an alteration in the \textit{HEXA} gene on chromosome 15. TSD is more commonly seen in people who are of Ashkenazi Jewish or French-Canadian descent. Males and females are equally affected.

Tay-Sachs disease is a degenerative condition, meaning that symptoms become worse over time. In people with TSD the nerve cells in the brain and spinal cord are progressively destroyed, leading to paralysis. Symptoms first appear at around six months of age in a previously healthy baby. The life expectancy for children with TSD is around five years of age and there is currently no effective treatment.

What causes Tay-Sachs disease?
The cells of the nervous system (neurones), including the brain and spinal cord, need an enzyme called B-Hexosaminidase A (HexA) to regularly break down a fatty substance called GM2 ganglioside. In children with TSD, the gene that tells the neurones to produce HexA is altered and stops the neurones from producing the enzyme. This means that the fatty substance builds up in the brain and spinal cord, causing cell damage and death.

The genetics of Tay-Sachs disease
The genes in our cells are in pairs. We inherit a set from each of our parents. TSD follows an autosomal recessive inheritance pattern. This means that:

- the affected gene is on one of the non-sex chromosomes
- both copies of the gene need to be altered for a person to have the condition.

If a person inherits a functional \textit{HEXA} gene copy from one parent and an altered \textit{HEXA} gene copy from the other parent, they are said to be a ‘genetic carrier’ of the altered \textit{HEXA} gene. Carriers of the altered \textit{HEXA} gene do not develop TSD.

To develop TSD, a child must inherit an altered gene copy from both parents. This means that both parents must be genetic carriers for TSD to have a child with the condition.

If a male and a female, each carrying an altered \textit{HEXA} gene, conceive a child, there is a:

- one-in-four chance that the child will have TSD
- two-in-four chance that the child will be a genetic carrier of TSD
- one-in-four chance that the child will be completely unaffected by TSD.

If only one parent has the altered gene, none of their children will develop the condition, but each child has a 50 per cent chance of being a genetic carrier of TSD.

Tay-Sachs disease is most commonly observed in individuals of Jewish and French-Canadian descent, but can also occur in people from other backgrounds.
Symptoms of Tay-Sachs disease

The symptoms of TSD in a young baby include:

- movement problems – loss of ability to smile, reach out, hold onto objects, crawl, turn over or sit up
- vision and hearing impairment
- exaggerated reactions to loud noises
- seizures.

Testing for Tay-Sachs disease

If TSD runs in your family, start by discussing testing options with your GP.

There are two ways to test whether a person is a genetic carrier of TSD:

- check the level of HexA enzyme in the person’s blood. Since a TSD genetic carrier has only one functioning copy of the gene, they will have half the usual amount of HexA in their bloodstream. This is enough for normal brain function. If levels of HexA are found to be at 50 per cent, a diagnosis of TSD can be confirmed by examining the HEXA gene
- examine the person’s DNA for the altered 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.

Examining the person’s DNA is a more accurate test.

Counselling and support for Tay-Sachs disease

If your child, or a family member, has been diagnosed with TSD, or it runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand TSD and what causes it, how it is inherited, and what a diagnosis means for your child’s health and development. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If TSD runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

If testing shows that you and your partner are both genetic carriers for TSD, a genetic counsellor can advise you about your reproductive options.

The Genetic Support Network of Victoria (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by TSD.

Genetic carriers of Tay-Sachs disease

If you are a TSD genetic carrier it doesn’t pose any health risk to you. One functioning gene copy is all that’s required for you to produce enough of the HexA enzyme to regularly break down the fatty substance in your brain and spinal cord.

Where to get help

- Your GP (doctor)
- Clinical geneticist
- Victorian Clinical Genetics Services (VCGS) , Royal Children’s Hospital Tel. 1300 118 247
- Genetic Support Network of Victoria (GSNV) Tel. (03) 8341 6315