# Mortal Defect

## A study of Tay-Sachs Disease

Psych 150

November 17, 2005

What is and causes Tay-Sachs disease (TSD)?

Tay-Sachs disease is an extremely rare genetic deviant that leaves the child's developing body incapable of creating the enzyme hexosaminidase A which is important for breaking down gangliosides, also known as GN12, in nerve cells. An increasing collection of gangliosides is lethal and the result is cell death (CSHL). The enzyme hexosaminidase A is contained on chromosome number 15. For someone with Tay-Sachs, the two copies in every body cell are affected. These people cannot produce the important enzyme to break down the fatty substance of GN12 which eventually damages the cells.

Who is most likely to get Tay-Sachs Disease (TSD)?

A British ophthalmologist, Dr. Tay, first characterized the red spots in the retina of babies in 1881. Later, a New York neurologist, Dr. Sachs, observed the occurrences in Eastern European Jews in 1887 (Densick 2001). Presently, Tay-Sachs is most seen in those of Central and Eastern European ancestry, French Canadians whose heritage lies in the St. Lawrence region, certain Cajuns, and some Amish communities (TCE 2004). In the general public, Tay-Sachs is seen in 1 out of every 250 persons (NMSI). Since Tay-Sachs is a recessive trait, both parents must be carriers of the mutated gene for their child to be affected (CGE 2004).

Chances of Inheriting Tay-Sachs Disease (TSD)

Four outcomes in each pregnancy are likely for the genes to be carried on to the child if both parents are carriers of TSD. The unborn child has a 50% change of being a genetic carrier and a 25% change of being either a non-carrier or affected. If only one parent is a carrier there is no chance the child will be affected but a 50% chance of either

being a non-carrier or a genetic carrier of TSD. To determine whether or not a person has Tay-Sachs, blood tests can be done to determine the amount of hexosaminidase A is present. A TSD carrier would have half the amount of enzyme as a non-carrier (CGE 2004).

Screening Unborn Babies for Tay-Sachs Disease (TSD)

Tay-Sachs disease can be screened as of the early 1970s. This was encouraged in high-risk populations. Couples already pregnant can now test the fetus through chorionic villus sampling or amniocentesis. Chorionic villus sampling involves inserting a fine needle into the abdomen or a plastic catheter into the cervix which collects the chorionic membrane surrounding the fetus which can then be tested for TSD. Amniocentesis involves removing a sample of the amniotic fluid around the fetus with a fine needle similar to the chorionic villus sampling. This procedure is normally done around the 14$^{th}$ or 15$^{th}$ week when abortion is still an option.

Symptoms of Tay-Sachs Disease (TSD)

The infant may appear healthy in all aspects until 6 months. Around this time, the child appears to developmentally regress. Babies stop smiling, crawling, or turning over and lose the ability to grasp or reach out. Following this are blindness, paralysis and epileptic seizures (NINDS 2006). Children may also experience an inability to swallow, difficulty breathing, and mental retardation. These symptoms are stretched out until the child's eventual death around the age of 5 (CGE 2004).

Curing Tay-Sachs Disease (TSD)?

There is no cure for Tay-Sachs disease at the present time. Medicines may help temporarily relieve some of the symptoms, for example; anticonvulsant medicines may

initially control the seizures (NINDS 2006). Mouse models, although limited in their alternative pathway for breaking down gangliosides, have shown ganglioside synthesis inhibitors promising. Ganglioside synthesis inhibitors are harder to treat the infant onset of Tay-Sachs disease because the extent of damage during the fetal period has yet to be determined. The inhibitors have helped with the later forms of Tay-Sachs and combined with DNA and enzymatic screening programs will allow the prospective control of the disease.

Summary

Tay-Sachs disease is an extremely rare genetic deviant that leaves the child's developing body incapable of creating the enzyme hexosaminidase A which is important for breaking down gangliosides, also known as GN12, in nerve cells. An increasing collection of gangliosides is lethal and the result is cell death (CSHL). Presently, Tay-Sachs is most seen in those of Central and Eastern European ancestry, French Canadians whose heritage lies in the St. Lawrence region, certain Cajuns, and some Amish communities (TCE 2004). Four outcomes in each pregnancy are likely for the genes to be carried on to the child if both parents are carriers of TSD. If only one parent is a carrier there is no chance the child will be affected but a 50% chance of either being a non-carrier or a genetic carrier of TSD. Couples already pregnant can now test the fetus through chorionic villus sampling or amniocentesis. Symptoms appear around 6 months in a degenerating pattern with blindness, paralysis, and epileptic seizures (NINDS 2006). There is no cure for infants with Tay-Sachs disease, and their eventual death occurs around the age of 5.

# Bibliography

Centre for Genetics Education, CGE. (2004). Tay-sachs Disease. *The Centre for Genetics Education.* November 14, 2006, from http://www.genetics.com.au/factsheet/35.htm

Cold Spring Harbor Laboratory, CSHL. Tay-Sachs Disease. *Your Genes, Your Health.* November 14, 2006, from http://www.ygyh.org/tay/whatisit.htm

Desnick, R. & Kaback, M. (2001). *Tay-Sachs Disease: volume 1.* Academic Press.

National Institute of Neurological Disorders and Stroke, NINDS, National Institutes of Health, NIH. (2006, January). NINDS Tay-Sachs Disease Information Page. *National Institute of Neurological Disorders and Stroke.* November 15, 2006, from http://www.ninds.nih.gov/disorders/taysachs/taysachs.htm

National Library of Medicine, NLM. (2006). Tay-Sachs Disease. *Genes and Disease.* Retrieved November 14, 2006, from http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.section.238&ref=sidebar

National Museum of Science and Industry, NMSI. Tay-Sachs Disease, Understanding

Tay-Sachs Disease, How is Tay-Sachs Disease Inherited. *Science Museum| who am i?*

November 14, 2006, from http://www.sciencemuseum.org.uk/exhibitions/genes/230.asp
The Columbia Encyclopedia, TCE. (2004). Tay-Sachs Disease. *Reference.com* November 14, 2005, from http://www.reference.com/browse/columbia/Tay-Sach