

Chem 412 Seminar

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Tay-Sachs Disease: Childhood Killer

Tay-Sachs Disease is an autosomal recessive disease that affects mostly people of Eastern Jewish, Louisiana Cajun, or French Canadian descent. Children receiving both faulty copies of the Tay-Sachs Disease allele (Hex A) are born normal, but soon begin to lose their mental and physical capabilities until their death later in childhood. Tay-Sachs disease is caused by a mutation in the β -hexosaminidase enzyme which degrades GM2 ganglioside as well as other substrates in the lysosomes of all neuron cells. Mutations in the α subunit of β -hexosaminidase (α/β heterodimer) lead to problems in enzyme activity. The most common mutations however cause either a loss of several amino acids that somehow have drastic effects on activity, or loss of large stretches of amino acids (exon 1). In Tay-Sachs patients GM2 ganglioside levels in the brain gradually increase, bringing about the mental and physical incapacitation and eventual death.

Several different techniques are used to identify Tay-Sachs carriers. Most guidance is directed toward genetic counseling of high risk carriers, and prevention or early term abortion of affected fetuses.

An animal model has been created in mice. The Hex A gene has been knocked out, and studies are underway to try to develop new therapies for Tay-Sachs Patients. Unfortunately, no cure or any other real therapy has yet been created to help abate the effects of this terrible disease.

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