Tay-Sachs (HEXA)

Tay-Sachs disease is caused by mutations in the HEXA gene, which encodes for one subunit of the enzyme beta-hexosaminidase A. The enzyme breaks down the toxic substance GM2 ganglioside in the brain and spinal cord. Symptoms usually develop from three months onwards, including loss of motor skills, increasing weakness, and strong startle response. Loss of vision and hearing, seizures, and paralysis normally follow. Life expectancy is 2 to 4 years. Very rare related diseases, which begin later in childhood, adolescence, or early adulthood are also known, but the symptoms are usually much milder.

Tay-Sachs is rare in the general population, but tends to be concentrated in various ethnic groups. Among those of Ashkenazi Jewish descent, about 1 in 30 are carriers for the disease. There is also a high level of carriers in the Acadian (Cajun) population of Louisiana, and among French Canadians. However, extensive genetic counseling has led to a large reduction in the number of live births over recent decades. The faulty gene is autosomal recessive, typically requiring both parents to be asymptomatic carriers of the faulty gene copy.

Sources

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