Tay-Sachs Disease

National Cancer Institute

Source

National Cancer Institute. Tay-Sachs Disease. NCI Thesaurus. Code C85184.

A rare, fatal, autosomal recessive lipid storage disorder caused by mutations in the HEXA gene. It is characterized by deficiency of beta-hexosaminidase A, resulting in accumulation of gangliosides in the neurons of the brain and spinal cord. Signs and symptoms include progressive deterioration of the mental and physical abilities early in life, accompanied by blindness, deafness, muscle atrophy, and paralysis.