



Disability Awareness Begins With You: Tay-Sachs Disease

Tay-Sachs disease is a rare, neurodegenerative disorder in which deficiency of an enzyme (hexosaminidase A) results in excessive accumulation of certain fats (lipids) known as gangliosides in the brain and nerve cells. This abnormal accumulation of gangliosides leads to progressive dysfunction of the central nervous system. This disorder is categorized as a lysosomal storage disease. Lysosomes are the major digestive units in cells. Enzymes within lysosomes break down or "digest" nutrients, including certain complex carbohydrates and fats.

Symptoms associated with Tay-Sachs disease may include an exaggerated startle response to sudden noises, listlessness, loss of previously acquired skills (i.e., psychomotor regression), and severely diminished muscle tone (hypotonia). With disease progression, affected infants and children may develop cherry-red spots within the middle layer of the eyes, gradual loss of vision, and deafness, increasing muscle stiffness and restricted movements (spasticity), eventual paralysis, uncontrolled electrical disturbances in the brain (seizures), and deterioration of cognitive processes (dementia). The classical form of Tay-Sachs disease occurs during infancy; an adult form (late-onset Tay-Sachs disease) may occur anytime from adolescence to the mid 30s.

Tay-Sachs disease is inherited as an autosomal recessive trait. The disorder results from changes (mutations) of a gene known as the HEXA gene, which regulates production of the hexosaminidase A enzyme. The HEXA gene has been mapped to the long arm (q) of chromosome 15 (15q23-q24).

Organizations related to Tay-Sachs Disease

Let Them Hear Foundation
1900 University Ave #101
East Palo Alto CA 94303
650-462-3143
877-735-2929
info@letthemhear.org
<http://www.letthemhear.org>

March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue
White Plains NY 10605
914-428-7100
888-663-4637
Askus@marchofdimes.com
<http://www.marchofdimes.com>

NIH/National Institute of Child Health and Human Development
31 Center Dr
Bethesda MD 20892
301-496-5133
<http://www.nih.gov/hichd/>

National Institute of Neurological Disorders and Stroke (NINDS)
31 Center Drive
Bethesda MD 20892-2540
301-496-5751
800-352-9424
braininfo@ninds.nih.gov
<http://www.ninds.nih.gov/>

National Tay-Sachs and Allied Diseases Association
2001 Beacon Street
Brighton MA 02135
617-277-4463
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info@ntsad.org
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