Condition: Tay-Sachs disease

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in *HEXA* gene, which encodes beta-hexosaminidase, alpha chain.

Frequency:
1/3,600 in Ashkenazi Jews; increased frequency also in French Canadians and Old Order Amish.

Clinical features:
The acute infantile form presents with neurological deterioration beginning around 3 months of age. There are myoclonic jerks and an enhanced startle reaction is noted. There is gradual enlargement of head size and development of a cherry-red spot in the macula of the eye. Most die between 2-4 years of age. Milder forms may present in later childhood, though progression to death still occurs. An adult-onset form presents primarily with motor symptoms.

Management:
Supportive care.

Genetic counseling:
Based on autosomal recessive inheritance; diagnosis of carrier status may be done by enzyme assay or molecular genetic testing. Carrier screening programs in high risk populations have reduced the incidence of the disease.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.