Condition: Canavan disease

Inheritance:
Autosomal recessive

Genetic etiology:
Mutation in *ASPA* gene that codes for enzyme aspartylacylase.

Frequency:
Approximately 1/10,000 in Ashkenazi Jewish population; rare in other populations

Clinical features:
Affected children are normal at birth and for the first few months of life. They then experience developmental regression and develop macrocephaly. MRI reveals abnormalities in white matter. Children are hypotonic, and later develop spasticity. Most do not survive past the teen years.

Management:
Supportive care.

Genetic counseling:
Parents of affected child have 25% risk of recurrence. Carrier testing can be accomplished by DNA testing, and is included in some routine carrier testing programs for individuals of Ashkenazi Jewish ancestry. Three mutations comprise the majority seen in this population.

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