Condition: Familial dysautonomia

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in *IKBKAP* gene, which encodes IkappaB kinase complex-associated protein. Two mutations occur in the Ashkenazi Jewish population, where the condition is most often seen. One is a mutation in the splice donor in intron 20 that results in skipping of exon 20, and the other is a missense mutation in exon 19.

Frequency:
1/3,700 in Ashkenazi Jews.

Clinical features:
Familial dysautonomia is characterized by dysfunction of the autonomic nervous system and sensory neurons beginning at birth. There are episodes of vomiting, aspiration pneumonia, temperature and cardiac instability, and altered sensitivity to pain. Premature death may occur due to infection or cardiac events.

Management:
Supportive care.

Genetic counseling:
Parents of affected child face 20% risk of recurrence; genetic testing is available.

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