Familial Dysautonomia (IKBKAP)

Familial dysautonomia is caused by defects in the IKBKAP gene. This gene encodes for a protein called IKK complex-associated protein, which plays a role in protein transcription. Nerve cells are adversely affected when the protein fails to function. Children with the disease typically suffer from gastrointestinal problems (such as vomiting), feeding difficulties, somewhat stunted growth, muscle weakness, and a lack of sensitivity to pain or temperature. Sufferers are liable to suffer from lung infections far more than normal. Curvature of the spine and deterioration in vision often occur. Walking becomes increasingly difficult as adulthood is reached, and many patients reach the point where they are no longer able to walk unaided. Kidney damage is also common during adulthood. Early death is likely, often due to lung infections, although improvements to treatment mean that around half of all patients now survive to age 40.

Familial Dysautonomia is normally found in those of Ashkenazi Jewish descent, where about 1 in 3,700 are affected; approximately 1 in 36 are carriers. The mutated gene is inherited in an autosomal recessive manner, which typically requires both parents to be asymptomatic carriers of the faulty gene copy. However, there have been cases of both male and female sufferers having children, although pregnancy is high risk for those with the condition. The offspring between affected patients and non-carriers will normally be asymptomatic carriers.

Sources

NIH, Genetics Home Reference: Familial Dysautonomia.
See http://ghr.nlm.nih.gov/condition/familial-dysautonomia

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See http://ghr.nlm.nih.gov/gene/IKBKAP

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