



POSNA

The Core Curriculum

Friedreich ataxia

Objectives

1. Describe early symptomatology and signs of Friedreich ataxia.
2. Discuss genetic transmission of Friedreich ataxia
3. Describe the orthopaedic problems associated with Friedreich ataxia

Discussion

Friedreich ataxia (FRDA) is an autosomal recessive disorder with a frequency of 1/50,000 live births, predominately in North America in patients of French Canadian heritage. It is caused by a defect in chromosome 9, leading to a reduction in a protein, frataxin, which is normally widely distributed in mitochondria. There is now a simple genetic test for FRDA, and rapid progress in the understanding of the pathophysiology of this disorder can be expected in the next decade.

In children, the mean age at onset of symptoms is about 6 years, presenting with an abnormal gait. Ataxia and depressed reflexes are important physical findings related to initial diagnosis, Harding listed a number of additional criteria which have become standard, but ataxia and depressed reflexes are paramount. Other signs may take 5 years to appear. The initial loss of strength is in the hip extensors, followed by additional lower limb weakness. Upper limb and trunk muscles weakened considerably later. Use of a wheelchair occurs at about age 18. The three major orthopaedic problems associated with FRDA are scoliosis, pes cavovarus, and painful muscle spasms. When the disease onset is before age 10, progression of scoliosis to 60 degrees can be expected. Surgical procedures for cavovarus feet are standard for treatment of that problem, including tenotomy, tendon transfer, osteotomy, or arthrodesis.

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