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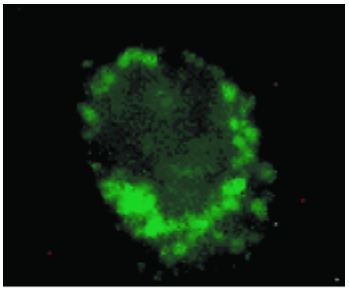
Friedreich's ataxia



Mitochondrial localization of human frataxin in live mammalian cells. [Reproduced from Babcock, M. et al. (1997) Regulation of Science 276: 1709-1712, with permission.]

Friedreich's ataxia (FRDA) is a rare inherited disease characterized by the progressive loss of voluntary muscular coordination (ataxia) and heart enlargement. It is named after the German doctor, Nikolaus Friedreich, who first described the disease in 1863. FRDA is generally diagnosed in childhood and affects both males and females.

FRDA is an autosomal recessive disease caused by a mutation of a gene called frataxin, which is located on chromosome 9. This mutation means that there are many extra copies of a DNA segment, the trinucleotide GAA. A normal individual has 8 to 30 copies of this trinucleotide, while FRDA patients have as many as 1000. The larger the number of GAA copies, the earlier the onset of the disease and the quicker the decline of the patient.

Although we know that frataxin is found in the mitochondria of humans, we do not yet know its function. However, there is a very similar protein in yeast, YFH1, which we know more about. YFH1 is involved in controlling iron levels and respiratory function. Since frataxin and YFH1 are so similar, studying YFH1 may help us understand the role of frataxin in FRDA.

Related diseases

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