Friedreich's Ataxia

Friedreich’s ataxia is a trinucleotide repeat disorder marked by characteristic ataxia in young individuals. The disorder displays an autosomal recessive inheritance pattern and involves the Frataxin gene. As trinucleotide repeats of GAA accumulate, protein synthesis decreases and subsequent CNS and multisystem damage results. The CNS damage is characteristically localized to the corticospinal tract, posterior column, and spinocerebellar tract, all which contribute to the patient’s difficulties with balance and mobility. Patients can also have a unique combination of additional findings including kyphoscoliosis, hammertoes, high arching feet, and type 1 diabetes. The most serious and potentially fatal finding in these patients is hypertrophic cardiomyopathy, which can lead to sudden cardiac death at a young age.



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Trinucleotide Repeat
DNA-strand with Repeats
Friedreich's ataxia is caused by a trinucleotide repeat expansion. Most individuals normally have approximately 25 repeats but affected individuals have up to 100 repeats and some up to 1,000 repeats have been identified. Expansion occurs in the frataxin gene region causing a decrease in the amount of protein synthesized.

GAA Repeat
GArAge with Glue-Apple-Apple lights Repeating
The nucleotides GAA repeat.

Frataxin
Frataxin-sign
Repeats occur in the frataxin gene.

Mitochondrial Dysfunction
Damaged Mitochondria
Reduced levels of the frataxin protein results in mitochondrial dysfunction.

Autosomal Recessive
Recessive-chocolate
This disease is inherited in an autosomal recessive fashion.

Hammertoes
Hammer-toes
Also called contracted toe, hammer toes refer to a deformity in the proximal interphalangeal joint of second, third, and fourth toes that cause it to be bent permanently. This deformity causes the toe to look like a hammer.

High Arches
High arches
Also called pes cavus deformity, high arches describes a foot type with extremely high arches between the ball and heel of the foot.

Kyphoscoliosis
Curvy-skull-spine
A musculoskeletal disorder caused by abnormal curvature of the spine.

Lateral Corticospinal Tract
Ladders on Corticospinal Tract
The lateral corticospinal tract consists of an upper motor neuron that descends from the cerebral cortex, decussates in the medulla, and proceeds down to the contralateral side of the spinal cord. The lateral corticospinal tract leaves out the anterior horns of the spinal cord and controls fine movement of the ipsilateral limbs, which are contralateral to the respective motor cortex.

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Posterior Column

Post-terrier column

The white matter tract in the dorsomedial side of the spinal cord. This tract contains ascending fibers important for fine touch, vibration, pressure, and proprioception.

Spinocerebellar Tract

Spinal column Silver-cerebellum-bell

Set of fibers that begin in the spinal cord and terminate in the ipsilateral cerebellum that conveys information about limb and joint position. This tract is especially important for fine movement and damage to this tract can cause ataxia.

Type I Diabetes Mellitus

Dyed-beads-pancreas with (1) Wand

About 20% of patients with this disease have difficulties controlling blood sugar and about 10% develop Type 1 Diabetes Mellitus.

Hypertrophic Cardiomyopathy

Hiker-trophy with Hypertrophic-heart

The majority of patients demonstrate cardiac involvement, especially hypertrophic cardiomyopathy. This is the most common cause for death of patients with Friedreich's ataxia.

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