Friedreich Ataxia

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Friedreich Ataxia Information Page.

Friedreich ataxia is a rare inherited disease that causes progressive damage to the nervous system. It is caused by a defect in the FXN gene that produces the protein frataxin. Frataxin controls important steps in mitochondrial iron metabolism and overall cell iron stability. Research suggests that cells that have a reduced level of frataxin produce energy less effectively, which may lead to a buildup of toxic byproducts.

Symptoms typically appear between ages 5 and 15 years but can begin in adulthood. Damage to the peripheral nerves and the cerebellum (part of the brain that coordinates balance and movement) results in awkward, unsteady movements and impaired muscle coordination (ataxia) that worsens and eventually spreads to the arms and the trunk of the body. Other symptoms include loss of sensory function, speech problems, and vision and hearing loss. Thinking and reasoning abilities are not affected. Many people with Friedreich ataxia develop scoliosis (a curving of the spine to one side), which, if severe, may impair breathing. Some individuals may develop diabetes.