

What is it?

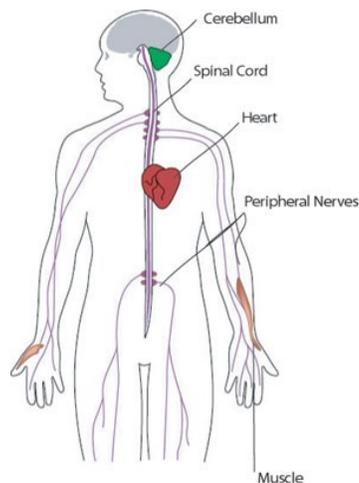
Friedreich's ataxia (FA) is a progressive genetic condition that impairs a part of the brain called the cerebellum which affects movement and coordination. An individual can suffer multiple neuromuscular losses. Major neurological symptoms characteristic of FA include muscle weakness, loss of coordination, and impaired balance.

The FA gene mutation that causes the disease actually affects the cell's production of the protein called frataxin. Frataxin is the key protein component of the cell's mitochondria. Since the mitochondria is considered the powerhouse of the cell, alterations of frataxin structure therefore greatly affect energy production. Without efficient frataxin levels, certain cells in the body cannot produce energy. In FA, these particular cells include those of peripheral nerves, spinal cord, brain, and heart muscles. As a result, a person has a difficult time with balance, movement, spatial perception, speech, and can even suffer cardiac conditions.

As the most common form of autosomal recessive ataxia, 1 in 50,000 individuals are affected. The progressive degeneration of physical abilities including weakness and fatigue can lead to motor incapacitation, the use of mobility aids, and often the use of a wheelchair. However, while motor function progressively declines the individual's cognitive function, reasoning and thinking ability remain intact.



What are the signs and symptoms?



Signs and symptoms of onset have been identified in ages 5-15 years, often before the end of puberty. Early symptoms of FA could include awkward, clumsy movement and difficulty walking. As the disease progresses, the arms and trunk will also be affected. Other signs and symptoms include loss of coordination of arm and legs, fatigue, vision and hearing impairment, scoliosis, diabetes mellitus, and several heart conditions. Not only is there a loss of function, but reflexes are also affected.

- Loss of coordination
- Unsteady walking
- Tripping
- Fatigue
- Poor gross motor skills
- Decreased fine motor skills
- Stiffness of extremities
- Urinary urgency
- Slow or slurred speech
- Chest pain
- Diabetes
- Cardiac conditions
- Tachycardia
- Decreased reflexes
- Scoliosis
- Vision loss
- Hearing impairment
- Shortness of breath

Suggested school accommodations

Students can experience difficulties with balance, standing and walking. They can also experience problems with tasks that involve manipulation like handwriting. A student can grow fatigued as they spend time and energy to complete such gross and fine motor tasks. The rate of this exhaustion will be different from person to person and school staff need to communicate with student and family to assess each student's individual needs.

- Offer rest breaks
- Know signs of fatigue
- Classroom and campus accessibility
- Elevator access
- Wheelchair accessibility
- Note taker
- Use of assistive technology
- Additional time on assignments
- Extra set of books for home
- Help carrying items like lunch trays
- Extra transition time between classes
- Alternative methods to demonstrate understanding
- Adapted PE
- Evaluate self esteem
- Offer counseling

SHNIC school nurses information:

Specific health issues for individual health care plans

- Diagnosis, note age of onset
- Child specific characteristics and symptoms
- Current medications for home and school, PRN medications
- Baseline assessment including cardiac
- Note any physical limitations
- Adaptive equipment or orthotics, including hours of wear time for use
- Safety restrictions for use of stairs, alone in hallway, etc.
- Location and times for rest breaks
- Assigned aid or required supervision for safety
- Educate school personnel about disease
- Emergency action plan including evacuation plan

Resources & Manuals

Friedreich's Ataxia Research Alliance (FARA)
<http://www.curefa.org/whatis>

National Ataxia Foundation
<http://ataxia.org/>

National Ataxia Foundation factsheet
https://www.ataxia.org/pdf/Friedreichs_Ataxia_FAQ_11.pdf

Muscular Dystrophy Association– FA
<https://www.mda.org/disease/friedreichs-ataxia>

Friedreich's Ataxia Parents' Group
<http://www.faparents.org/fapg/>