

FRIEDREICH'S ATAXIA

What is it?

Friedreich's ataxia (FA) is a genetic condition impairing the cerebellum causing progressive nervous system damage that affects movement and coordination. Major neurological symptoms characteristic of FA include muscle weakness, loss of coordination, and impaired speech, hearing, and vision. An individual can suffer multiple neuromuscular losses. Individuals with FA often have a form of heart disease called hypertrophic cardiomyopathy.

The cause of FA is a defect in the gene responsible for a protein called frataxin. Frataxin is the key protein component of the cell's mitochondria. Without efficient frataxin levels, certain cells in the body cannot produce energy; causing degeneration of the brain, spinal cord, peripheral nerves, and heart cells.

The progressive degeneration of physical abilities including weakness and fatigue can lead to motor incapacitation which may require the use of mobility aids and often the use of a wheelchair. While motor function progressively declines, the individual's cognitive function, reasoning and thinking ability remain intact.

What are the signs and symptoms?

Symptoms typically begin between the ages of 5 and 15 years, often before the end of puberty. Early neurological symptoms of FA include awkward, clumsy movement and difficulty walking. As the disease progresses, the arms and trunk will also be affected. 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

What is the treatment?

There is no cure or effective treatment for this degenerative nervous system disease. Symptoms or complications can be treated including some of the cardiac conditions or diabetes. Physical, occupational, and speech therapy are important to the multi-disciplinary team to help prolong use and function of things like walking, talking, and swallowing.



Kennedy Krieger Institute

The Specialized Health Needs Interagency Collaboration (SHNIC) program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

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. Supporting students with this condition in the school require educators and parents/guardian to work as a team. Some accommodations to consider for a 504/IEP could include:

- PT/OT/SLP evaluation
- Audiology/impaired vision consultations
- Offer rest breaks
- Note student signs of fatigue
- Classroom and campus accessibility
- Elevator access
- Wheelchair accessibility
- Note taker
- Use of assistive technology
- Safety restrictions or supervision for use of stairs, alone in hallway, etc.
- 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
- Emotional support
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis, note age of onset
- Child specific characteristics and symptoms of FA
- Current medications including PRN medications
- Baseline assessment including musculoskeletal and cardiac
- Note physical limitations including swallowing or special nutrition orders
- Adaptive equipment or orthotics, including hours of wear time for use
- Communicate with school staff, parents/guardian, and provider any changes or concerns about the disease
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Friedreich's Ataxia Research
<https://www.curefa.org/>

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

Muscular Dystrophy Association
<https://www.mda.org/>