

FRIEDREICH ATAXIA

Background

Friedreich ataxia (FA) is a genetic disease affecting the central nervous system that causes progressive damage to the cerebellum, spinal cord, and sensory nerves. The disease causes damage characterized by ataxia, spasticity, and nystagmus. An individual can suffer poor balance and coordination due to a decrease in the ability to identify where their limbs are in space (proprioception).

The cause of FA is a defect in the gene responsible for a mitochondrial protein called frataxin. Frataxin regulates the levels of iron inside the cell's mitochondria, the energy source within the body's cells. When frataxin is missing or defective, too much iron is left to freely float or builds up in the mitochondria causing oxidative stress and reduced mitochondrial function. Certain nerve and muscle cells cannot function properly with a shortage of frataxin.

Symptoms typically begin between the ages of 10 and 15 years, often before the end of puberty. Progressive muscle weakness, impaired muscle coordination and fatigue are common. Initially the child will have difficulty walking with awkward, clumsy movement. As the disease progresses, symptoms will affect the arms and trunk and further lead to other chronic conditions such as heart disease, diabetes and scoliosis. Other symptoms may include:

- Decreased gross and fine motor skills
- Impaired speech, hearing, vision
- Shortness of breath
- Extremity stiffness and cramping
- Urinary urgency

There is no cure for FA, but new treatment options are being studied. One option to improve neurological function in adolescents and adults was approved by the FDA in 2023. Management of the disease will often include a combination of physical, occupational and speech therapy to improve or maintain strength and coordination. Medications are often prescribed to treat heart disease and diabetes and surgery may be necessary to correct skeletal problems.



Top Takeaways for School Considerations

FA is a rare and debilitating neurological disease affecting the ability of the nervous system to plan and coordinate movements.

Ataxia is the first symptom of FA. A student may appear unsteady and clumsy. The student may require the use of assistive devices.

Ataxia can make the heart work harder, leading to fatigue. The student may benefit from scheduling adjustments (e.g., flexibility, blended learning, leaving class early to avoid crowded hallways).

The progressive degeneration of physical abilities can lead to difficulty with speech and fine motor skills like handwriting. Alternative means of written communication should be considered (e.g., keyboarding, note taker and reduced paper/pencil tasks).

Though motor function progressively declines, the individual's cognitive function generally remains intact.

Kennedy Krieger Institute's Specialized Health Needs Interagency Collaboration

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



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Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnosis of impaired physical mobility, impaired swallowing, impaired gas exchange, ineffective breathing pattern, ineffective airway clearance, fatigue, and risk for ineffective peripheral tissue perfusion
- Current diagnosed health condition, including date of diagnosis, progress of disease process and other chronic health conditions
- Current medication and treatment orders (consider schedule, equipment needs and side effects)
- Nutrition interventions and equipment needs (consider brand/size of feeding tube, tube replacement, water flushes, fluid intake goal and supplements); note school district policy on tube replacement and consider keeping backup feeding tube kit at school if applicable
- Use of specialized equipment, adaptive equipment, and orthotics
- Equipment troubleshooting (consider equipment/device user manual, battery, charger)
- Consider emergency care plan(s) (ECP) and emergency evacuation plan(s) (EEP) as related to medical needs in the school setting, and staff education/training, as appropriate

Discussion Starters for Educational Team

1. Has the school staff been trained to implement the student-specific emergency plan?
2. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
3. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
4. Can rest breaks, safe spaces, or reduced stimulation times be built into the student's schedule?
5. Does the student need support with gross or fine motor skills in the classroom?
6. Is the physical school environment safely accessible for the student's mobility needs (e.g., entry and exit, ramps, location of classes, access to elevator, doorways)?
7. Does the classroom environment support the student's needs and/or equipment (e.g., desk/seating options, maneuverability space, electrical outlets, flash pass for bathroom or nurse)?

Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinic
kennedykrieger.org

Friedreich's Ataxia Research Alliance
curefa.org/

National Ataxia Foundation
ataxia.org/



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