Friedreich's Ataxia

What is ataxia?

Ataxia (say: ah-TACKS-ee-ah) is a movement disorder. Movement disorders are conditions that cause involuntary body movements. Uncontrolled movement starts in the brain, in the area you use to move, speak, and pay attention. When a child has ataxia, he has trouble controlling his muscles and can be clumsy and awkward. Common examples of ataxia include epilepsy and multiple sclerosis (MS).

What is Friedreich's ataxia?

Friedreich's ataxia (say: FREED-ricks) hardens and damages nerves in the brain and spinal cord. This disrupts brain signals. If a child has Friedreich's ataxia (FA), he will have difficulty controlling his legs and arms. The child might slur when he talks, bend over with poor posture, or curl his feet into high arches.

FA spreads from the legs to the arms, and eventually to the torso. This happens at different speeds in different children. It can eventually impair day-to-day life.

FA usually starts when a child is between 5 and 15 years old. It almost never starts later than age 20.

Friedreich's ataxia is a genetic disease

FA is a genetic disease, meaning it is passed from parents to their children. One in every 90 people carries the gene that causes FA. A gene is a section of DNA that gives an instruction to a cell. Most of the time, the instruction is a "recipe" for making a protein.

FA is caused by a recessive gene. This means that a person needs to have 2 copies of the gene to develop FA. If a person has only 1 copy of the gene, they will not have FA, but they may pass the gene on to their children. People with only 1 copy of a recessive gene are called carriers of the gene.

If a child has FA, it means that he inherited 2 copies of the gene, 1 from each parent. Most parents do not know they carry the FA gene because they have only 1 copy, so they do not have symptoms.

With each pregnancy, 2 parents who carry the FA gene have a 1 in 4 chance of having a child with FA and a 1 in 2 chance of having a child who is a carrier.

If there is any history of FA in your family, you may want to seek genetic counselling.

Signs and symptoms of Friedreich's ataxia

Children with FA may show some or all of the following signs and symptoms:

- slow, clumsy walking
- wide stance, with feet spread far apart when standing
- abrupt, wild steps
- curved posture and curled feet
- no knee-jerk reflex
- heart troubles like racing (tachycardia) and extra beats (murmurs)
- difficulty speaking
- rapid eye movements when awake
- diabetes

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- hearing loss
- visual problems

There are several different treatments for symptoms of FA and its related conditions

There is no cure for FA, but there are treatments for some of the symptoms. Foot and spine problems are treated with braces and surgery. If a child with FA develops diabetes, it will be treated with insulin and a specialized diet.

Some studies show that antioxidants like vitamin E, CoQ10, and a CoQ10 drug called idebenone (Catena) can relieve muscle spasms and slow heart problems.

Related conditions

Some other conditions that can look like Friedreich's ataxia include: Ataxia with vitamin E deficiency; Abetalipoproteinemia, Refsun disease, Infantile onset spinocerebellar ataxia; Ataxia-telangiectasia; and many others. A specialist physician can help to make a definitive diagnosis.

Key points

- Friedreich's ataxia (FA) impairs muscle control.
- FA damages areas of the brain and spinal cord.
- FA gets worse, spreading from legs to arms to the torso.
- FA is a genetic disease. Ask for genetic counselling. If you and your partner both carry the gene, each child has a 1 in 4 chance of getting the disease.
- There is no cure for FA, but symptoms are treatable with antioxidants.

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