International Parkinson and Movement Disorder Society

Hereditary Ataxias: Essential Facts for Patients

What Is It?

Hereditary ataxias (HA) include a wide variety of inherited diseases where the main symptom is ataxia. Ataxia refers to uncoordinated, clumsy movements and walking problems with loss of balance. Changes in certain genes cause HA. In most cases, the disease affects more than one family member; however, sometimes there is no family history at all. In HA, ataxia is usually not the only symptom. Other neurological signs may include:

- Slowness and shaking (tremor)
- Twisting, turning or other uncontrolled movement (dystonia)
- Impaired sensation such as numbness, tingling and burning in legs and arms, with or without muscle weakness (neuropathy)

Other organs may also be affected, such as the heart (cardiomyopathy) or the eyes (retinopathy).

How is it Inherited?

There are four main ways ataxia can be inherited:

- **Autosomal dominant inheritance:** Only need to inherit one abnormal gene from either parent. A person with the abnormal gene has a 50% chance of passing the gene to a child.
- **Autosomal recessive inheritance:** Must inherit an abnormal gene from both parents. If each parent has one abnormal gene then each child has a 25% chance of inheriting both abnormal genes and developing the disease. Usually parents are just carriers and are healthy with no signs of the disease.
- **X-linked ataxia:** The abnormal gene is on the X chromosome and the gene passes from mother (usually healthy) to child.
- **Mitochondrial ataxia:** The disease develops when mitochondrial DNA has an abnormal gene. Mitochondria are parts of cells that produce energy. The disease is usually passed on by the mother.

What are Some Common Ataxias?

Autosomal dominantly inherited ataxias

Spinocerebellar ataxias (SCA): Currently, ~36 different gene abnormalities are known to cause SCAs. SCAs usually start in early to late adulthood. In addition to ataxia, you may experience:

- Uncontrolled, abnormal body movements
- Problems with attention, thinking and memory
- Changes in vision and/or abnormal eye movements
- Leg and arm numbness, tingling, burning (neuropathy)

Episodic ataxias: These ataxias start in childhood and include brief repeating events of ataxia and dizziness often triggered by exercise .

Autosomal recessively inherited ataxias

These diseases usually start before age 20. They are generally complex and disabling diseases. The most common type in Europe and North America is Friedreich's Ataxia. There is a genetic blood test that can confirm the diagnosis. Symptoms may include:

- Loss of sensation
- Abnormal spine curving (kyphoscoliosis)
- Heart problems (cardiomyopathy)
- Diabetes

X-linked ataxia: These diseases include Fragile X-associated Tremor-Ataxia (FXTAS) syndrome.

Mitochondrial ataxias: These diseases include:

- Myoclonic epilepsy ragged red fire (MERRF) syndrome
- Neuropathy, ataxia and retinitis pigmentosa (NARP)
- Kearns-Sayre syndrome
- POLG-related disorders (ataxia neuropathy spectrum)

How is it Diagnosed?

To diagnose ataxia, a doctor thoroughly reviews your symptoms. You can expect to:

- Share a three-generation family history
- Have a physical and neurological exam
- Get any required imaging (brain CT or MRI) and lab tests

The only way to get a definite diagnosis is through genetic testing from a blood or saliva sample. However, if the genetic test is negative you may still have a genetic disease since only some genes are known and can be tested for. Genetic counseling may help you understand the genetic risk for you and your family members to develop HA and to help with family planning.

Is There a Treatment?

Some rare hereditary ataxias have specific treatments. However, most ataxias are treated only for symptoms. You may improve your quality of life with:

- Physical therapy
- Speech therapy
- Occupational therapy
- Medical devices to solve specific problems

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