What is inherited ataxia?

Find out about the types, symptoms, causes, and diagnosis of inherited ataxia – a group of progressive neurological conditions that affect coordination, balance, and speech.

Inherited ataxia refers to a group of progressive neurological conditions caused by genetic changes that affect coordination, balance, speech, and motor control. These genetic mutations lead to the production of abnormal or faulty proteins, which can damage or kill nerve cells over time. This mainly affects the cerebellum – the part of the brain that controls movement - but other areas like the peripheral nerves and vestibular system can also be involved.

Common inherited ataxias include:

- Friedreich ataxia (FA) – typically begins in childhood or adolescence and can involve gait instability, heart complications, and scoliosis
- Spinocerebellar ataxias (SCA) Symptoms vary but can include tremor, abnormal eye movements, and sometimes cognitive changes. There are over 40 types of SCA, each with unique features
- CANVAS (Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome) – often presents in later adulthood with progressive imbalance, sensory loss, dizziness, and falls

Each person's experience of ataxia is different. Symptoms may change gradually over time, fluctuate with fatigue or illness, and impact daily life in unique ways.

Typical features and presentations

While all inherited ataxias involve a degree of impaired coordination and unsteady movement, specific features vary depending on the underlying cause and can include:

- Gait and balance issues – increased unsteadiness, tripping and falls
- Speech difficulties (dysarthria) – slurred or slow speech
- Limb incoordination – difficulty with writing, buttoning clothes, or using utensils
- Fatigue and slower movements are common across all types
- Vestibular symptoms – dizziness or blurred vision when turning the head
- Sensory changes – such as numbness or poor vibration sense
- Eye movement abnormalities – including nystagmus or difficulty tracking

Though inherited ataxias may look similar outwardly, their progression and associated symptoms can differ significantly based on the type.

What causes inherited ataxia?

Inherited ataxias result from genetic mutations, typically passed down in families. The pattern of inheritance may be:

- Autosomal recessive – as seen in Friedreich ataxia, where both parents carry
- Autosomal dominant – seen in SCAs, where one affected parent can pass on the condition
- Biallelic repeat expansions – associated with CANVAS, which is often adult-onset

The affected genes interfere with how neurons function and communicate, particularly in areas of the brain and body responsible for movement and balance**.**

Getting a diagnosis

Diagnosis is based on a combination of clinical assessment and investigations. This may include:

- A detailed neurological examination with a neurologist
- MRI to check for cerebellar atrophy or other structural changes
- **Genetic testing** to identify the type of ataxia
- Nerve conduction studies – especially important in FA and CANVAS, where neuropathy is common

Getting an accurate diagnosis can often take time, especially when symptoms are subtle or overlap with other conditions. A neurologist experienced in neurogenetics or hereditary movement disorders is best placed to guide this process.

Centre for Brain Research

The University of Auckland Centre for Brain Research are undertaking studies for inherited ataxias, we recommend you contact the university if you are interested in finding out more about participating in these studies.

Cann Charitable Trust is proud to partner with the Neurogenetics Research Clinic (NGRC) you can find more information about our collaboration here.