Ataxia

Frequently Asked Questions (FAQs)

INDIAN ACADEMY OF NEUROLOGY

A Public Information Initiative

Q. 1. What do you mean by ataxia?

Ans. Ataxia refers to a lack of muscle control during voluntary movements, e.g., walking like an intoxicated person. It may also affect speech, eye movements, or manifest as clumsiness in performing daily activities.

Persistent ataxia usually results from damage to the cerebellum - a part of the brain that controls muscle coordination. Many conditions can cause ataxia, including alcohol abuse, stroke, tumour, multiple sclerosis or inherited defective genes.

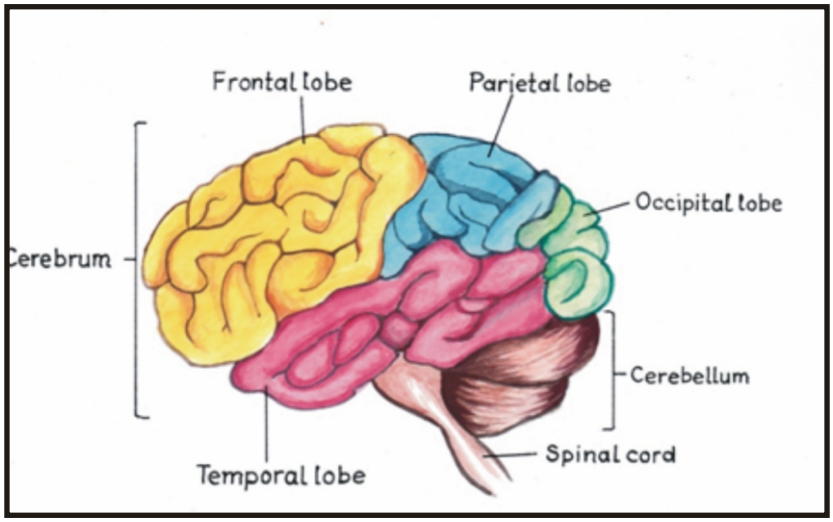

Figure: Sagittal section of the brain showing the cerebellum

Q.2. What are the symptoms of ataxia?

Ans. The following are the common symptoms of ataxias:

- Poor coordination, limb tremors which increase on reaching the target.
- Unsteady walk and a tendency to fall down.
- Difficulty with fine motor tasks such as, eating, writing, or buttoning a shirt.
- Altered (slurred, effortful and halting) speech.

Q. 3. When to consult a neurologist?

Ans. A neurologist should be consulted if:

- Ability to balance one's body is impaired.
- Coordination movements of upper and lower extremities are impaired.
- The person has difficulty in walking.
- The person experiences difficulty while speaking.

Q. 4. What are the causes of ataxia?

Ans. Damage, degeneration or loss of nerve cells in the part of the brain that controls muscle coordination,

i.e., the cerebellum, results in ataxia. The right side of the cerebellum controls coordination on the right side of the body, and the left side of the cerebellum controls coordination on the left.

Diseases that damage the spinal cord and peripheral nerves that connect the cerebellum to muscles also may cause ataxia.

Other causes of ataxia include:

- Head trauma
- Stroke
- Multiple sclerosis (MS)
- Chickenpox
- Tumour
- Toxic reaction
- Vitamin E, or vitamin B-12 deficiency

Q. 5. Is ataxia related to cancers (paraneoplastic syndromes)?

Ans. Some patients wtih lung, ovarian, breast or lymphatic cancers may experience unsteadiness due to ataxia. These are because of immune system's response to a cancerous tumor. Ataxia may appear many months or years before the cancer itself is diagnosed.

Q. 6. Can medications/toxins lead to ataxia as a side effect?

Ans. Certain medications, especially phenytoin, barbiturates, e.g., phenobarbital, and sedatives, e.g., benzodiazepines are responsible for ataxia as their side effects. Alcohol and drug intoxication, heavy metal poisoning, e.g., from lead or mercury, and solvent poisoning, e.g., from paint thinner can also cause ataxia.

Q. 7. What are hereditary/genetic ataxias?

Ans. A person can inherit genetic ataxia from either a dominant gene from one parent (autosomal dominant disorder) or a recessive gene from each parent (autosomal recessive disorder). In the latter case, it is possible that neither parent suffers from the disease (silent mutation), so there may be no obvious family history.

Different gene defects cause different types of ataxia, most of which are progressive. Each type results in poor coordination, but each has specific signs and symptoms.

Q. 8. Name few autosomal dominant ataxias?

Ans. These include:

- ***Spinocerebellar ataxias (SCA).*** Researchers have labelled more than 20 autosomal dominant ataxia genes, and the number is likely to grow. Cerebellar ataxia and cerebellar degeneration are common to all types, but other symptoms and signs, as well as age-of-onset may differ depending on the specific gene mutation.

- ***Episodic ataxia (EA).*** There are seven recognized types of ataxia that are episodic rather than progressive - EA1 through EA7. EA1 and EA2 are the most common. EA1 involves brief ataxic episodes that may last seconds or minutes. The episodes are triggered by stress, being startled, or by sudden movement. EA2 involves longer episodes, usually lasting from 30 minutes to six hours that are also aggravated by stress.

Q. 9. Which is the most common autosomal recessive hereditary ataxia?

Ans. **Friedreich's ataxia** is the most common autosomal recessive hereditary ataxia and is associated with damage to the cerebellum, spinal cord and peripheral nerves. Peripheral nerves carry signals from the brain and spinal cord to muscles. In most cases, symptoms and signs of the disease occur before the age of 25.

The rate of disease progression is variable. The first indication generally present is difficulty walking (gait ataxia). The condition typically progresses to the arms and the trunk. The muscles weaken and waste away over time, causing deformities, particularly, in the feet, lower legs, and hands.

Q.10. What is ataxia telangiectasia?

Ans. This rare progressive childhood disease causes degeneration in the brain and other body systems. The disease also causes immune system breakdown (immunodeficiency disease), which increases susceptibility to other diseases. It affects various organs.

Telangiectasias are tiny, red "spider" veins that may appear in the corners of the child's eyes, or on the ears and cheeks. Although they are characteristic of the disease, every affected child may not develop them. Delayed motor skill development, poor balance, and slurred speech are typically the first indications of the disease. Recurrent sinus and respiratory infections are common.
Children with ataxia-telangiectasia are at high risk of developing cancer, particularly leukemia or lymphoma.

Q.11. What is Wilson's disease?

Ans. People with this condition accumulate copper in their brain, liver and other organs, which can cause neurological problems including ataxia.

Q.12. What are the common tests or investigations that the neurologists may recommend for ataxia?

Ans: In someone with ataxia, the neurologist will look for a treatable cause. The neurologist conducts a thorough physical and neurological examination, including assessment of attention, concentration and memory, checking of vision, hearing, balance, coordination and reflexes.

The doctor may also request certain laboratory tests:

- ***Imaging studies:*** A computerized tomography (CT) scan or magnetic resonance imaging (MRI) of the brain may help determine the likely cause.
- ***Lumbar puncture (spinal tap):*** A needle is inserted into the lower back (lumbar region) between two lumbar bones (vertebrae) to remove a sample of cerebrospinal fluid. The fluid, which surrounds and protects the brain and spinal cord, is sent to a laboratory for testing.

- ***Genetic testing:*** The neurologist may ask for genetic testing to determine whether the parents or the affected child have the gene mutation that causes one of the hereditary ataxic conditions. Gene tests are available for many but not all of the hereditary ataxias.

Q.13. What are the treatments and/or prevention for a patient with ataxia?

Ans. There is no specific treatment for ataxia. Sometimes, treating the underlying cause resolves the ataxia. In other cases, e.g., ataxia that is associated with chickenpox or other viral infections, it is likely to resolve on its own over time. The neurologist may recommend adaptive devices or therapies to help with ataxia.

People with ataxia should avoid alcohol and exposure to heavy metals. In case of hereditary ataxias, they may need pre-marital counselling for knowing the severity, age-of-onset in subsequent generations, and future risk of development of ataxia. The patients with a family history of ataxia may not develop all the symptoms because the defective gene mutation might not always manifest in subsequent generations.

Q.14. Name some adaptive devices for a person with ataxia.

Ans. For ataxia caused by conditions, such as multiple sclerosis or cerebral palsy, it may not be treatable. In such cases, the neurologist may recommend adaptive devices such as:

- Canes or walkers for walking.
- Modified utensils for eating.
- Communication aids for speaking.

Q.15. What therapies may benefit a person with ataxia?

Ans. The patient with ataxia might benefit from certain therapies, including:

- Physical therapy to help him build strength and enhance mobility.
- Occupational therapy to help the person with daily-living tasks, such as feeding, dressing etc.
- Speech therapy to improve speech.
- Stem-cell and genetic therapies are still in an experimental stage.

Disclaimer:
This brochure is for the general information of the public and the patients. People should not self-medicate themselves with the medicines and treatments mentioned here. Before taking any of the medications mentioned in the information brochure, please consult your neurologist.

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