

Autosomal recessive axonal neuropathy with neuromyotonia

Description

Autosomal recessive axonal neuropathy with neuromyotonia is a disorder that affects the peripheral nerves. Peripheral nerves connect the brain and spinal cord to muscles and to sensory cells that detect sensations such as touch, pain, heat, and sound.

Axonal neuropathy, a characteristic feature of this condition, is caused by damage to a particular part of peripheral nerves called axons, which are the extensions of nerve cells (neurons) that transmit nerve impulses. In people with autosomal recessive axonal neuropathy with neuromyotonia, the damage primarily causes progressive weakness and wasting (atrophy) of muscles in the feet, legs, and hands. Muscle weakness may be especially apparent during exercise (exercise intolerance) and can lead to an unusual walking style (gait), frequent falls, and joint deformities (contractures) in the hands and feet. In some affected individuals, axonal neuropathy also causes decreased sensitivity to touch, heat, or cold, particularly in the lower arms or legs.

Another feature of this condition is neuromyotonia (also known as Isaac syndrome). Neuromyotonia results from overactivation (hyperexcitability) of peripheral nerves, which leads to delayed relaxation of muscles after voluntary tensing (contraction), muscle cramps, and involuntary rippling movement of the muscles (myokymia).

Frequency

Autosomal recessive axonal neuropathy with neuromyotonia is a rare form of inherited peripheral neuropathy. This group of conditions affects an estimated 1 in 2,500 people. The prevalence of autosomal recessive axonal neuropathy with neuromyotonia is unknown.

Causes

Autosomal recessive axonal neuropathy with neuromyotonia is caused by mutations in the *HINT1* gene. This gene provides instructions for making a protein that is involved in the function of the nervous system; however its specific role is not well understood. Laboratory studies show that the HINT1 protein has the ability to carry out a chemical reaction called hydrolysis that breaks down certain molecules; however, it is not known what effects the reaction has in the body.

HINT1 gene mutations that cause autosomal recessive axonal neuropathy with

neuromyotonia lead to production of a HINT1 protein with little or no function. Sometimes the abnormal protein is broken down prematurely. Researchers are working to determine how loss of functional HINT1 protein affects the peripheral nerves and leads to the signs and symptoms of this condition.

[Learn more about the gene associated with Autosomal recessive axonal neuropathy with neuromyotonia](#)

- HINT1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ARAN-NM
- Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia
- Autosomal recessive neuromyotonia and axonal neuropathy
- Gamstorp-Wohlfart syndrome
- Myokymia, myotonia, and muscle wasting
- NMAN

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Autosomal recessive axonal neuropathy with neuromyotonia (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C5700127/>)

Genetic and Rare Diseases Information Center

- Autosomal recessive axonal neuropathy with neuromyotonia (<https://rarediseases.info.nih.gov/diseases/12353/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Autosomal recessive axonal neuropathy with neuromyotonia%22>)

Catalog of Genes and Diseases from OMIM

- NEUROMYOTONIA AND AXONAL NEUROPATHY, AUTOSOMAL RECESSIVE; NMAN (<https://omim.org/entry/137200>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28axonal+neuropathy+with+neuromyotonia%29+OR+%28%28neuropathy%29+AND+%28HINT1%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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