Distal myopathy 2

Description

Distal myopathy 2 is a condition characterized by weakness of specific muscles that begins in adulthood. It is a form of muscular dystrophy that specifically involves muscles in the throat, lower legs, and forearms. Muscles farther from the center of the body, like the muscles of the lower legs and forearms, are known as distal muscles.

Muscle weakness in the ankles is usually the first symptom of distal myopathy 2. The weakness can also affect muscles in the hands, wrists, and shoulders. At first, the muscle weakness may be on only one side of the body, but both sides are eventually involved. This muscle weakness can slowly worsen and make actions like walking and lifting the fingers difficult.

Another characteristic feature of distal myopathy 2 is weakness of the vocal cords and throat. This weakness initially causes the voice to sound weak or breathy (hypophonic). Eventually, the voice becomes gurgling, hoarse, and nasal. The weakness can also cause difficulty swallowing (dysphagia).

Frequency

The prevalence of distal myopathy 2 is unknown. At least two families with the condition have been described in the scientific literature.

Causes

A mutation in the *MATR3* gene has been identified in people with distal myopathy 2. This gene provides instructions for making a protein called matrin 3, which is found in the nucleus of the cell as part of the nuclear matrix. The nuclear matrix is a network of proteins that provides structural support for the nucleus and aids in several important nuclear functions.

The function of the matrin 3 protein is unknown. This protein can attach to (bind) RNA, which is a chemical cousin of DNA. Some studies indicate that matrin 3 binds and stabilizes a type of RNA called messenger RNA (mRNA), which provides the genetic blueprint for proteins. Matrin 3 may also bind certain abnormal RNAs that might lead to nonfunctional or harmful proteins, thereby blocking the formation of such proteins. Other studies suggest that the matrin 3 protein may be involved in cell survival.

The *MATR3* gene mutation identified in people with distal myopathy 2 changes a single protein building block (amino acid) in the matrin 3 protein. The effect of this mutation on the function of the protein is unknown, although one study suggests that the mutation may change the location of the protein in the nucleus. Researchers are working to determine how this gene mutation leads to the signs and symptoms of distal myopathy 2.

Learn more about the gene associated with Distal myopathy 2

- MATR3

Inheritance

Distal myopathy 2 is inherited in an autosomal dominant pattern, which means one copy of the altered *MATR3* gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Distal myopathy with vocal cord and pharyngeal signs
- Distal myopathy with vocal cord weakness
- Matrin 3 distal myopathy
- MPD2
- Myopathia distalis type 2
- VCPDM
- Vocal cord and pharyngeal weakness with distal myopathy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Amyotrophic lateral sclerosis type 21 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3807521/)

Genetic and Rare Diseases Information Center

- Vocal cord and pharyngeal distal myopathy (https://rarediseases.info.nih.gov/diseases/1887/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- AMYOTROPHIC LATERAL SCLEROSIS 21; ALS21 (https://omim.org/entry/606070)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Distal+Myopathies%5BMAJR%5D%29+OR+%28vcpdm%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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