Hereditary neuropathy with liability to pressure palsies

Description

Hereditary neuropathy with liability to pressure palsies is a disorder that affects peripheral nerves. These nerves connect the brain and spinal cord to muscles and sensory cells that detect touch, pain, and temperature. In people with this disorder, the peripheral nerves are unusually sensitive to pressure, such as the pressure that occurs when carrying heavy grocery bags, leaning on an elbow, or sitting without changing position, particularly with crossed legs. These activities would not normally cause sensation problems in people without the disorder.

Hereditary neuropathy with liability to pressure palsies is characterized by recurrent episodes of numbness, tingling, and loss of muscle function (palsy) in the region associated with the affected nerve, usually an arm, hand, leg, or foot. An episode can last from several minutes to several months, but recovery is usually complete. Repeated incidents, however, can cause permanent muscle weakness or loss of sensation. This disorder is also associated with pain in the limbs, especially the hands.

A pressure palsy episode results from pressure on a single nerve, and any peripheral nerve can be affected. Although episodes often recur, they can affect different nerves. The most common problem sites involve nerves in the wrists, elbows, and knees. The fingers, shoulders, hands, feet, and scalp can also be affected. Many people with this disorder experience carpal tunnel syndrome, which occurs when a nerve in the wrist (the median nerve) is involved. Carpal tunnel syndrome is characterized by numbness, tingling, and weakness in the hand and fingers. An episode in the hand may affect fine motor activities such as writing, opening jars, and fastening buttons. An episode of nerve compression in the knee can lead to a condition called foot drop, which makes walking, climbing stairs, or driving difficult or impossible.

The symptoms of hereditary neuropathy with liability to pressure palsies usually begin during adolescence or early adulthood but may develop anytime from childhood to late adulthood. Symptoms vary in severity; many people never realize they have the disorder, while some people experience prolonged disability. Hereditary neuropathy with liability to pressure palsies does not affect life expectancy.

Frequency

Hereditary neuropathy with liability to pressure palsies is estimated to occur in 2 to 5 per 100,000 individuals.

Causes

Hereditary neuropathy with liability to pressure palsies is most commonly caused by the deletion of one copy of the *PMP22* gene. This loss reduces the amount of PMP22 protein produced. Other *PMP22* gene mutations that lead to a reduction of PMP22 protein can also cause the condition.

The PMP22 protein is a component of myelin, a protective substance that covers nerves and promotes the efficient transmission of nerve impulses. Studies suggest that the PMP22 protein is particularly important in protecting nerves from physical pressure, helping them restore their structure after compression. Compression can interrupt nerve signaling, leading to the sensation commonly referred to as a limb "falling asleep." The ability of nerves to recover from normal, day-to-day pressure, for example when sitting for long periods, keeps the limbs from constantly losing sensation.

The consequences of *PMP22* gene mutations are not clearly understood. A shortage of PMP22 protein appears to make nerves less able to recover from compression, which impairs the transmission of nerve impulses, causing the signs and symptoms of hereditary neuropathy with liability to pressure palsies.

Learn more about the gene associated with Hereditary neuropathy with liability to pressure palsies

- PMP22

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Compression neuropathy
- Entrapment neuropathy
- Familial pressure sensitive neuropathy
- Hereditary motor and sensory neuropathy
- Hereditary pressure sensitive neuropathy
- HNPP
- Inherited tendency to pressure palsies
- Tomaculous neuropathy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hereditary liability to pressure palsies (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0393814/)

Genetic and Rare Diseases Information Center

- Hereditary neuropathy with liability to pressure palsies (https://rarediseases.info.nih.gov/diseases/5221/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Hereditary neuropathy with liability to pressure palsies%22)

Catalog of Genes and Diseases from OMIM

- NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP (https://omim.org/entry/162500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Hereditary+Motor+and+Sensory+Neuropathies%5BMAJR%5D%29+AND+%28%28hnpp%5BTIAB%5D%29+OR+%28tomaculous+neuropathy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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