Hereditary sensory and autonomic neuropathy type V

Description

Hereditary sensory and autonomic neuropathy type V (HSAN5) is a condition that primarily affects the sensory nerve cells (sensory neurons), which transmit information about sensations such as pain, temperature, and touch. These sensations are impaired in people with HSAN5.

The signs and symptoms of HSAN5 appear early, usually at birth or during infancy. People with HSAN5 lose the ability to feel pain, heat, and cold. Deep pain perception, the feeling of pain from injuries to bones, ligaments, or muscles, is especially affected in people with HSAN5. Because of the inability to feel deep pain, affected individuals suffer repeated severe injuries such as bone fractures and joint injuries that go unnoticed. Repeated trauma can lead to a condition called Charcot joints, in which the bones and tissue surrounding joints are destroyed.

Frequency

HSAN5 is very rare. Only a few people with the condition have been identified.

Causes

Mutations in the *NGF* gene cause HSAN5. The *NGF* gene provides instructions for making a protein called nerve growth factor beta (NGFβ) that is important in the development and survival of nerve cells (neurons), including sensory neurons. The NGFβ protein functions by attaching (binding) to its receptors, which are found on the surface of neurons. Binding of the NGFβ protein to its receptor transmits signals to the cell to grow and to mature and take on specialized functions (differentiate). This binding also blocks signals in the cell that initiate the process of self-destruction (apoptosis). Additionally, NGFβ signaling plays a role in pain sensation. Mutation of the *NGF* gene leads to the production of a protein that cannot bind to the receptor and does not transmit signals properly. Without the proper signaling, sensory neurons die and pain sensation is altered, resulting in the inability of people with HSAN5 to feel pain.

Learn more about the gene associated with Hereditary sensory and autonomic neuropathy type V

- NGF

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

- Congenital insensitivity to pain
- Congenital sensory neuropathy with selective loss of small myelinated fibers
- Hereditary sensory and autonomic neuropathy, type 5
- HSAN type V
- HSAN V
- HSAN5

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital sensory neuropathy with selective loss of small myelinated fibers (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0020075/)

Genetic and Rare Diseases Information Center

- Hereditary sensory and autonomic neuropathy type 5 (https://rarediseases.info.nih.gov/diseases/12328/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE V; HSAN5 (https://omim.org/entry/608654)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28hereditary+sensory+and+autonomic+neuropathy+type+v%5BTIAB%5D%29+OR+%28hsan+type+v%5BTIAB%5D%29+OR+%28hsan+v%5BTIAB%5D%29+OR+%28hsan5%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+day

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