# MPZ gene

myelin protein zero

## Normal Function

The *MPZ* gene provides instructions for making a protein called myelin protein zero. It is the most abundant protein in myelin, a protective substance that covers nerves and promotes the efficient transmission of nerve impulses. Myelin protein zero is produced by specialized cells called Schwann cells, which wrap around and insulate 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. Myelin protein zero is required for the proper formation and maintenance of myelin. This protein is an adhesion molecule, which means it acts like molecular glue. It plays a role in tightly packing the myelin around nerve cells (myelin compaction).

## Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

Researchers have identified more than 120 *MPZ* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 1B. Charcot-Marie-Tooth syndrome is a disorder characterized by muscle weakness and sensory problems, especially in the hands and feet. Many of the *MPZ* gene mutations that cause Charcot-Marie-Tooth syndrome change single protein building blocks (amino acids) in myelin protein zero. Other mutations lead to a protein that is missing one or more amino acids. The altered myelin protein zero probably cannot interact properly with other myelin components, which may disrupt myelin compaction. As a result, peripheral nerves cannot trigger muscle movement or relay information from sensory cells back to the brain, leading to the weakness and sensory problems characteristic of Charcot-Marie-Tooth disease.

Some *MPZ* gene mutations cause a severe form of type 1B Charcot-Marie-Tooth disease. Symptoms begin during infancy or early childhood and include delayed development of motor skills such as walking. This form of Charcot-Marie-Tooth disease is sometimes called Dejerine-Sottas syndrome, congenital hypomyelination, or Roussy-Levy syndrome. Researchers believe that the *MPZ* gene mutations that cause the severe form of the disorder probably disrupt the formation of myelin during early development.

Several mutations in the *MPZ* gene cause other forms of Charcot-Marie-Tooth disease

known as type 2I, type 2J, and dominant intermediate D. These forms of Charcot-Marie-Tooth disease, which often do not become evident until adulthood, affect the specialized outgrowths from nerve cells (axons) that transmit impulses to muscles and other nerve cells. People with type 2J Charcot-Marie-Tooth disease may also have hearing loss and abnormalities in the opening of the eye through which light passes (the pupil). It is unclear how *MPZ* gene mutations cause these abnormalities.

## Other Names for This Gene

- CMT1B
- CMT2I
- CMT2J
- HMSN1B
- MPP
- myelin glycoprotein P-zero
- myelin peripheral protein
- myelin protein zero (Charcot-Marie-Tooth neuropathy 1B)
- MYP0_HUMAN
- P0 Glycoprotein
- P0 Protein

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MPZ (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4359[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MPZ%5BTIAB%5D%29+OR+%28myelin+protein+zero%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MYELIN PROTEIN ZERO; MPZ (https://omim.org/entry/159440)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4359)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MPZ[gene])

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## Genomic Location

The *MPZ* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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