**Genetic Conditions**

**Your Guide to Understanding Genetic Conditions**

**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. Specialized cells called Schwann cells, which wrap around and insulate nerves, are the only cells that make myelin protein zero. Schwann cells are part of the peripheral nervous system which connects 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. Many of these mutations change single protein building blocks (amino acids) in myelin protein zero. Other *MPZ* gene 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 the formation and maintenance of myelin. As a result, peripheral nerve cells cannot activate muscles used for 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. The early-onset forms of Charcot-Marie-Tooth disease are 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.

**Chromosomal Location**

Cytogenetic Location: 1q23.3, which is the long (q) arm of chromosome 1 at position 23.3

Molecular Location: base pairs 161,303,594 to 161,309,972 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- CMT1B
- 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**

**Educational Resources**

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1205

- Charcot-Marie-Tooth Neuropathy Type 2
  https://www.ncbi.nlm.nih.gov/books/NBK1285

Scientific Articles on PubMed

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

OMIM

- MYELIN PROTEIN ZERO
  http://omim.org/entry/159440

Research Resources

- ClinVar

- HGNC Gene Family: Ig-like cell adhesion molecule family
  https://www.genenames.org/cgi-bin/genefamilies/set/1410

- HGNC Gene Family: V-set domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/590

- HGNC Gene Symbol Report

- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=2

- NCBI Gene

- UniProt
  http://www.uniprot.org/uniprot/P25189
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301462

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11835375

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18337304

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15298082

- OMIM: MYELIN PROTEIN ZERO
  http://omim.org/entry/159440

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19293842
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2986589/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16775378

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12402337

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16414078

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8816708

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14685682