PMM2 gene

phosphomannomutase 2

Normal Function

The *PMM2* gene provides instructions for making an enzyme called phosphomannomutase 2 (PMM2). This enzyme is involved in a process called glycosylation, which attaches groups of sugar molecules (oligosaccharides) to proteins. Oligosaccharides are made up of many small sugar molecules that are attached to one another in a long chain. Glycosylation modifies proteins so they can perform a wider variety of functions. In one of the early steps of glycosylation, the PMM2 enzyme converts a molecule called mannose-6-phosphate to mannose-1-phosphate. Subsequently, mannose-1-phosphate is converted into GDP-mannose, which can transfer its small sugar molecule called mannose to the growing oligosaccharide chain. Once the correct number of small sugar molecules are linked together to form the oligosaccharide, it can be attached to a protein.

Health Conditions Related to Genetic Changes

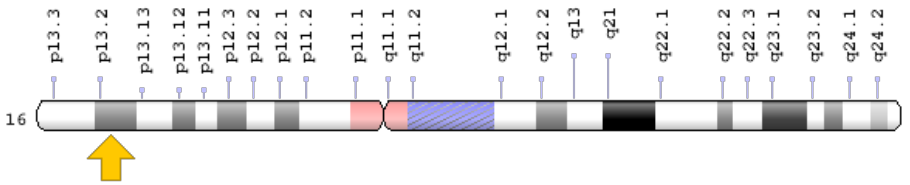
PMM2-congenital disorder of glycosylation

More than 115 mutations in the *PMM2* gene have been found to cause *PMM2*-congenital disorder of glycosylation (*PMM2*-CDG, also known as congenital disorder of glycosylation type Ia). This is a severe condition that is characterized by developmental delay, weak muscle tone (hypotonia), abnormal distribution of fat, and various other signs and symptoms. The mutations that cause *PMM2*-CDG change the structure of the PMM2 enzyme in different ways; however, all of the mutations appear to result in reduced enzyme activity. Decreased activity of the PMM2 enzyme leads to a shortage of GDP-mannose within cells. As a result, there is not enough activated mannose to form oligosaccharides. Glycosylation cannot proceed normally because incorrect oligosaccharides are produced. The signs and symptoms in *PMM2*-CDG are likely due to the production of abnormally glycosylated proteins in many organs and tissues.

Chromosomal Location

Cytogenetic Location: 16p13.2, which is the short (p) arm of chromosome 16 at position 13.2

Molecular Location: base pairs 8,797,813 to 8,849,326 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CDG1a
- phosphomannomutase
- PMM
- PMM2_HUMAN

Additional Information & Resources

Educational Resources

- EUROGLYCANET
  http://www.euroglycanet.org/
- Molecular Biology of the Cell (fourth edition, 2002): What is the purpose of glycosylation?
  https://www.ncbi.nlm.nih.gov/books/NBK26941/#A2354

Clinical Information from GeneReviews

- PMM2-CDG (CDG-Ia)
  https://www.ncbi.nlm.nih.gov/books/NBK1110

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PMM2%5BTIAB%5D%29+OR+%28phosphomannomutase+2%5BTIAB%5D%29%29+OR+%28CDG1a%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PHOSPHOMANNOMUTASE 2
  http://omim.org/entry/601785

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PMM2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9115
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5373
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5373
- UniProt
  https://www.uniprot.org/uniprot/O15305

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