

PGM3 gene

phosphoglucomutase 3

Normal Function

The *PGM3* gene provides instructions for making an enzyme called phosphoglucomutase 3 (PGM3). This enzyme is involved in a process called glycosylation. During this process, complex chains of sugar molecules (oligosaccharides) are added to proteins and fats (lipids). Glycosylation modifies proteins and lipids so they can perform a wider variety of functions.

The PGM3 enzyme converts a molecule called N-acetylglucosamine-6-phosphate into a different molecule called N-acetylglucosamine-1-phosphate. This conversion is required to make a sugar called uridine diphosphate-N-acetylglucosamine (UDP-GlcNAc), which is needed to transfer sugars to growing oligosaccharides during glycosylation.

Health Conditions Related to Genetic Changes

PGM3-congenital disorder of glycosylation

At least 16 mutations in the *PGM3* gene have been found to cause *PGM3*-congenital disorder of glycosylation (*PGM3*-CDG). This condition primarily affects the immune system but can also involve other areas of the body. Affected individuals often have impaired immune function (immune deficiency), distinct facial features, intellectual disability, and delayed development.

PGM3 gene mutations result in the production of an enzyme with reduced activity. Without a properly functioning enzyme, there is a shortage of UDP-GlcNAc and glycosylation cannot proceed normally. The wide variety of signs and symptoms in *PGM3*-CDG are likely due to impaired glycosylation of proteins and lipids that are needed for the normal function of many organs and tissues. Immune system proteins are highly dependent on glycosylation to function normally, which likely explains why people with *PGM3*-CDG have immune deficiency.

Other Names for This Gene

- acetylglucosamine phosphomutase
- AGM1
- N-acetylglucosamine-phosphate mutase 1

- PAGM

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PGM3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5238\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5238[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PGM3%5BTIAB%5D%29+OR+%28phosphoglucomutase+3%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%29%29>)

Catalog of Genes and Diseases from OMIM

- PHOSPHOGLUCOMUTASE 3; PGM3 (<https://omim.org/entry/172100>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5238>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PGM3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PGM3[gene]))

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Genomic Location

The *PGM3* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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