G6PD gene

glucose-6-phosphate dehydrogenase

Normal Function

The *G6PD* gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. This enzyme, which is active in virtually all types of cells, is involved in the normal processing of carbohydrates. It plays a critical role in red blood cells, which carry oxygen from the lungs to tissues throughout the body. This enzyme helps protect red blood cells from damage and premature destruction.

Glucose-6-phosphate dehydrogenase is responsible for the first step in the pentose phosphate pathway, a series of chemical reactions that convert glucose (a type of sugar found in most carbohydrates) to another sugar, ribose-5-phosphate. Ribose-5-phosphate is an important component of nucleotides, which are the building blocks of DNA and its chemical cousin RNA. This chemical reaction produces a molecule called NADPH, which plays a role in protecting cells from potentially harmful molecules called reactive oxygen species. These molecules are byproducts of normal cellular functions. Reactions involving NADPH produce compounds that prevent reactive oxygen species from building up to toxic levels within cells. The production of NADPH by glucose-6-phosphate dehydrogenase is essential in red blood cells, which are particularly susceptible to damage by reactive oxygen species because they lack other NADPH-producing enzymes.

Health Conditions Related to Genetic Changes

Glucose-6-phosphate dehydrogenase deficiency

More than 200 variants (also called mutations) that cause glucose-6-phosphate dehydrogenase deficiency have been identified in the *G6PD* gene. Almost all of these variants lead to changes in single building blocks (amino acids) in the glucose-6-phosphate dehydrogenase enzyme. These changes disrupt the normal structure and function of the enzyme or reduce the amount of the enzyme produced in cells.

Without enough functional glucose-6-phosphate dehydrogenase, red blood cells are unable to protect themselves from the damaging effects of reactive oxygen species. The damaged cells are likely to rupture and break down prematurely (undergo hemolysis). Factors such as infections, certain drugs, and ingesting fava beans can increase the levels of reactive oxygen species, causing red blood cells to undergo hemolysis faster

than the body can replace them. This loss of red blood cells causes the signs and symptoms of hemolytic anemia, which is a characteristic feature of glucose-6-phosphate dehydrogenase deficiency.

Other Names for This Gene

- G6PD1
- G6PD_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of G6PD (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2539[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28G6PD%5BTIAB%5D%29+OR+%28glucose-6-phosphate+dehydrogenase%5BTIAB%5D%29%29+AND+%28%28glucose-6-phosphate+dehydrogenase%5BMAJR%5D%29+OR+%28glucosephosphate+dehydrogenase%5BMAJR%5D%29+OR+%28d-glucose-6-phosphate:nadp++1-oxidoreductase%5BMAJR%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

- GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD (https://omim.org/entry/305900)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2539)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=G6PD[gene])

References

- Biochemistry (fifth edition, 2002): Glucose 6-Phosphate Dehydrogenase Plays a Key Role in Protection Against Reactive Oxygen Species (https://www.ncbi.nlm.nih.gov/books/NBK22389/)
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- Efferth T, Schwarzl SM, Smith J, Osieka R. Role of glucose-6-phosphatedehydrogenase for oxidative stress and apoptosis. Cell Death Differ. 2006Mar;13(3):527-8; author reply 529-30. doi: 10.1038/sj.cdd.4401807. No abstractavailable. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16311511)
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- Verrelli BC, McDonald JH, Argyropoulos G, Destro-Bisol G, Froment A,Drousiotou A, Lefranc G, Helal AN, Loiselet J, Tishkoff SA. Evidence forbalancing selection from nucleotide sequence analyses of human G6PD. Am J HumGenet. 2002 Nov;71(5):1112-28. doi: 10.1086/344345. Epub 2002 Oct 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12378426) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC385087/)

Genomic Location

The *G6PD* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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