HPRT1 gene

hypoxanthine phosphoribosyltransferase 1

Normal Function

The *HPRT1* gene provides instructions for producing an enzyme called hypoxanthine phosphoribosyltransferase 1. This enzyme allows cells to recycle purines, a type of building block of DNA and its chemical cousin RNA. Manufacturing purines uses more energy and takes more time than recycling purines, which makes recycling these molecules more efficient. Recycling purines ensures that cells have a plentiful supply of building blocks for the production of DNA and RNA. The process of recycling purines is also known as the purine salvage pathway.

Health Conditions Related to Genetic Changes

Lesch-Nyhan syndrome

More than 200 mutations in the *HPRT1* gene have been found to cause Lesch-Nyhan syndrome. These mutations include changes in single DNA building blocks (nucleotides) or insertions or deletions of small amounts of DNA within the gene. These changes result in either nonfunctional or very low-function hypoxanthine phosphoribosyltransferase 1. Under these conditions, uric acid, a waste product of purine breakdown, accumulates in the body and can cause gouty arthritis (arthritis caused by uric acid in the joints), kidney stones, and bladder stones. It is unclear how this enzyme deficiency causes the neurological and behavioral problems characteristic of Lesch-Nyhan syndrome.

Other disorders

Certain mutations in the *HPRT1* gene can also cause a condition featuring gouty arthritis called HPRT-related gout, previously known as Kelley-Seegmiller syndrome. Individuals with this condition have lower than normal levels of hypoxanthine phosphoribosyltransferase 1. Kidney problems commonly occur in people with this condition because a buildup of uric acid crystals can form kidney stones. Rarely, this condition will cause problems with the nervous system.

Other Names for This Gene

- Guanine Phosphoribosyltransferase

- HGPRT
- HGPRTase
- HOX5.4
- HPRT
- HPRT_HUMAN
- HPRTase
- hypoxanthine phosphoribosyltransferase 1 (Lesch-Nyhan syndrome)
- Hypoxanthine-Guanine Phosphoribosyltransferase
- IMP Pyrophosphorylase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HPRT1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3251[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28HPRT1%5BTI%5D%29+OR+%28%28Guanine+Phosphoribosyltransferase%5BTIAB%5D%29+OR+%28HGPRT%5BTIAB%5D%29+OR+%28hypoxanthine+phosphoribosyltransferase+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- HYPERURICEMIA, HPRT-RELATED; HRH (https://omim.org/entry/300323)
- HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1; HPRT1 (https://omim.org/entry/308000)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3251)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HPRT1[gene])

References

- Duan J, Nilsson L, Lambert B. Structural and functional analysis of mutationsat the human hypoxanthine phosphoribosyl transferase (HPRT1) locus. Hum Mutat.2004 Jun;23(6):599-611. doi: 10.1002/humu.20047. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15146465)

- Jinnah HA, De Gregorio L, Harris JC, Nyhan WL, O'Neill JP. The spectrum ofinherited mutations causing HPRT deficiency: 75 new cases and a review of 196previously reported cases. Mutat Res. 2000 Oct;463(3):309-26. doi:10.1016/s1383-5742(00)00052-1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11018746)
- Nyhan WL. Dopamine function in Lesch-Nyhan disease. Environ Health Perspect. 2000 Jun;108 Suppl 3(Suppl 3):409-11. doi: 10.1289/ehp.00108s3409. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10852837) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1637829/)
- Nyhan WL. The recognition of Lesch-Nyhan syndrome as an inborn error of purinemetabolism. J Inherit Metab Dis. 1997 Jun;20(2):171-8. doi:10.1023/a:1005348504512. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9211189)
- Puig JG, Torres RJ, Mateos FA, Ramos TH, Arcas JM, Buno AS, O'Neill P. Thespectrum of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency. Clinical experience based on 22 patients from 18 Spanish families. Medicine(Baltimore). 2001 Mar;80(2):102-12. doi: 10.1097/00005792-200103000-00003. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11307586)
- Sculley DG, Dawson PA, Emmerson BT, Gordon RB. A review of the molecular basisof hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency. Hum Genet.1992 Nov;90(3):195-207. doi: 10.1007/BF00220062. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/1487231)

Genomic Location

The *HPRT1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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