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+%28Guanine+Phosphoribosyltransferase%5BTIAB%5D%29+OR+%28HGPRT%5BTIAB%5D%29+OR+%28hypoxanthine+phosphoribosyltransferase%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM
• HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1 (https://omim.org/entry/308000)
• KELLEY-SEEGMILLER SYNDROME (https://omim.org/entry/300323)

Gene and Variant Databases

References


Genomic Location

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

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