

Lesch-Nyhan Syndrome

Lesch-Nyhan syndrome is an X-linked recessive disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase (HGPRT). HGPRT is a transferase enzyme that catalyzes the conversion of hypoxanthine to inosine monophosphate (IMP) and guanine to guanosine monophosphate (GMP). This enzyme plays a key role in the purine salvage pathway, and a deficiency causes a buildup of uric acid in the body. Therefore, individuals with Lesch-Nyhan syndrome suffer from severe gout and display neurological signs, including moderate intellectual disability. A particularly notable feature of Lesch-Nyhan syndrome is self-mutilation, commonly manifested by lip and finger biting. Other neurological symptoms include choreoathetosis and facial grimacing.



PLAY PICMONIC

ETIOLOGY

X-linked Recessive

X-suit with Recessive-chocolate

This disease is inherited in an X-linked recessive pattern.

Pathophysiology

Absence of HGPRT

Anti-sign on HGPRt-curtain

HGPRT is a transferase enzyme that catalyzes the conversion of hypoxanthine to inosine monophosphate (IMP) and guanine to guanosine monophosphate (GMP). This enzyme plays a key role in the purine salvage pathway.

Hypoxanthine to IMP

Hippo-xylophone Connected to IMP

HGPRT is a transferase enzyme that catalyzes the conversion of hypoxanthine to inosine monophosphate (IMP) in the purine salvage pathway.

Guanine to GMP

Guano to GruMP

HGPRT is a transferase enzyme that catalyzes the conversion of guanine to guanosine monophosphate (GMP) in the purine salvage pathway.

PRPP is Increased

PrePPy-boy

Phosphoribosyl pyrophosphate (PRPP) accumulates due to defective HGPRT and stimulates the de novo purine synthesis pathway.

Excess Uric Acid

Unicorn Acidic-lemon

Because the purine salvage pathway is defective, cell breakdown products cannot be reused and are instead degraded into uric acid.

Signs & Symptoms

Gout

Gout-goat

Gout is caused by excess uric acid, leading to the deposition of urate crystals in joints, causing inflammation and pain. It is commonly seen in Lesch-Nyhan syndrome.

Intellectual Disability

[Book Covered in Tar](#)

Individuals with Lesch-Nyhan syndrome exhibit severe cognitive impairment, often with deficits in adaptive behaviors such as communication and social skills. These deficits contribute to developmental delay.

Self-mutilation

[Self-hitting](#)

A hallmark feature of Lesch-Nyhan syndrome is self-mutilation, commonly manifested by lip and finger biting.

Choreoathetosis

[Korean-Thor](#)

Choreoathetosis presents involuntary movements with features of chorea, which are irregular, dance-like movements, and athetosis, which is characterized by a twisting and writhing motion.

Treatment

Allopurinol

[Aloe-piranha](#)

Allopurinol is a medication that inhibits xanthine oxidase. Because this enzyme is involved in uric acid production, allopurinol helps to manage hyperuricemia by decreasing uric acid.

Febuxostat

[Fat-boxer](#)

Febuxostat is a xanthine oxidase inhibitor. Blocking xanthine oxidase reduces uric acid production. It serves as an alternative to allopurinol in managing hyperuricemia associated with Lesch-Nyhan syndrome.