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.



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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.

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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
Choreoatheotosis 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.

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