

## 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 buildup of uric acid in the body. Therefore, individuals with Lesch-Nyhan syndrome suffer from severe gout and display neurological signs including moderate mental retardation. A particularly striking 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

### X-linked Recessive

[X-suit with Recessive-chocolate](#)

This disease is inherited in an X linked recessive pattern.

### 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 GiMP](#)

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

### Gout

[Gout-goat](#)

Gout is caused by excess uric acid and is commonly seen in Lesch-Nyhan syndrome.

### Retardation

[Read-tarred book](#)

Individuals with Lesch-Nyhan syndrome display significantly impaired cognitive functioning with deficits in two or more adaptive behaviors.

### Self-mutilation

[Self-hitting](#)

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

## **Choreoathetosis**

### **Korean-Thor**

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

## **PRPP is increased**

### **PRePPy-boy**

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