

**ANALYTE:**  
**HPRT1**

<b>NAME:</b>	hypoxanthine phosphoribosyltransferase 1
<b>SYMBOL:</b>	HPRT1
<b>VERSION OF ORPHANET:</b>	2023-06-22 14:14:43
<b>SYNONYMS:</b>	HGPRT Lesch-Nyhan syndrome
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">Ensembl</a> <a href="#">Genatlas</a> <a href="#">HGNC</a> <a href="#">OMIM</a> <a href="#">Reactome</a> <a href="#">SwissProt</a>
<b>CREATED:</b>	13 May 2019 - 01:01
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- [Ataxia \(autosomic dominant and recessive / except expansion of triplets\) \(gene panel - 722 genes\)](#)
- [Cerebral palsy \(gene panel\)](#)
- [Dystonia \(gene panel\)](#)
- [Inherited Kidney Diseases \(Gene Panel\)](#)
- [Intellectual Disability \(gene panel\)](#)
- [Intellectual disability & Epilepsy \(gene panel\)](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability \(virtual gene panel\)](#)
- [Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders \(213 genes\)](#)
- [Movement Disorders \(gene panel\)](#)
- [Movement Disorders \(gene panel\)](#)
- [Nephrocalcinosis and nephrolithiasis \(gene panel\)](#)
- [Nephrogenetics / Nephropathy \(gene panel\)](#)
- [Nephropathies, hereditary \(gene panel\)](#)
- [Neurodevelopmental disorders \(1300 genes\)](#)
- [Neurodevelopmental disorders gene panel](#)
- [Tubulopathy \(gene panel\)](#)

### Related Diseases

- [Hypoxanthine guanine phosphoribosyltransferase partial deficiency](#)
- [Lesch-Nyhan syndrome](#)

## Related Gene Panels

- [Ataxia \(348 genes\) - ULB](#)
- [Cerebral palsy \(212 genes\) - UZA](#)
- [Dystonia \(68 genes\) - KUL](#)
- [Intellectual Disability \(104 genes\) - IPG](#)
- [Intellectual disability & Epilepsy - UGent](#)
- [Intellectual disability \(gene panel\)](#)
- [Intellectual disability/Epilepsy \(1091 genes\) - ULG](#)
- [Metabolic disorders \(213 genes\) - VUB](#)
- [Movement Disorders - UGent](#)
- [Movement Disorders - ULG](#)
- [Nephrocalcinosis and nephrolithiasis \(37 genes\) - IPG](#)
- [Nephropathies, hereditary \(219 genes\) - KUL](#)
- [Nephropathy panel - UGent](#)
- [Neurodevelopmental disorders \(1300 genes\) - ULB](#)
- [Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders \(1162 genes\) - VUB](#)
- [Panel Nephro-ULG-V1](#)
- [Tubulopathy/Nephrolithiasis \(106 genes\) - IPG](#)

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