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Diagnostik & molekulare Diagnostik



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### Zuschläge

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# HPRT (h5): 293 Lysate: sc-158621

## BACKGROUND

HPRT (hypoxanthine phosphoribosyltransferase 1), also known as HGPRT or HPRT1, is a 218 amino acid cytoplasmic protein that belongs to the purine/pyrimidine phosphoribosyltransferase family. Involved in purine metabolism, HPRT functions as a purine salvage enzyme that catalyzes the conversion of hypoxanthine and guanine to their respective mononucleotides (inosine monophosphate and guanosine monophosphate, respectively). HPRT exists as a homotetramer that can bind two magnesium ions as cofactors. Defects in the gene encoding HPRT are the cause of gout and Lesch-Nyhan syndrome (LNS), both of which are characterized by a partial or complete lack of HPRT enzymatic activity. While a partial loss of HPRT enzymatic activity results in a buildup of uric acid (gout), a total loss of enzymatic activity results in hyperuricaemia, mental retardation, choreoathetosis and compulsive self-mutilation, all of which are symptoms associated with LNS. The severity of these diseases suggests an essential role for HPRT in purine metabolism.

## REFERENCES

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2. Fujimori, S., et al. 1997. An asymptomatic germline missense base substitution in the hypoxanthine phosphoribosyltransferase (HPRT) gene that reduces the amount of enzyme in humans. *Hum. Genet.* 99: 8-10.
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4. Koina, E., et al. 2005. An inactive X specific replication origin associated with a matrix attachment region in the human X linked HPRT gene. *J. Cell. Biochem.* 95: 391-402.
5. Dawson, P.A., et al. 2005. Normal HPRT coding region in a male with gout due to HPRT deficiency. *Mol. Genet. Metab.* 85: 78-80.
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## CHROMOSOMAL LOCATION

Genetic locus: HPRT1 (human) mapping to Xq26.1.

## PRODUCT

HPRT (h5): 293 Lysate represents a lysate of human HPRT transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

## APPLICATIONS

HPRT (h5): 293 Lysate is suitable as a Western Blotting positive control for human reactive HPRT antibodies. Recommended use: 10-20 µl per lane.

Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 cells.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.