

HPRT (m): 293T Lysate: sc-120891

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

1. Stout, J.T. and Caskey, C.T. 1985. HPRT: gene structure, expression, and mutation. *Annu. Rev. Genet.* 19: 127-148.
2. Fujimori, S., Sakuma, R., Yamaoka, N., Hakoda, M., Yamanaka, H. and Kamatani, N. 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.
3. Mizunuma, M., Fujimori, S., Ogino, H., Ueno, T., Inoue, H. and Kamatani, N. 2001. A recurrent large Alu-mediated deletion in the hypoxanthine phosphoribosyltransferase (HPRT1) gene associated with Lesch-Nyhan syndrome. *Hum. Mutat.* 18: 435-443.
4. Koina, E. and Piper, A. 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., Gordon, R.B., Keough, D.T. and Emmerson, B.T. 2005. Normal HPRT coding region in a male with gout due to HPRT deficiency. *Mol. Genet. Metab.* 85: 78-80.
6. Cossu, A., Orrù, S., Jacomelli, G., Carcassi, C., Contu, L., Sestini, S., Corradi, M.R., Pompucci, G., Carcassi, A. and Micheli, V. 2006. HPRTSardinia: a new point mutation causing HPRT deficiency without Lesch-Nyhan disease. *Biochim. Biophys. Acta* 1762: 29-33.
7. Keebaugh, A.C., Sullivan, R.T. and Thomas, J.W. 2007. Gene duplication and inactivation in the HPRT gene family. *Genomics* 89: 134-142.
8. Liu, S., Ao, L., Du, B., Zhou, Y., Yuan, J., Bai, Y., Zhou, Z. and Cao, J. 2008. HPRT mutations in lymphocytes from 1,3-butadiene-exposed workers in China. *Environ. Health Perspect.* 116: 203-208.

CHROMOSOMAL LOCATION

Genetic locus: *Hprt* (mouse) mapping to X A5.

PRODUCT

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

APPLICATIONS

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

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

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.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.