

MVK (h3): 293T Lysate: sc-170561

BACKGROUND

Mevalonate kinase (MVK) is an early enzyme in isoprenoid and sterol synthesis. Mevalonate kinase catalyzes the ATP-dependent phosphorylation of mevalonic acid to form mevalonate 5-phosphate. Mevalonate is a key intermediate, and mevalonate kinase a key early enzyme, in isoprenoid and sterol synthesis. Deficiency in MVK activity contributes to mevalonic aciduria and hyperimmunoglobulinemia D/periodic fever syndrome (HIDS). Mevalonic acid accumulates because of failure of conversion to 5-phosphomevalonic acid, which is catalyzed by mevalonate kinase. Mevalonic acid is synthesized from 3-hydroxy-3-methylglutaryl-CoA, a reaction catalyzed by HMG-CoA reductase.

REFERENCES

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2. Zheng, Q. 1995. On the MVK stochastic carcinogenesis model with Erlang distributed cell life lengths. *Risk Anal.* 15: 495-502.
3. Houten, S.M., et al. 1999. Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinemia D and periodic fever syndrome. *Nat. Genet.* 22: 175-177.
4. Houten, S.M., et al. 2001. Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinemia D and periodic fever syndrome. *Eur. J. Hum. Genet.* 9: 253-259.
5. Cuisset, L., et al. 2001. Molecular analysis of MVK mutations and enzymatic activity in hyper-IgD and periodic fever syndrome. *Eur. J. Hum. Genet.* 9: 260-266.
6. Fu, Z., et al. 2002. The structure of a binary complex between a mammalian mevalonate kinase and ATP: insights into the reaction mechanism and human inherited disease. *J. Biol. Chem.* 277: 18134-18142.
7. Houten, S.M., et al. 2003. Carrier frequency of the V377I (1129G>A) MVK mutation, associated with hyper-IgD and periodic fever syndrome, in the Netherlands. *Eur. J. Hum. Genet.* 11: 196-200.
8. Stojanov, S., et al. 2004. Molecular analysis of the MVK and TNFRSF1A genes in patients with a clinical presentation typical of the hyperimmunoglobulinemia D with periodic fever syndrome: a low-penetrance TNFRSF1A variant in a heterozygous MVK carrier possibly influences the phenotype of hyperimmunoglobulinemia D with periodic fever syndrome or vice versa. *Arthritis Rheum.* 50: 1951-1958.
9. LocusLink Report (LocusID: 4598). <http://www.ncbi.nlm.nih.gov/LocusLink/>

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.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: MVK (human) mapping to 12q24.11.

PRODUCT

MVK (h3): 293T Lysate represents a lysate of human MVK transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

MVK (h3): 293T Lysate is suitable as a Western Blotting positive control for human reactive MVK 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.

RESEARCH USE

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