

MAN2B1 (h2): 293T Lysate: sc-174574

BACKGROUND

MAN2B1 (mannosidase α class 2B member 1), also known as MANB (mannosidase α -B) or LAMAN (lysosomal α -mannosidase), is a 1,011 amino acid lysosomal protein that cleaves α -mannosidic linkages. Essential for N-linked carbohydrate catabolism during glycoprotein turnover, MAN2B1 binds one zinc ion per subunit and belongs to the glycosyl hydrolase 38 family. MAN2B1 is processed into five peptide chains and undergoes post-translational glycosylation. Defects in the gene encoding MAN2B1 are the cause of a lysosomal storage disease known as lysosomal α -mannosidosis (AM). Patients with AM have an accumulation of unbranched oligosaccharide chains primarily in the central nervous system, typically resulting in impaired hearing, mental retardation and frequent infections.

REFERENCES

1. Autio, S., Nordén, N.E., Ockerman, P.A., Riekkinen, P., Rapola, J. and Louhimo, T. 1973. Mannosidosis: clinical, fine-structural and biochemical findings in three cases. *Acta Paediatr. Scand.* 62: 555-565.
2. Bach, G., Kohn, G., Lasch, E.E., El Massri, M., Ornoy, A., Sekeles, E., Legum, C. and Cohen, M.M. 1978. A new variant of mannosidosis with increased residual enzymatic activity and mild clinical manifestation. *Pediatr. Res.* 12: 1010-1015.
3. Nebes, V.L. and Schmidt, M.C. 1994. Human lysosomal α -mannosidase: isolation and nucleotide sequence of the full-length cDNA. *Biochem. Biophys. Res. Commun.* 200: 239-245.
4. Beccari, T., Appolloni, M.G., Stirling, J.L. and Orlacchio, A. 1996. Assignment of lysosomal α -D-mannosidase to mouse chromosome 8. *Mamm. Genome* 7: 707-708.
5. Riise, H.M., Berg, T., Nilssen, O., Romeo, G., Tollersrud, O.K. and Ceccherini, I. 1997. Genomic structure of the human lysosomal α -mannosidase gene (MANB). *Genomics* 42: 200-207.
6. Berg, T., Riise, H.M., Hansen, G.M., Malm, D., Tranebjaerg, L., Tollersrud, O.K. and Nilssen, O. 1999. Spectrum of mutations in α -mannosidosis. *Am. J. Hum. Genet.* 64: 77-88.
7. Olmez, A., Nilssen, O., Coskun, T. and Klenow, H. 2003. α -mannosidosis and mutational analysis in a Turkish patient. *Turk. J. Pediatr.* 45: 46-50.
8. Sbaragli, M., Bibi, L., Pittis, M.G., Balducci, C., Heikinheimo, P., Ricci, R., Antuzzi, D., Parini, R., Spaccini, L., Bembì, B. and Beccari, T. 2005. Identification and characterization of five novel MAN2B1 mutations in Italian patients with α -mannosidosis. *Hum. Mutat.* 25: 320.
9. Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 609458. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

Genetic locus: MAN2B1 (human) mapping to 19p13.2.

PRODUCT

MAN2B1 (h2): 293T Lysate represents a lysate of human MAN2B1 transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

MAN2B1 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive MAN2B1 antibodies. Recommended use: 10-20 μ l 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.