Mpi (h): 293T Lysate: sc-115944



The Power to Question

BACKGROUND

Mpi (mannose phosphate isomerase), also known as PMI (phosphomannose isomerase) or PMI1, is a 423 amino acid zinc metalloenzyme belonging to the mannose-6-phosphate isomerase type 1 family, and is expressed in all tissues, more abundantly in heart, brain and skeletal muscle. A steady supply of D-mannose derivatives, which are required for most glycosylation reactions, is maintained by Mpi. Localized to the cytoplasm, Mpi utilizes zinc as a cofactor and catalyzes the interconversion of fructose-6-phosphate and mannose-6-phosphate. Mutations in the gene encoding Mpi lead to congenital disorder of glycosylation type 1B (CDG1B), also designated carbohydrate-deficient glycoprotein syndrome type lb (CDGS1B), which is characterized by protein-losing enteropathy. Congenital disorders of glycosylation are metabolic deficiencies in glycoprotein biosynthesis that usually results in severe mental and psychomotor retardation.

REFERENCES

- 1. Proudfoot, A.E., Goffin, L., Payton, M.A., Wells, T.N. and Bernard, A.R. 1996. *In vivo* and *in vitro* folding of a recombinant metalloenzyme, phosphomannose isomerase. Biochem. J. 318: 437-442.
- Jaeken, J., Matthijs, G., Saudubray, J.M., Dionisi-Vici, C., Bertini, E., de Lonlay, P., Henri, H., Carchon, H., Schollen, E. and Van Schaftingen, E. 1998. Phosphomannose isomerase deficiency: a carbohydrate-deficient glycoprotein syndrome with hepatic-intestinal presentation. Am. J. Hum. Genet. 62: 1535-1539.
- Niehues, R., Hasilik, M., Alton, G., Körner, C., Schiebe-Sukumar, M., Koch, H.G., Zimmer, K.P., Wu, R., Harms, E., Reiter, K., von Figura, K., Freeze, H.H., Harms, H.K. and Marquardt, T. 1998. Carbohydrate-deficient glycoprotein syndrome type lb. Phosphomannose isomerase deficiency and mannose therapy. J. Clin. Invest. 101: 1414-1420.
- Schollen, E., Dorland, L., de Koning, T.J., Van Diggelen, O.P., Huijmans, J.G., Marquardt, T., Babovic-Vuksanovic, D., Patterson, M., Imtiaz, F., Winchester, B., Adamowicz, M., Pronicka, E., Freeze, H. and Matthijs, G. 2000. Genomic organization of the human phosphomannose isomerase (MPI) gene and mutation analysis in patients with congenital disorders of glycosylation type lb (CDG-lb). Hum. Mutat. 16: 247-252.
- de Lonlay, P., Seta, N., Barrot, S., Chabrol, B., Drouin, V., Gabriel, B.M., Journel, H., Kretz, M., Laurent, J., Le Merrer, M., Leroy, A., Pedespan, D., Sarda, P., Villeneuve, N., Schmitz, J., van Schaftingen, E., Matthijs, G., Jaeken, J., Korner, C., Munnich, A., Saudubray, J.M. and Cormier-Daire, V. 2001. A broad spectrum of clinical presentations in congenital disorders of glycosylation I: a series of 26 cases. J. Med. Genet. 38: 14-19.
- Schollen, E., Martens, K., Geuzens, E. and Matthijs, G. 2002. DHPLC analysis as a platform for molecular diagnosis of congenital disorders of glycosylation (CDG). Eur. J. Hum. Genet. 10: 643-648.
- 7. Vuillaumier-Barrot, S., Le Bizec, C., de Lonlay, P., Barnier, A., Mitchell, G., Pelletier, V., Prevost, C., Saudubray, J.M., Durand, G. and Seta, N. 2002. Protein losing enteropathy-hepatic fibrosis syndrome in Saguenay-Lac St-Jean, Quebec is a congenital disorder of glycosylation type lb. J. Med. Genet. 39: 849-851.

CHROMOSOMAL LOCATION

Genetic locus: MPI (human) mapping to 15g24.1.

PRODUCT

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

APPLICATIONS

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

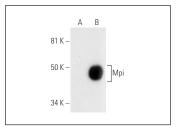
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

Mpi (E-4): sc-393477 is recommended as a positive control antibody for Western Blot analysis of enhanced human Mpi expression in Mpi transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

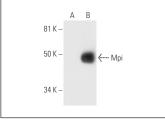
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-lgG κ BP-HRP: sc-516102 or m-lgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz MarkerTM Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA







Mpi (B-2): sc-393484. Western blot analysis of Mpi expression in non-transfected: sc-117752 (**A**) and human Mpi transfected: sc-115944 (**B**) 293T whole cell Ivsates

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.