SANTA CRUZ BIOTECHNOLOGY, INC.

POMGnT1 (JD.23): sc-130459

BACKGROUND

The product of the POMGNT1 gene, protein O-mannose β-1,2-N-acetylglucosaminyltransferase, participates in O-mannosyl glycan synthesis. POMGnT1 is an N(in)/C(out) (type II) membrane protein localized in the medial-Golgi that initiates the conversion of high mannose N-glycans to complex N-glycans. Specifically, POMGnT1 is a glycosylation enzyme that participates in the synthesis of O-mannosyl glycan, a laminin-binding ligand of α-dystroglycan that is rarely synthesized in mammals. Mutations in the POMGNT1 gene cause muscle-eye-brain disease (MEB), an autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities and lissencephaly. Altered glycosylation of α-dystroglycan may play a critical role in the pathomechanism of MEB as well as Walker-Warburg syndrome (WWS), characterized by the absence of glycosylation of α-dystroglycan. The human POMGNT1 gene maps to chromosome 1p34.1 and encodes a 660-amino acid type II transmembrane protein.

REFERENCES

1. Burke, J., et al. 1992. The transmembrane and flanking sequences of β-1, 2-N-acetylglucosaminyltransferase I specify medial-Golgi localization. J. Biol. Chem. 267: 24433-24440.
2. Yoshida, A., et al. 2001. Muscular dystrophy and neuronal migration disorder caused by mutations in a glycosyltransferase, POMGnT1. Dev. Cell 1: 717-724.
3. Kano, H., et al. 2002. Deficiency of α-dystroglycan in muscle-eye-brain disease. Biochem. Biophys. Res. Commun. 291: 1283-1286.
4. Michele, D.E., et al. 2002. Post-translational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies. Nature 418: 417-422.
5. Beltran-Valero De Bernabe, D., et al. 2002. Mutations in the O-mannosyltransferase gene POMT1 give rise to the severe neuronal migration disorder Walker-Warburg syndrome. Am. J. Hum. Genet. 71: 1033-1043.
6. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 606822. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

Genetic locus: POMGNT1 (human) mapping to 1p34.1; Pomgnt1 (mouse) mapping to 4 D1.

SOURCE

POMGnT1 (JD.23) is a mouse monoclonal antibody raised against recombinant POMGnT1 of human origin.

PRODUCT

Each vial contains 100 µg IgG_{2a} kappa light chain in 1.0 ml PBS with < 0.1% sodium azide and 0.1% gelatin.

RESEARCH USE

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

APPLICATIONS

POMGnT1 (JD.23) is recommended for detection of POMGnT1 of mouse, rat and human origin by immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500).

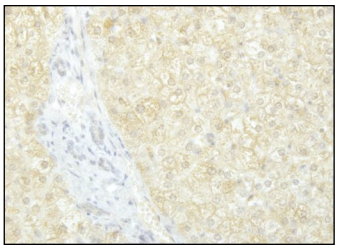
Suitable for use as control antibody for POMGnT1 siRNA (h): sc-40865, POMGnT1 siRNA (m): sc-40866, POMGnT1 shRNA Plasmid (h): sc-40865-SH, POMGnT1 shRNA Plasmid (m): sc-40866-SH, POMGnT1 shRNA (h) Lentiviral Particles: sc-40865-V and POMGnT1 shRNA (m) Lentiviral Particles: sc-40866-V.

Molecular Weight of POMGnT1: 75 kDa.

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



POMGnT1 (JD.23): sc-130459. Immunoperoxidase staining of formalin-fixed, paraffin-embedded human liver tissue showing cytoplasmic localization.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

PROTOCOLS

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