SANTA CRUZ BIOTECHNOLOGY, INC.

POMT1 (K-15): sc-48914



BACKGROUND

O-mannosylation is an essential protein modification in eukaryotes that is initiated by an evolutionarily conserved family of protein O-mannosyltransferases. The POMT1 (protein O-mannosyltransferase 1) protein consists of 725 amino acids. POMT1 contains seven to twelve presumed transmembrane regions and a C-terminal ER membrane retention signal; RT-PCR reveals several mRNA splice variants. RNA dot blot analysis indicates ubiquitous expression of POMT1, with maximum levels in testis and high levels in fetal brain and pituitary tissues. Walker-Warburg syndrome (WWS), a severe, recessive, congenital muscular dystrophy associated with defects in neuronal migration that produce complex brain and eye abnormalities, is caused by mutations in the POMT1 gene.

REFERENCES

- Akasaka-Manya, K., et al. 2004. Mutations of the POMT1 gene found in patients with Walker-Warburg syndrome lead to a defect of protein Omannosylation. Biochem. Biophys. Res. Commun. 325: 75-79.
- Ichimiya, T., et al. 2004. The twisted abdomen phenotype of *Drosophila* POMT1 and POMT2 mutants coincides with their heterophilic protein Omannosyltransferase activity. J. Biol. Chem. 279: 42638-42647.
- 3. Willer, T., et al. 2004. Targeted disruption of the Walker-Warburg syndrome gene POMT1 in mouse results in embryonic lethality. Proc. Natl. Acad. Sci. USA 101: 14126-14131.
- Yamamoto, T., et al. 2004. Expression and localization of fukutin, POMGnT1, and POMT1 in the central nervous system: consideration for functions of fukutin. Med. Electron Microsc. 37: 200-207.
- Balci, B., et al. 2005. An autosomal recessive limb girdle muscular dystrophy (LGMD2) with mild mental retardation is allelic to Walker-Warburg syndrome (WWS) caused by a mutation in the POMT1 gene. Neuromuscul. Disord. 15: 271-275.
- Currier, S.C., et al. 2005. Mutations in POMT1 are found in a minority of patients with Walker-Warburg syndrome. Am. J. Med. Genet. A 133: 53-57.
- 7. D'Amico, A., et al. 2006. Expanding the clinical spectrum of POMT1 phenotype. Neurology 66: 1564-1567.
- 8. Manya, H., et al. 2006. Molecular cloning and characterization of rat POMT1 and POMT2. Glycobiology 16: 863-873.
- 9. van Reeuwijk, J., et al. 2006. The expanding phenotype of POMT1 mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly and mental retardation. Hum. Mutat. 27: 453-459.

CHROMOSOMAL LOCATION

Genetic locus: POMT1 (human) mapping to 9q34.13; Pomt1 (mouse) mapping to 2 B.

SOURCE

POMT1 (K-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of POMT1 of human origin.

PRODUCT

Each vial contains 200 μg lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-48914 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

POMT1 (K-15) is recommended for detection of POMT1 isoforms 1, 2 and 3 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

POMT1 (K-15) is also recommended for detection of POMT1 isoforms 1, 2 and 3 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for POMT1 siRNA (h): sc-61379, POMT1 siRNA (m): sc-61380, POMT1 shRNA Plasmid (h): sc-61379-SH, POMT1 shRNA Plasmid (m): sc-61380-SH, POMT1 shRNA (h) Lentiviral Particles: sc-61379-V and POMT1 shRNA (m) Lentiviral Particles: sc-61380-V.

Molecular Weight of POMT1: 75 kDa.

Positive Controls: Mouse testis extract: sc-2405 or F9 cell lysate: sc-2245.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluo-rescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. 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 or our catalog for detailed protocols and support products.