SANTA CRUZ BIOTECHNOLOGY, INC.

CLN1 (1117CT11.2.1.4): sc-517323

BACKGROUND

CLN1 (ceroid lipofuscinosis 1), also known as PPT, INCL or PPT1 (palmitoyl-protein thioesterase 1), is a 306 amino acid glycosylated protein that localizes to lysosome and is a member of the palmitoyl-protein thioesterase family. CLN1 functions to remove thioester-linked fatty acyl groups from a variety of substrates, such as as palmitate, from modified cysteine residues in proteins or peptides during lysosomal degradation. Defects in the gene encoding CLN1 are a cause of infantile neuronal ceroid lipofuscinosis 1 (CLN1 or INCL) and neuronal ceroid lipofuscinosis 4 (CLN4). Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, with clinical symptoms including seizures, dementia, visual loss and/or cerebral atrophy.

REFERENCES

1. Vesa, J., et al. 1995. Mutations in the palmitoyl protein thioesterase gene causing infantile neuronal ceroid lipofuscinosis. Nature 376: 584-587.
2. Schriner, J.E., et al. 1996. cDNA and genomic cloning of human palmitoyl-protein thioesterase (PPT), the enzyme defective in infantile neuronal ceroid lipofuscinosis. Genomics 34: 317-322.
3. Crews, C.M., et al. 1996. Didemnin binds to the protein palmitoyl thioesterase responsible for infantile neuronal ceroid lipofuscinosis. Proc. Natl. Acad. Sci. USA 93: 4316-4319.
4. van Diggelen, O.P., et al. 2001. Adult neuronal ceroid lipofuscinosis with palmitoyl-protein thioesterase deficiency: first adult-onset patients of a childhood disease. Ann. Neurol. 50: 269-272.
5. Gupta, P., et al. 2001. Disruption of PPT1 or PPT2 causes neuronal ceroid lipofuscinosis in knockout mice. Proc. Natl. Acad. Sci. USA 98: 13566-13571.
6. Taschner, P.E., et al. 2005. From gene to disease; from CLN1, CLN2 and CLN3 to neuronal ceroid lipofuscinosis. Ned. Tijdschr. Geneeskd. 149: 300-303.
7. Tsukamoto, T., et al. 2006. Overexpression in colorectal carcinoma of two lysosomal enzymes, CLN2 and CLN1, involved in neuronal ceroid lipofuscinosis. Cancer 106: 1489-1497.
8. Ohno, K., et al. 2009. Structural basis of neuronal ceroid lipofuscinosis 1. Brain Dev. 32: 524-530.
9. Kohan, R., et al. 2009. An integrated strategy for the diagnosis of neuronal ceroid lipofuscinosis types 1 (CLN1) and 2 (CLN2) in eleven Latin American patients. Clin. Genet. 76: 372-382.

CHROMOSOMAL LOCATION

Genetic locus: PPT1 (human) mapping to 1p34.2.

SOURCE

CLN1 (1117CT11.2.1.4) is a mouse monoclonal antibody raised against a recombinant protein representing full-length CLN1 of human origin.

STORAGE

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

PRODUCT

Each vial contains 100 µg IgG_1 in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

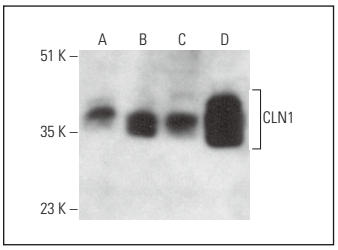
CLN1 (1117CT11.2.1.4) is recommended for detection of CLN1 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and flow cytometry (1 µg per 1 x 10^6 cells).

Suitable for use as control antibody for CLN1 siRNA (h): sc-105216, CLN1 shRNA Plasmid (h): sc-105216-SH and CLN1 shRNA (h) Lentiviral Particles: sc-105216-V.

Molecular Weight of glycoslylated CLN1 doublet: 37 kDa.

Positive Controls: HeLa whole cell lysate: sc-2200, Hep G2 cell lysate: sc-2227 or HEK293 whole cell lysate: sc-45136.

DATA



CLN1 (1117CT11.2.1.4): sc-517323. Western blot analysis of CLN1 expression in HeLa (**A**), Hep G2 (**B**), HEK293 (**C**) and THP-1 (**D**) whole cell lysates.

SELECT PRODUCT CITATIONS

1. Davis, O.B., et al. 2021. NPC1-mTORC1 signaling couples cholesterol sensing to organelle homeostasis and is a targetable pathway in Niemann-Pick type C. Dev. Cell 56: 260-276.e7.

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.