

PTCHD1 (Y-18): sc-248324

BACKGROUND

PTCHD1 (patched domain-containing protein 1) is an 888 amino acid multi-pass membrane protein. Belonging to the patched family, PTCHD1 contains one SSD (sterol-sensing) domain and is widely expressed, with high levels found in the gray and white cerebellum, spinal cord, stomach, uterus, prostate and lung. PTCHD1 may play a role in the hedgehog signaling pathway by inhibiting GLI promoter transcription. The gene encoding PTCHD1 maps to human chromosome Xp22.11 and mouse chromosome X F3. Deletions of the PTCHD1 gene have been linked to intellectual disability, autism and mental retardation. PTCHD1 exists as three isoforms due to alternative splicing events.

REFERENCES

1. Marshall, C.R., et al. 2008. Structural variation of chromosomes in autism spectrum disorder. *Am. J. Hum. Genet.* 82: 477-488.
2. Whibley, A.C., et al. 2010. Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability. *Am. J. Hum. Genet.* 87: 173-188.
3. Pinto, D., et al. 2010. Functional impact of global rare copy number variation in autism spectrum disorders. *Nature* 466: 368-372.
4. Noor, A., et al. 2010. Disruption at the PTCHD1 Locus on Xp22.11 in Autism spectrum disorder and intellectual disability. *Sci. Transl. Med.* 2: 49ra68.
5. Filges, I., et al. 2011. Deletion in Xp22.11: PTCHD1 is a candidate gene for X-linked intellectual disability with or without autism. *Clin. Genet.* 79: 79-85.

CHROMOSOMAL LOCATION

Genetic locus: PTCHD1 (human) mapping to Xp22.11; Ptchd1 (mouse) mapping to X F3.

SOURCE

PTCHD1 (Y-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of PTCHD1 of human origin.

PRODUCT

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

Blocking peptide available for competition studies, sc-248324 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

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 or our catalog for detailed protocols and support products.

APPLICATIONS

PTCHD1 (Y-18) is recommended for detection of PTCHD1 of mouse, rat and 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) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with PTCHD2 or PTCHD3.

PTCHD1 (Y-18) is also recommended for detection of PTCHD1 in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for PTCHD1 siRNA (h): sc-91304, PTCHD1 siRNA (m): sc-152574, PTCHD1 shRNA Plasmid (h): sc-91304-SH, PTCHD1 shRNA Plasmid (m): sc-152574-SH, PTCHD1 shRNA (h) Lentiviral Particles: sc-91304-V and PTCHD1 shRNA (m) Lentiviral Particles: sc-152574-V.

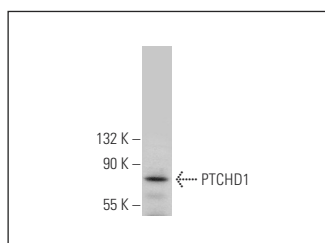
Molecular Weight of PTCHD1 isoforms 1/2/3: 101/89/29 kDa.

Positive Controls: K-562 whole cell lysate: sc-2203.

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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: 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.

DATA



PTCHD1 (Y-18): sc-248324. Western blot analysis of PTCHD1 expression in K-562 whole cell lysate.

RESEARCH USE

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