

COH1 (Y-18): sc-51480

BACKGROUND

COH1 (cohen syndrome protein 1), also known as VPS13B (vacuolar protein sorting-associated protein 13B) or CHS1, is a 4,022 amino acid protein that belongs to the VPS13 family. COH1 is widely expressed and may be involved in protein sorting in post Golgi membrane traffic. Defects in the gene that encodes COH1 are the cause of Cohen syndrome, which is characterized by intellectual deficit, abnormalities of the hands and feet, hypotonia and obesity. Craniofacial dysmorphism commonly seen with Cohen syndrome include a low hairline, thick hair, high-arched or wave-shaped eyelids and a short philtrum. COH1 is expressed as five isoforms produced by alternative splicing. Isoform 1 is expressed in brain and retina while isoform 2 is expressed ubiquitously.

REFERENCES

1. Kolehmainen, J., et al. 2003. Cohen syndrome is caused by mutations in a novel gene, COH1, encoding a transmembrane protein with a presumed role in vesicle-mediated sorting and intracellular protein transport. *Am. J. Hum. Genet.* 72: 1359-1369.
2. Hennies, H.C., et al. 2004. Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. *Am. J. Hum. Genet.* 75: 138-145.
3. Velayos-Baeza, A., et al. 2004. Analysis of the human VPS13 gene family. *Genomics* 84: 536-549.
4. Farooqi, I.S. 2005. Genetic and hereditary aspects of childhood obesity. *Best Pract. Res. Clin. Endocrinol. Metab.* 19: 359-374.
5. Kondo, I., et al. 2005. COH1 analysis and linkage study in two Japanese families with Cohen syndrome. *Clin. Genet.* 67: 270-272.
6. Zarzour, W., et al. 2005. Two novel CHS1 (LYST) mutations: clinical correlations in an infant with Chediak-Higashi syndrome. *Mol. Genet. Metab.* 85: 125-132.
7. Khan, A., et al. 2006. Corneal ectasia associated with Cohen syndrome: a role for COH1 in corneal development and maintenance? *Br. J. Ophthalmol.* 90: 390-391.

CHROMOSOMAL LOCATION

Genetic locus: VPS13B (human) mapping to 8q22.2; Vps13b (mouse) mapping to 15 B3.1.

SOURCE

COH1 (Y-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of COH1 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.

PROTOCOLS

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

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-51480 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

COH1 (Y-18) is recommended for detection of COH1 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).

COH1 (Y-18) is also recommended for detection of COH1 in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for COH1 siRNA (h): sc-105225, COH1 siRNA (m): sc-142457, COH1 shRNA Plasmid (h): sc-105225-SH, COH1 shRNA Plasmid (m): sc-142457-SH, COH1 shRNA (h) Lentiviral Particles: sc-105225-V and COH1 shRNA (m) Lentiviral Particles: sc-142457-V.

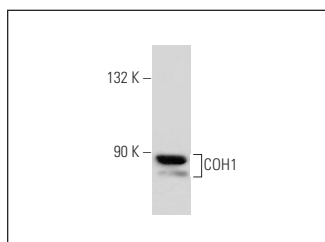
Molecular Weight of COH1 isoforms: 449/445/160/98/47 kDa.

Positive Controls: Jurkat whole cell lysate: sc-2204.

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



COH1 (Y-18): sc-51480. Western blot analysis of COH1 expression in Jurkat whole cell lysate.

RESEARCH USE

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