

COH1 Antibody

Catalog # ASC11509

Product Information

| Application | WB, IF, E |
|-----------------------|--|
| Primary Accession | <u>Q7Z7G8</u> |
| Other Accession | <u>NP_056058</u> , <u>35493725</u> |
| Reactivity | Human, Mouse, Rat |
| Host | Rabbit |
| Clonality | Polyclonal |
| Isotype | IgG |
| Calculated MW | 448664 |
| Concentration (mg/ml) | 1 mg/mL |
| Conjugate | Unconjugated |
| Application Notes | COH1 antibody can be used for detection of COH1 by Western blot at 1 - 2 ᠋g/mL. For immunofluorescence start at 20 ᠋g/mL. |

Additional Information

| Gene ID Other Names | 157680 Vacuolar protein sorting-associated protein 13B, Cohen syndrome protein 1, VPS13B, CHS1, COH1, KIAA0532 |
|--------------------------|---|
| Target/Specificity | VPS13B; At least five alternatively spliced transcript variants have been observed. COH1 detects two isoforms. |
| Reconstitution & Storage | COH1 antibody can be stored at 4°C for three months and -20°C, stable for up to one year. As with all antibodies care should be taken to avoid repeated freeze thaw cycles. Antibodies should not be exposed to prolonged high temperatures. |
| Precautions | COH1 Antibody is for research use only and not for use in diagnostic or therapeutic procedures. |

Protein Information

| Name | VPS13B |
|----------|--|
| Synonyms | CHS1, COH1, KIAA0532 |
| Function | Mediates the transfer of lipids between membranes at organelle contact sites (By similarity). Binds phosphatidylinositol 3- phosphate (By similarity). Functions as a tethering factor in the slow endocytic recycling pathway, to assist traffic between early and recycling endosomes (PubMed: <u>24334764</u> , PubMed: <u>30962439</u> , PubMed: <u>32375900</u>). Involved in the transport of proacrosomal vesicles to the nuclear dense lamina (NDL) during spermatid development (By similarity). Plays a role in the assembly of the Golgi |

| | apparatus, possibly by mediating trafficking to the Golgi membrane (PubMed: <u>21865173</u>). Plays a role in the development of the nervous system, and may be required for neuron projection development (PubMed: <u>25492866</u> , PubMed: <u>32560273</u>). May also play a role during adipose tissue development (PubMed: <u>26358774</u>). Required for maintenance of the ocular lens (By similarity). |
|-------------------|--|
| Cellular Location | Recycling endosome membrane {ECO:0000250 UniProtKB:Q80TY5}; Peripheral membrane protein. Cytoplasmic vesicle, secretory vesicle, acrosome membrane {ECO:0000250 UniProtKB:Q80TY5}; Peripheral membrane protein. Golgi apparatus, cis-Golgi network membrane; Peripheral membrane protein. Endoplasmic reticulum- Golgi intermediate compartment membrane; Peripheral membrane protein. Golgi apparatus, trans-Golgi network membrane; Peripheral membrane protein. Early endosome membrane; Peripheral membrane protein. Lysosome membrane; Peripheral membrane protein. Note=Localizes to proacrosomal and acrosomal vesicles and not the Golgi apparatus during acrosome formation. {ECO:0000250 UniProtKB:Q80TY5} |
| Tissue Location | Widely expressed (PubMed:12730828). There is apparent differential expression of different transcripts (PubMed:12730828, PubMed:19006247). In fetal brain, lung, liver, and kidney, two transcripts of 2 and 5 kb are identified (PubMed:12730828) These transcripts are also seen in all adult tissues analyzed (PubMed:12730828). A larger transcript (12-14 kb) is expressed in prostate, testis, ovary, and colon in the adult (PubMed:12730828) Expression is very low in adult brain tissue (PubMed:12730828) Expressed in peripheral blood lymphocytes (PubMed:33025479). Isoform 1 and isoform 2 are expressed in brain and retina (PubMed:12730828, PubMed:19006247). Isoform 2 is expressed ubiquitously (PubMed:12730828, PubMed:19006247). |

Background

COH1 Antibody: COH1 (Cohen syndrome protein 1), also known as VPS13B (vacuolar protein sorting-associated protein 13B) or CHS1, belongs to the VPS13 family and may function in vesicle-mediated transport and sorting of proteins within the cell. COH1 is widely expressed and multiple alternatively spliced transcript variants have been observed. Mutations in this gene have been associated with Cohen syndrome. COH1 is a Golgi-localized peripheral membrane protein and plays a critical role in Golgi (re)assembly.

References

Velayos-Baeza A, Vettori A, Copley RR, et al. Analysis of the human VPS13 gene family. Genomics 2004; 84:536-49.

Kolehmainen J, Black GC, Saarinen A, et al. 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. 2003; 72:1359-69.

Seifert W, Kühnisch J, Maritzen T, et al. Cohen syndrome-associated protein, COH1, is a novel, giant Golgi matrix protein required for Golgi integrity. J. Biol. Chem. 2011; 286:37665-75.

Images

Western blot analysis of COH1 in SK-N-SH cell lysate with COH1 antibody at (A) 1 and (B) 2 $\mu g/mL$.





Immunofluorescence of COH1 in human brain tissue with COH1 antibody at 20 $\mu\text{g/mL}.$

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