

# B9D1 Antibody

Catalog # ASC11440

## Product Information

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| <b>Application</b>           | WB, IF, ICC, E                                                                                                                                                                                              |
| <b>Primary Accession</b>     | <a href="#">Q9UPM9</a>                                                                                                                                                                                      |
| <b>Other Accession</b>       | <a href="#">NP_056496</a> , <a href="#">343478275</a>                                                                                                                                                       |
| <b>Reactivity</b>            | Human                                                                                                                                                                                                       |
| <b>Host</b>                  | Rabbit                                                                                                                                                                                                      |
| <b>Clonality</b>             | Polyclonal                                                                                                                                                                                                  |
| <b>Isotype</b>               | IgG                                                                                                                                                                                                         |
| <b>Calculated MW</b>         | 22775                                                                                                                                                                                                       |
| <b>Concentration (mg/ml)</b> | 1 mg/mL                                                                                                                                                                                                     |
| <b>Conjugate</b>             | Unconjugated                                                                                                                                                                                                |
| <b>Application Notes</b>     | B9D1 antibody can be used for detection of B9D1 by Western blot at 1 $\mu$ g/mL. Antibody can also be used for immunocytochemistry starting at 5 $\mu$ g/mL. For immunofluorescence start at 20 $\mu$ g/mL. |

## Additional Information

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| <b>Gene ID</b>                      | 27077                                                                                                                                                                                                                                        |
| <b>Other Names</b>                  | B9 domain-containing protein 1, MKS1-related protein 1, B9D1, MKSR1                                                                                                                                                                          |
| <b>Target/Specificity</b>           | B9D1; At least two isoforms of B9D1 are known to exist; this antibody will only recognize the longest isoform. B9D1 antibody is predicted to not cross-react with other DNAJC family members.                                                |
| <b>Reconstitution &amp; Storage</b> | B9D1 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. |
| <b>Precautions</b>                  | B9D1 Antibody is for research use only and not for use in diagnostic or therapeutic procedures.                                                                                                                                              |

## Protein Information

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| <b>Name</b>              | B9D1                                                                                                                                                                                                                                                                                              |
| <b>Synonyms</b>          | MKSR1                                                                                                                                                                                                                                                                                             |
| <b>Function</b>          | Component of the tectonic-like complex, a complex localized at the transition zone of primary cilia and acting as a barrier that prevents diffusion of transmembrane proteins between the cilia and plasma membranes. Required for ciliogenesis and sonic hedgehog/SHH signaling (By similarity). |
| <b>Cellular Location</b> | Cytoplasm, cytoskeleton, cilium basal body. Cytoplasm, cytoskeleton, cilium                                                                                                                                                                                                                       |

axoneme. Note=Localizes at the transition zone, a region between the basal body and the ciliary axoneme.

## Background

**B9D1 Antibody:** Meckel syndrome (MKS) is an embryonic lethal, autosomal recessive disorder characterized by polycystic kidney disease, central nervous system defects, polydactyly and liver fibrosis. B9D1 is a B9 domain-containing protein, one of several that are involved in ciliogenesis. Alterations in expression of this gene have been found in a family with Meckel syndrome. B9D1, and its related protein B9D2, form a complex with MKS1, disruption of which causes MKS. B9D1 is thought to be required for normal hedgehog signaling, ciliogenesis, and ciliary protein localization.

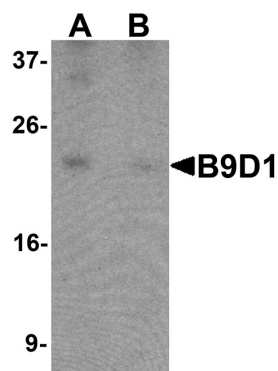
## References

Williams CL, Winkelbauer ME, Schafer JC, et al. Functional redundancy of the B9 proteins and nephrocystins in *Caenorhabditis elegans* ciliogenesis. *Mol. Biol. Cell* 2008; 19:2154-68.

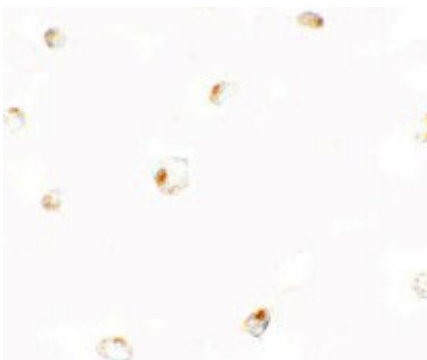
Hopp K, Heyer CM, Hommerding CJ, et al. B9D1 is revealed as a novel Meckel syndrome (MKS) gene by targeted exon-enriched next-generation sequencing and deletion analysis. *Hum. Mol. Genet.* 2011; 20:2524-34.

Dowdle WE, Robinson JF, Kneist A, et al. Disruption of a ciliary B9 protein complex causes Meckel syndrome. *Am. J. Hum. Genet.* 2011; 89:94-110.

## Images

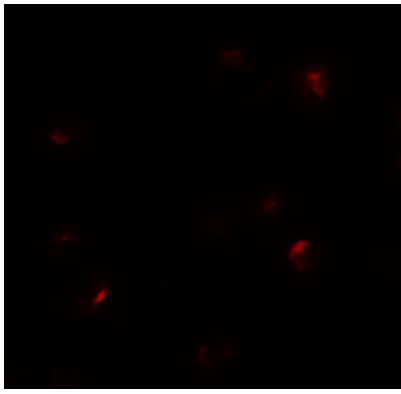


Western blot analysis of B9D1 in 293 cell lysate with B9D1 antibody at 1  $\mu\text{g/mL}$  in (A) the absence and (B) the presence of blocking peptide.



Immunocytochemistry of B9D1 in 293 cells with B9D1 antibody at 5  $\mu\text{g/mL}$ .

Immunofluorescence of B9D1 in 293 cells with B9D1 antibody at 20  $\mu\text{g/mL}$ .



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