

OFD1 (N-12): sc-168837

BACKGROUND

OFD1 (oral-facial-digital syndrome 1), also known as 71-7A, SGBS2, CXorf5, or JBTS10, is a 1,012 amino acid cytoplasmic and nuclear protein that is widely expressed and is a member of the OFD1 family. Containing one LisH domain, OFD1 exists as a homooligomer that can undergo alternative splicing to produce three alternatively spliced isoforms. OFD1 participates in controlling centriole length and is involved in the biogenesis of cilium, a centriole-associated structure that is found in many vertebrate cells and are required to transduce signals important for development and tissue homeostasis. OFD1 is thought to play a critical role in development by regulating Wnt signaling. Mutations in the gene encoding OFD1 are the cause of several diseases, including oral-facial-digital syndrome type 1 (OFD1), Simpson-Golabi-Behmel syndrome type 2 and Joubert syndrome type 10 (JBTS10).

REFERENCES

1. de Conciliis, L., et al. 1998. Characterization of Cxorf5 (71-7A), a novel human cDNA mapping to Xp22 and encoding a protein containing coiled-coil α -helical domains. *Genomics* 51: 243-250.
2. Ferrante, M.I., et al. 2001. Identification of the gene for oral-facial-digital type I syndrome. *Am. J. Hum. Genet.* 68: 569-576.
3. Rakkolainen, A., et al. 2002. Four novel mutations in the OFD1 (Cxorf5) gene in Finnish patients with oral-facial-digital syndrome 1. *J. Med. Genet.* 39: 292-296.
4. Romio, L., et al. 2003. OFD1, the gene mutated in oral-facial-digital syndrome type 1, is expressed in the metanephros and in human embryonic renal mesenchymal cells. *J. Am. Soc. Nephrol.* 14: 680-689.
5. Andersen, J.S., et al. 2003. Proteomic characterization of the human centrosome by protein correlation profiling. *Nature* 426: 570-574.
6. Budny, B., et al. 2006. A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oral-facial-digital type I syndrome. *Hum. Genet.* 120: 171-178.

CHROMOSOMAL LOCATION

Genetic locus: OFD1 (human) mapping to Xp22.2; Ofd1 (mouse) mapping to X F5.

SOURCE

OFD1 (N-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of OFD1 of human origin.

PRODUCT

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

Blocking peptide available for competition studies, sc-168837 P, (100 μ g 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.

APPLICATIONS

OFD1 (N-12) is recommended for detection of OFD1 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g 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).

OFD1 (N-12) is also recommended for detection of OFD1 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for OFD1 siRNA (h): sc-91245, OFD1 siRNA (m): sc-150182, OFD1 shRNA Plasmid (h): sc-91245-SH, OFD1 shRNA Plasmid (m): sc-150182-SH, OFD1 shRNA (h) Lentiviral Particles: sc-91245-V and OFD1 shRNA (m) Lentiviral Particles: sc-150182-V.

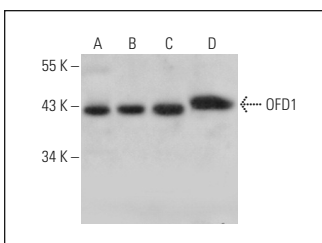
Molecular Weight of OFD1: 116 kDa.

Positive Control: U266 whole cell lysate: sc-364800, SH-SY5Y cell lysate: sc-3812 or NIH/3T3 nuclear extract: sc-2138.

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



OFD1 (N-12): sc-168837. Western blot analysis of OFD1 expression in U266 (A) and SH-SY5Y (B) whole cell lysates and SH-SY5Y (C) and NIH/3T3 (D) nuclear extracts.

RESEARCH USE

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

PROTOCOLS

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