

OFD1 (Q-15): sc-168838

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

- 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.
- Ferrante, M.I., et al. 2001. Identification of the gene for oral-facial-digital type I syndrome. *Am. J. Hum. Genet.* 68: 569-576.
- 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.
- 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.
- Andersen, J.S., et al. 2003. Proteomic characterization of the human centrosome by protein correlation profiling. *Nature* 426: 570-574.
- 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.
- Giorgio, G., et al. 2007. Functional characterization of the OFD1 protein reveals a nuclear localization and physical interaction with subunits of a chromatin remodeling complex. *Mol. Biol. Cell* 18: 4397-4404.

CHROMOSOMAL LOCATION

Genetic locus: OFD1 (human) mapping to Xp22.2; *Odf1* (mouse) mapping to X F5.

SOURCE

OFD1 (Q-15) 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-168838 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

OFD1 (Q-15) 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 (Q-15) is also recommended for detection of OFD1 in additional species, including equine, canine, bovine, porcine and avian.

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.

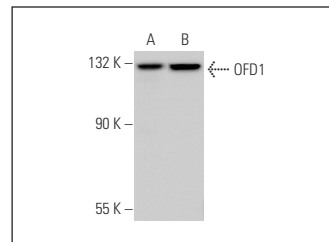
Molecular Weight of OFD1: 116 kDa.

Positive Controls: HeLa whole cell lysate: sc-2200 or A549 cell lysate: sc-2413.

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 (Q-15): sc-168838. Western blot analysis of OFD1 expression in A549 (A) and HeLa (B) whole cell lysates.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

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