SANTA CRUZ BIOTECHNOLOGY, INC.

PHF6 (S-12): sc-131747

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The Power to Question

BACKGROUND

Zinc finger proteins contain DNA-binding domains and have a wide variety of functions, most of which encompass some form of transcriptional activation or repression. PHF6 (PHD finger protein 6), also known as BORJ, is a 365 amino acid protein that localizes to the nucleus and contains 2 PHD-type zinc fingers. Expressed ubiquitously, PHF6 exists as two alternatively spliced isoforms and is thought to play a role in transcriptional regulation. Upon DNA damage, PHF6 is subject to phosphorylation, probably by ATM or ATR. Mutations in the gene encoding PHF6 are the cause of Boerjeson-Forssman-Lehmann syndrome (BFLS), an X-linked recessive disorder that is characterized by mental retardation, epilepsy, hypogonadism, hypometabolism, obesity with marked gynecomastia, swelling of subcutaneous tissue of the face and narrow palpebral fissure.

REFERENCES

1. Borjeson, M., et al. 1962. An X-linked, recessively inherited syndrome characterized by grave mental deficiency, epilepsy, and endocrine disorder. Acta Med. Scand. 171: 13-21.
2. Lower, K.M., et al. 2002. Muta-tions in PHF6 are associated with Börjeson-Forssman-Lehmann syndrome. Nat. Genet. 32: 661-665.
3. Lower, K.M., et al. 2004. 1024C> T (R342X) is a recurrent PHF6 mutation also found in the original Börjeson-Forssman-Lehmann syndrome family. Eur. J. Hum. Genet. 12: 787-789.
4. Vallee, D., et al. 2004. A novel PHF6 mutation results in enhanced exon skipping and mild Börjeson-Forssman-Lehmann syndrome. J. Med. Genet. 41: 778-783.
5. Crawford, J., et al. 2006. Mutation screening in Borjeson-Forssman-Lehmann syndrome: identification of a novel *de novo* PHF6 mutation in a female patient. J. Med. Genet. 43: 238-243.
6. Voss, A.K., et al. 2007. Protein and gene expression analysis of PHF6, the gene mutated in the Börjeson-Forssman-Lehmann Syndrome of intellectual disability and obesity. Gene Expr. Patterns 7: 858-871.

CHROMOSOMAL LOCATION

Genetic locus: PHF6 (human) mapping to Xq26.2; Phf6 (mouse) mapping to X A5.

SOURCE

PHF6 (S-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of PHF6 of human origin.

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-131747 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

RESEARCH USE

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

APPLICATIONS

PHF6 (S-12) is recommended for detection of PHF6 isoforms 1 and 2 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); non cross-reactive with other PHF family members.

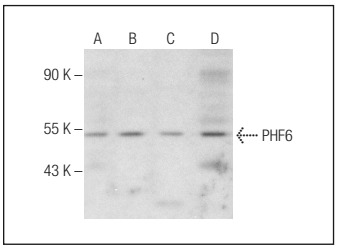
PHF6 (S-12) is also recommended for detection of PHF6 isoforms 1 and 2 in additional species, including canine, bovine and porcine.

Suitable for use as control antibody for PHF6 siRNA (h): sc-90882, PHF6 siRNA (m): sc-152219, PHF6 shRNA Plasmid (h): sc-90882-SH, PHF6 shRNA Plasmid (m): sc-152219-SH, PHF6 shRNA (h) Lentiviral Particles: sc-90882-V and PHF6 shRNA (m) Lentiviral Particles: sc-152219-V.

Molecular Weight of PHF6 isoforms: 41/35 kDa.

Positive Controls: K-562 nuclear extract: sc-2130, Hep G2 cell lysate: sc-2227 or HeLa nuclear extract: sc-2120.

DATA



PHF6 (S-12): sc-131747. Western blot analysis of PHF6 expression in K-562 (**A**), Hep G2 (**B**), HeLa (**C**) and Jurkat (**D**) nuclear extracts.

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.