

PHF6 (B-9): sc-365013

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 two 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 Börjeson-Forsssman-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. Mutations in PHF6 are associated with Börjeson-Forsssman-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-Forsssman-Lehmann syndrome family. *Eur. J. Hum. Genet.* 12: 787-789.
4. Vallée, D., et al. 2004. A novel PHF6 mutation results in enhanced exon skipping and mild Börjeson-Forsssman-Lehmann syndrome. *J. Med. Genet.* 41: 778-783.
5. Crawford, J., et al. 2006. Mutation screening in Börjeson-Forsssman-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-Forsssman-Lehmann syndrome of intellectual disability and obesity. *Gene Expr. Patterns* 7: 858-871.
7. Carter, M.T., et al. 2009. Further clinical delineation of the Börjeson-Forsssman-Lehmann syndrome in patients with PHF6 mutations. *Am. J. Med. Genet. A* 149A: 246-250.
8. Mangelsdorf, M., et al. 2009. Börjeson-Forsssman-Lehmann syndrome due to a novel plant homeodomain zinc finger mutation in the PHF6 gene. *J. Child Neurol.* 24: 610-614.
9. Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 300414. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

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

SOURCE

PHF6 (B-9) is a mouse monoclonal antibody raised against amino acids 1-300 mapping at the N-terminus of PHF6 of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2b} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

PHF6 (B-9) is recommended for detection of PHF6 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, 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).

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.

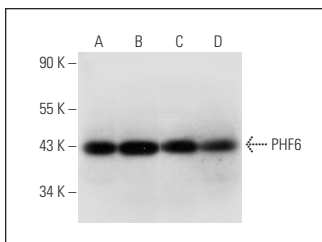
Molecular Weight of PHF6 isoforms: 41/35 kDa.

Positive Controls: HeLa nuclear extract: sc-2120, Jurkat nuclear extract: sc-2132 or K-562 whole cell lysate: sc-2203.

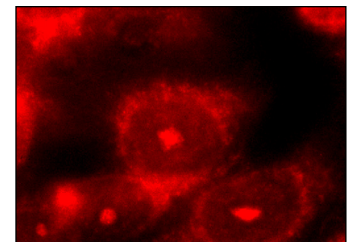
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 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 m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



PHF6 (B-9): sc-365013. Western blot analysis of PHF6 expression in HeLa (A) and Jurkat (B) nuclear extracts and K-562 (C) and RT-4 (D) whole cell lysates.



PHF6 (B-9): sc-365013. Immunofluorescence staining of methanol-fixed HeLa cells showing nucleolar and cytoplasmic localization.

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.