

# PHF6 (C-6): sc-514644

## 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 Börjeson-Forsman-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

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2. Lower, K.M., et al. 2002. Mutations in PHF6 are associated with Börjeson-Forsman-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-Forsman-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-Forsman-Lehmann syndrome. *J. Med. Genet.* 41: 778-783.
5. Crawford, J., et al. 2006. Mutation screening in Börjeson-Forsman-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-Forsman-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-Forsman-Lehmann syndrome in patients with PHF6 mutations. *Am. J. Med. Genet. A* 149A: 246-250.
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## CHROMOSOMAL LOCATION

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

## SOURCE

PHF6 (C-6) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 52-69 within an internal region of PHF6 of human origin.

## RESEARCH USE

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

## PRODUCT

Each vial contains 200 µg IgG<sub>1</sub> kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-514644 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

## APPLICATIONS

PHF6 (C-6) is recommended for detection of PHF6 isoforms 1 and 2 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.

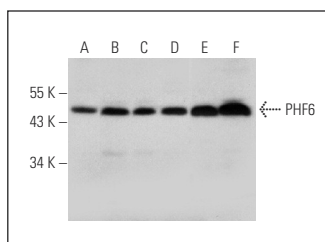
Molecular Weight of PHF6 isoforms: 41/35 kDa.

Positive Controls: A-431 nuclear extract: sc-2122, RT-4 whole cell lysate: sc-364257 or K-562 whole cell lysate: sc-2203.

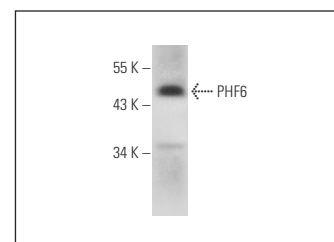
## RECOMMENDED SUPPORT REAGENTS

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, 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 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 (C-6): sc-514644. Western blot analysis of PHF6 expression in K-562 (A), A-431 (B), HeLa (C) and Jurkat (D) nuclear extracts and K-562 (E) and RT-4 (F) whole cell lysates.



PHF6 (C-6): sc-514644. Western blot analysis of PHF6 expression in MCF7 whole cell lysate.

## STORAGE

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