

# NIPBL (I-17): sc-69437

## BACKGROUND

NIPBL (Nipped-B-like protein), also known as IDN3, Delangin or CDLS, is a 2,804 amino acid nuclear protein that is the mammalian homolog of the *Drosophila* Nipped-B gene product, a protein that plays a role in developmental regulation by facilitating the communication between promoters and transcriptional enhancers. Widely expressed with particularly high levels present in skeletal muscle, heart, liver and kidney, NIPBL contains five HEAT repeats and interacts with HP1 $\alpha$ , possibly playing a role in sister chromatid adhesion and in the maintenance of proper chromatin structure. NIPBL exists as three isoforms and is subject to DNA damage-dependent phosphorylation, probably by ATM or ATR. Mutations in the gene encoding NIPBL are the cause of Cornelia de Lange syndrome type 1 (CDLS1), a developmental disorder that is characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation and various other malformations, including gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies.

## REFERENCES

1. Krantz, I.D., et al. 2001. Exclusion of linkage to the CDLS1 gene region on chromosome 3q26.3 in some familial cases of Cornelia de Lange syndrome. *Am. J. Med. Genet.* 101: 120-129.
2. Gillis, L.A., et al. 2004. NIPBL mutational analysis in 120 individuals with Cornelia de Lange syndrome and evaluation of genotype-phenotype correlations. *Am. J. Hum. Genet.* 75: 610-623.
3. Borck, G., et al. 2004. NIPBL mutations and genetic heterogeneity in Cornelia de Lange syndrome. *J. Med. Genet.* 41: e128.
4. Rollins, R.A., et al. 2004. *Drosophila* Nipped-B protein supports sister chromatid cohesion and opposes the stromalin/Scs3 cohesion factor to facilitate long-range activation of the cut gene. *Mol. Cell. Biol.* 24: 3100-3111.
5. Krantz, I.D., et al. 2004. Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of *Drosophila melanogaster* Nipped-B. *Nat. Genet.* 36: 631-635.
6. Tonkin, E.T., et al. 2004. NIPBL, encoding a homolog of fungal Scs2-type sister chromatid cohesion proteins and fly Nipped-B, is mutated in Cornelia de Lange syndrome. *Nat. Genet.* 36: 636-641.
7. Borck, G., et al. 2006. Father-to-daughter transmission of Cornelia de Lange syndrome caused by a mutation in the 5' untranslated region of the NIPBL gene. *Hum. Mutat.* 27: 731-735.
8. Bhuiyan, Z.A., et al. 2007. Large genomic rearrangements in NIPBL are infrequent in Cornelia de Lange syndrome. *Eur. J. Hum. Genet.* 15: 505-508.
9. Online Mendelian Inheritance in Man, OMIM<sup>™</sup>. 2007. Johns Hopkins University, Baltimore, MD. MIM Number: 608667. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

## CHROMOSOMAL LOCATION

Genetic locus: NIPBL (human) mapping to 5p13.2; Nipbl (mouse) mapping to 15 A1.

## SOURCE

NIPBL (I-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of NIPBL of human origin.

## PRODUCT

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

Blocking peptide available for competition studies, sc-69437 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

NIPBL (I-17) is recommended for detection of NIPBL of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), 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).

NIPBL (I-17) is also recommended for detection of NIPBL in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for NIPBL siRNA (h): sc-75921, NIPBL siRNA (m): sc-75922, NIPBL shRNA Plasmid (h): sc-75921-SH, NIPBL shRNA Plasmid (m): sc-75922-SH, NIPBL shRNA (h) Lentiviral Particles: sc-75921-V and NIPBL shRNA (m) Lentiviral Particles: sc-75922-V.

Molecular Weight of NIPBL: 316 kDa.

## 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<sup>™</sup> compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker<sup>™</sup> Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) 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<sup>™</sup> Mounting Medium: sc-24941.

## 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.