

Product name:	NIPBL Rabbit Polyclonal Antibody
Cat number:	ABN14709
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human protein . at AA range: 560-640
Reactivity:	Human,Mouse
Applications:	IHC 1:50-1:300,ICC/IF 1:50-1:200
Molecular Weight:	308kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes the homolog of the *Drosophila melanogaster* Nipped-B gene product and fungal Scc2-type sister chromatid cohesion proteins. The *Drosophila* protein facilitates enhancer-promoter communication of remote enhancers and plays a role in developmental regulation. It is also homologous to a family of chromosomal adherins with broad roles in sister chromatid cohesion, chromosome condensation, and DNA repair. The human protein has a bipartite nuclear targeting sequence and a putative HEAT repeat. Condensins, cohesins and other complexes with chromosome-related functions also contain HEAT repeats. Mutations in this gene result in Cornelia de Lange syndrome, a disorder characterized by dysmorphic facial features, growth delay, limb reduction defects, and mental retardation. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jul 2008],developmental stage:In embryos, it is expressed in developing limbs and later in cartilage primordia of the ulna and of various hand bones. Sites of craniofacial expression include the cartilage primordium of the basioccipital and basisphenoid skull bones and elsewhere in the head and face, including a region encompassing the mesenchyme adjacent to the cochlear canal. Also expressed in the spinal column, notochord and surface ectoderm sclerotome and what seem to be migrating myoblasts. Expressed in the developing heart in the atrial and ventricular myocardium and in the ventricular tuberculae but absent in the endocardial cushions. Also expressed in the developing esophagus, trachea and midgut loops, in the bronchi of the lung and in the tubules of the metanephros. Expression in organs and tissues not typically affected in CDL (e.g., the developing trachea, bronchi, esophagus, heart and kidney) may reflect a bias towards underreporting of more subtle aspects of the phenotype or problems that typically present later in life. Expressed in the mesenchyme surrounding the cochlear canal possibly reflecting the hearing impairment commonly found. Weakly or not expressed in embryonic brain.,disease:Defects in NIPBL are the cause of Cornelia de Lange syndrome type 1 (CDLS1) [MIM:122470]. CDLS is a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. CDLS 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.,domain:Contains one Pro-Xaa-Val-Xaa-Leu (PxVxL) motif, which is required for interaction with chromoshadow domains. This motif requires additional residues -7, -6, +4 and +5 of the central Val which contact the chromoshadow domain.,function:Probably plays a structural role in chromatin. Involved in sister chromatid cohesion, possibly by interacting with the cohesin complex.,miscellaneous:The sequence shown here is derived from an EMBL/GenBank/DDBJ third party annotation (TPA) entry.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,sequence caution:Chimeric cDNA.,similarity:Belongs to the SCC2/Nipped-B family.,similarity:Contains 5 HEAT repeats.,subunit:Interacts directly with CBX5 via the PxVxL motif.,tissue specificity:Widely expressed. Highly expressed in heart, skeletal muscle, fetal and adult liver, fetal and adult kidney. Expressed at intermediated level in thymus, placenta, peripheral leukocyte and small intestine. Weakly or not expressed in brain, colon, spleen and lung.,