

## NIPBL 抗原（重组蛋白）

中文名称：NIPBL 抗原（重组蛋白）

英文名称：NIPBL Antigen (Recombinant Protein)

别名：CDLS; IDN3; Scc2; CDLS1; IDN3-B

储存：冷冻（-20℃）

相关类别：抗原

概述：

Fusion protein corresponding to a region derived from 2523-2687 amino acids of human NIPBL

技术规格：

<b>Full name:</b>	NIPBL, cohesin loading factor
<b>Synonyms:</b>	CDLS; IDN3; Scc2; CDLS1; IDN3-B
<b>Swissprot:</b>	Q6KC79
<b>Gene Accession:</b>	BC033847
<b>Purity:</b>	>85%, as determined by Coomassie blue stained SDS-PAGE
<b>Expression system:</b>	Escherichia coli
<b>Tags:</b>	His tag C-Terminus, GST tag N-Terminus
<b>Background:</b>	This gene encodes the homolog of the <i>Drosophila melanogaster</i> Nipped-B gene product and fungal Scc2-type sister chromatid cohesion proteins. The <i>Drosophila</i> 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.

ts. Mutations in this gene result in Cornelia de Lange syndrome, a disorder characterized by dysmorphic facial features, growth delay, limb reduction defects, and cognitive disability. Two transcript variants encoding different isoforms have been found for this gene.