

Recombinant Human NIPBL

Catalog No: #GP13257



Package Size: #GP13257-1 100ug

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Description

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| Product Name | Recombinant Human NIPBL |
| Brief Description | Recombinant Protein |
| Immunogen Description | Fusion protein corresponding to a region derived from 2523-2687 amino acids of human NIPBL |
| Target Name | NIPBL, cohesin loading factor |
| Other Names | CDLS; IDN3; Scc2; CDLS1; IDN3-B |
| Accession No. | Swissprot:Q6KC79Gene Accession:BC033847 |
| Uniprot | Q6KC79 |
| GeneID | 25836; |
| Storage | -20~-80°C, pH 7.6 PBS |

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 cognitive disability. Two transcript variants encoding different isoforms have been found for this gene.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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