NIPBL gene

NIPBL cohesin loading factor

Normal Function

The *NIPBL* gene provides instructions for making a protein called delangin, which plays an important role in human development. Delangin helps control the activity of chromosomes during cell division. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids. The sister chromatids are attached to one another during the early stages of cell division by a group of proteins known as the cohesin complex. Delangin plays a critical role in the regulation of this complex. Specifically, it controls the interaction between the cohesion complex and the DNA that makes up the sister chromatids.

Researchers believe that delangin, as a regulator of the cohesin complex, also plays important roles in stabilizing cells' genetic information, repairing damaged DNA, and controlling the activity of certain genes that are essential for normal development.

Health Conditions Related to Genetic Changes

Cornelia de Lange syndrome

Variants (also called mutations) in the *NIPBL* gene have been found in people with Cornelia de Lange syndrome, a developmental disorder that affects many parts of the body. Variants in this gene are the most common known cause of Cornelia de Lange syndrome, accounting for more than half of all cases.

Many different kinds of *NIPBL* gene variants have been reported; most lead to the production of an abnormally short (truncated), nonfunctional version of the delangin protein from one copy of the gene in each cell. These variants reduce the overall amount of delangin produced in cells, which likely alters the activity of the cohesin complex and impairs its ability to regulate genes that are critical for normal development. Although researchers do not fully understand how these changes cause Cornelia de Lange syndrome, they suspect that altered gene regulation probably underlies many of the developmental problems characteristic of the condition. Studies suggest that variants leading to a nonfunctional version of delangin tend to cause more severe signs and symptoms than variants that result in a partially functional version of the protein.

Other Names for This Gene

- CDLS
- IDN3
- IDN3-B
- NIPBL_HUMAN
- Nipped-B homolog (Drosophila)
- Nipped-B-like
- Scc2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NIPBL (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=25836[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28NIPBL%5BTI%5D%29+OR+%28delangin%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- NIPPED-B-LIKE; NIPBL (https://omim.org/entry/608667)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/25836)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NIPBL[gene])

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Genomic Location

The *NIPBL* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromoso

me/5/).

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