NIPBL Gene

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Contributor: Lily Guo

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1. Introduction

The *NIPBL* gene provides instructions for making a protein called delangin, which plays an important role in human development. Before birth, delangin is found in the developing arms and legs, the bones of the skull and face, the spinal column, the heart, and other parts of the body.

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.

2. Health Conditions Related to Genetic Changes

2.1. Cornelia de Lange syndrome

More than 300 mutations in the *NIPBL* gene have been identified in people with Cornelia de Lange syndrome, a developmental disorder that affects many parts of the body. Mutations 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 mutations 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 mutations 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 mutations leading to a nonfunctional version of delangin tend to cause more severe signs and symptoms than mutations that result in a partially functional version of the protein.

3. Other Names for This Gene

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

- Scc2

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