SMC3 Gene

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structural maintenance of chromosomes 3

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1. Normal Function

The *SMC3* gene provides instructions for making a protein that is part of the structural maintenance of chromosomes (SMC) family. Within the nucleus, SMC proteins help regulate the structure and organization of chromosomes.

The protein produced from the *SMC3* gene helps control 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, which are attached to one another during the early stages of cell division. The SMC3 protein is part of a protein group called the cohesin complex that holds the sister chromatids together.

Researchers believe that the SMC3 protein, as a structural component of the cohesin complex, also plays important roles in stabilizing cells' genetic information, repairing damaged DNA, and regulating the activity of certain genes that are essential for normal development.

Although the SMC3 protein is found primarily in the nucleus, some of this protein is transported out of cells. The exported protein, which is usually called bamacan, may be involved in sticking cells together (cell adhesion) and cell growth. Bamacan is a component of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. Little else is known about the function of this protein outside the cell, but it appears to be important for normal development.

2. Health Conditions Related to Genetic Changes

2.1. Cornelia de Lange syndrome

At least 15 mutations in the *SMC3* gene have been found to cause Cornelia de Lange syndrome, a developmental disorder that affects many parts of the body. Researchers estimate that mutations in this gene account for 1 to 2 percent of all cases of this condition.

Most of the *SMC3* gene mutations that cause Cornelia de Lange syndrome either change single protein building blocks (amino acids) in the SMC3 protein or add or delete a small number of amino acids in the protein. Each of these mutations alters the structure and function of the protein, which likely interferes with 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 in the *SMC3* gene tend to cause a form of Cornelia de Lange syndrome with relatively mild features. Compared to mutations in the *NIPBL* gene, which are the most common known cause of the disorder, *SMC3* gene mutations often cause less significant delays in development and growth and are less likely to cause major birth defects.

3. Other Names for This Gene

- BAM
- bamacan
- basement membrane-associated chondroitin proteoglycan

- BMH
- chondroitin sulfate proteoglycan 6
- chromosome-associated polypeptide
- CSPG6
- HCAP
- SMC3_HUMAN
- SMC3L1

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