RAD21 Gene

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RAD21 cohesin complex component

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1. Normal Function

The *RAD21* gene provides instructions for making a protein that is involved in regulating the structure and organization 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, which are attached to one another during the early stages of cell division. The RAD21 protein is part of a protein group called the cohesin complex that holds the sister chromatids together.

Researchers believe that the RAD21 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.

2. Health Conditions Related to Genetic Changes

2.1. Cornelia de Lange syndrome

At least eight mutations in the *RAD21* 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 appear to be an uncommon cause of this condition.

Some cases of Cornelia de Lange syndrome have resulted from a deletion that removes a segment of DNA on chromosome 8 including the *RAD21* gene. In these cases, the entire gene is missing from one copy of the chromosome in each cell, so cells produce a reduced amount of RAD21 protein. In other cases, the condition is caused by mutations within the *RAD21* gene that impair or eliminate the function of the RAD21 protein. A defective or missing RAD21 protein likely alters the activity of the cohesin complex, impairing 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 *RAD21* gene 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, *RAD21* gene mutations cause less significant delays in development and growth and are less likely to cause major birth defects.

2.2. Trichorhinophalangeal syndrome type II

The *RAD21* gene is located in a region of chromosome 8 that is deleted in people with trichorhinophalangeal syndrome type II (TRPS II). TRPS II is a condition that causes bone and joint malformations; distinctive facial features; intellectual disability; and abnormalities of the skin, hair, teeth, sweat glands, and nails. As a result of this deletion, affected individuals are missing one copy of the *RAD21* gene in each cell, so cells produce a reduced amount of RAD21 protein. A shortage of RAD21 protein may contribute to intellectual disability, but the mechanism is unclear. The deletion of other genes near the *RAD21* gene likely contributes to the additional features of this condition.

3. Other Names for This Gene

- CDLS4
- double-strand-break repair protein rad21 homolog
- hHR21
- HR21
- HRAD21
- KIAA0078
- kleisin
- MCD1
- nuclear matrix protein 1
- NXP-1
- NXP1
- RAD21 homolog
- SCC1
- SCC1 homolog
- sister chromatid cohesion 1

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