RECQL4

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RecQ like helicase 4

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1. Normal Function

The *RECQL4* gene provides instructions for making one member of a protein family called RecQ helicases. Helicases are enzymes that bind to DNA and temporarily unwind the two spiral strands (double helix) of the DNA molecule. This unwinding is necessary for copying (replicating) DNA in preparation for cell division, and for repairing damaged DNA. Because RecQ helicases maintain the structure and integrity of DNA, they are known as the "caretakers of the genome."

The RECQL4 protein is active in several types of cells before and after birth. Researchers believe that this protein is particularly important in cells of the developing bones and skin. It has also been found in enterocytes, which are cells that line the intestine and absorb nutrients.

2. Health Conditions Related to Genetic Changes

2.1. Baller-Gerold syndrome

Several mutations in the *RECQL4* gene have been identified in people with Baller-Gerold syndrome. Most of these mutations prevent the production of any RECQL4 protein or change the way the protein is pieced together, which disrupts its usual function. A shortage of this protein may prevent normal DNA replication and repair, causing widespread damage to a person's genetic information over time. It is unclear how these changes result in the varied signs and symptoms of Baller-Gerold syndrome, including the abnormal fusion of certain skull bones (craniosynostosis), small stature, missing thumbs or bones in the forearm (radial ray malformations), and a skin rash.

2.2. RAPADILINO syndrome

At least 10 mutations in the *RECQL4* gene have been identified in people with RAPADILINO syndrome. This condition has many features, including radial ray malformations, malformed or missing kneecaps, diarrhea, and short stature. The condition was first identified in Finland, and the most common mutation in RAPADILINO syndrome is found in all affected individuals of Finnish descent as well as some people from other populations. This mutation, which is written as IVS7+2delT, is known as a splice-site mutation, and it causes the RECQL4 protein to be pieced together incorrectly. This genetic change results in the production of a protein that is missing a region called exon 7. The altered protein does not have helicase activity, which may prevent normal DNA replication and repair. These changes may result in the accumulation of DNA errors and cell death, although it is unclear exactly how *RECQL4* gene mutations lead to the specific features of RAPADILINO syndrome.

2.3. Rothmund-Thomson syndrome

More than 40 mutations in the *RECQL4* gene have been found in people with Rothmund-Thomson syndrome. These mutations likely prevent the production of any RECQL4 protein or lead to the production of an abnormally short, nonfunctional version of the protein. A shortage of this protein may prevent normal DNA replication and repair, causing widespread damage to a person's genetic information over time. Further study is needed to determine how these changes result in the characteristic features of Rothmund-Thomson syndrome, which include a skin rash, sparse hair, small stature, skeletal abnormalities, and an increased risk of certain cancers.

Because Rothmund-Thomson syndrome, Baller-Gerold syndrome, and RAPADILINO syndrome have overlapping features and can be caused by mutations in the same gene, researchers are investigating whether they are separate disorders or part of a single syndrome with overlapping signs and symptoms.

3. Other Names for This Gene

- ATP-Dependent DNA Helicase Q4
- RecQ helicase-like 4
- RecQ protein 4
- RecQ Protein Like 4
- RecQ protein-like 4
- RECQ4
- RECQ4_HUMAN
- RTS

The entry is from https://medlineplus.gov/genetics/gene/recql4

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