CTC1 gene

CST telomere replication complex component 1

Normal Function

The *CTC1* gene provides instructions for making a protein that plays an important role in structures known as telomeres, which are found at the ends of chromosomes. Telomeres are short, repetitive segments of DNA that help protect chromosomes from abnormally sticking together or breaking down (degrading). In most cells, telomeres become progressively shorter as the cell divides. After a certain number of cell divisions, the telomeres become so short that they trigger the cell to stop dividing or to self-destruct (undergo apoptosis).

The CTC1 protein works as part of a group of proteins known as the CST complex, which is involved in the maintenance of telomeres. This complex is part of the special machinery that some cells use to copy (replicate) telomeres so they do not become too short as cells divide. Studies suggest that the CTC1 protein may also have roles in DNA replication unrelated to telomeres, but these functions are not well understood.

Health Conditions Related to Genetic Changes

Coats plus syndrome

At least 20 mutations in the *CTC1* gene have been identified in people with Coats plus syndrome. This disorder is characterized by an eye condition called Coats disease plus abnormalities of the brain, bones, gastrointestinal system, and other parts of the body.

Most people with Coats plus syndrome have a mutation in one copy of the *CTC1* gene in each cell that eliminates the function of the CTC1 protein and a mutation in the other copy of the gene that reduces but does not eliminate the protein's function. This combination of mutations leaves only a small amount of functional CTC1 protein available to work as part of the CST complex. The resulting impairment of this complex affects the replication of telomeres, although the effect on telomere structure and function is unclear. Some studies have found that people with *CTC1* gene mutations have abnormally short telomeres, while other studies have found no change in telomere length. Researchers are working to determine how telomeres are different in people with *CTC1* gene mutations and how these changes could underlie the varied signs and symptoms of Coats plus syndrome.

Dyskeratosis congenita

MedlinePlus Genetics provides information about Dyskeratosis congenita

Other Names for This Gene

- AAF-132
- AAF132
- alpha accessory factor 132
- C17orf68
- conserved telomere capping protein 1
- CRMCC
- CST complex subunit CTC1
- CTS telomere maintenance complex component 1
- FLJ22170
- HBV DNAPTP1-transactivated protein B
- RP11-849F2.8
- tmp494178

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CTC1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80169[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CTC1%5BTIAB%5D%29+AND+%28telomere*%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- CONSERVED TELOMERE MAINTENANCE COMPONENT 1; CTC1 (https://omim.org/entry/613129)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/80169)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CTC1[gene])

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Genomic Location

The *CTC1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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