TINF2 gene

TERF1 interacting nuclear factor 2

Normal Function

The *TINF2* gene provides instructions for making part of the shelterin protein complex. This complex consists of a group of proteins that work together to help maintain structures known as telomeres, which are found at the ends of chromosomes. Telomeres help protect chromosomes from abnormally sticking together or breaking down (degrading).

The shelterin complex helps protect telomeres from the cell's DNA repair process. Without the protection of shelterin, the repair mechanism would sense the chromosome ends as abnormal breaks in the DNA sequence and either attempt to join the ends together or initiate cellular self-destruction (apoptosis).

Health Conditions Related to Genetic Changes

Dyskeratosis congenita

At least 15 mutations in the *TINF2* gene have been identified in people with dyskeratosis congenita, including a severe form of this disorder called Revesz syndrome. Dyskeratosis congenita is characterized by changes in skin coloring (pigmentation), white patches inside the mouth (oral leukoplakia), and abnormally formed fingernails and toenails (nail dystrophy). People with dyskeratosis congenita have an increased risk of developing several life-threatening conditions, including cancer and a progressive lung disease called pulmonary fibrosis. Many affected individuals also develop a serious condition called aplastic anemia, also known as bone marrow failure, which occurs when the bone marrow does not produce enough new blood cells.

Most of the *TINF2* gene mutations that cause dyskeratosis congenita change single protein building blocks (amino acids) in the TINF2 protein, likely disrupting the function of the protein. The mutations result in dysfunction of the shelterin complex, interfering with its protection of telomeres and leading to reduced telomere length. Shortened telomeres can result in damage to genetic material, causing the cell to stop dividing or to undergo apoptosis.

Cells that divide rapidly are especially vulnerable to the effects of shortened telomeres.

As a result, people with dyskeratosis congenita may experience a variety of problems affecting quickly dividing cells in the body such as cells of the nail beds, hair follicles, skin, lining of the mouth (oral mucosa), and bone marrow.

Breakage and instability of chromosomes resulting from inadequate telomere maintenance may lead to genetic changes that allow cells to divide in an uncontrolled way, resulting in the development of cancer in some people with dyskeratosis congenita.

Idiopathic pulmonary fibrosis

MedlinePlus Genetics provides information about Idiopathic pulmonary fibrosis

Other Names for This Gene

- (TRF1)-interacting nuclear factor 2 variant 1
- TERF1 (TRF1)-interacting nuclear factor 2
- TERF1-interacting nuclear factor 2
- TERF1-interacting nuclear factor 2 isoform 1
- TERF1-interacting nuclear factor 2 isoform 2
- TIN2
- TINF2_HUMAN
- TRF1-interacting nuclear protein 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TINF2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26277[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TINF2%5BTIAB%5D%29+OR+%28TIN2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TERF1-INTERACTING NUCLEAR FACTOR 2; TINF2 (https://omim.org/entry/604319)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/26277)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TINF2[gene])

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Genomic Location

The *TINF2* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

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