TNFRSF13B gene

TNF receptor superfamily member 13B

Normal Function

The *TNFRSF13B* gene provides instructions for making a protein called TACI. The TACI protein is found on the surface of immune system cells called B cells. These specialized white blood cells help protect the body against infection from foreign invaders such as bacteria and viruses. When B cells mature, they produce special proteins called antibodies (also known as immunoglobulins). Antibodies attach to specific foreign invaders, marking them for destruction. Through interactions with other proteins, TACI promotes cell signaling, plays a role in B cell survival and maturation, and is involved in the production of antibodies.

Health Conditions Related to Genetic Changes

Common variable immune deficiency

More than 25 mutations in the *TNFRSF13B* gene have been associated with common variable immune deficiency (CVID). This condition impairs the immune system, resulting in increased risk for recurrent infections; autoimmune disorders, which occur when the immune system malfunctions and attacks the body's tissues and organs; and certain cancers.

Most of the *TNFRSF13B* gene mutations associated with CVID change single protein building blocks (amino acids) in the TACI protein. The most common mutation seen in people with CVID replaces the amino acid cysteine with the amino acid arginine at position 104 in the TACI protein (written as Cys104Arg or C104R). This mutation impairs the ability of TACI to interact with other proteins, disrupting cell signaling and preventing normal B cell maturation and antibody production. A shortage (deficiency) of certain antibodies makes it difficult for people to fight off infections. Abnormal and deficient immune responses over time likely contribute to the increased cancer risk in people with CVID.

Some people with *TNFRSF13B* gene mutations do not develop the signs and symptoms of CVID. In these individuals, additional genetic or environmental factors are probably needed for the condition to occur.

Other Names for This Gene

- CD267
- IGAD2
- TACI
- TR13B_HUMAN
- transmembrane activator and CAML interactor
- tumor necrosis factor receptor 13B
- tumor necrosis factor receptor superfamily member 13B
- tumor necrosis factor receptor superfamily, member 13B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TNFRSF13B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23495[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TNFRSF13B%5BTIAB%5D%29+OR+%28TACI%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 13B; TNFRSF13B (https://omim.org/entry/604907)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23495)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TNFRSF13B[gene])

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Genomic Location

The *TNFRSF13B* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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