TCN2 gene

transcobalamin 2

Normal Function

The *TCN2* gene provides instructions for making a protein called transcobalamin (formerly known as transcobalamin II). This protein transports cobalamin (also known as vitamin B12) from the bloodstream to cells throughout the body. Cobalamin is obtained from the diet; this vitamin is found in animal products such as meat, eggs, and shellfish.

During digestion, cobalamin is transported through intestinal cells into the bloodstream. Transcobalamin attaches (binds) to cobalamin when it is released into the bloodstream and transports the vitamin to cells. The transcobalamin-cobalamin complex binds to a receptor on the cell surface, which allows the complex to enter the cell. Transcobalamin releases cobalamin when the complex enters the cell and transcobalamin is broken down.

Within cells, cobalamin helps certain enzymes carry out chemical reactions. Cobalamin plays a role in the processes that produce the building blocks of DNA (nucleotides) and break down various compounds such as fatty acids; these processes are needed for cell growth and division (proliferation) and cellular energy production. Cobalamin's role in these processes is particularly important in the formation of new blood cells and in the nervous system.

Health Conditions Related to Genetic Changes

Transcobalamin deficiency

More than 20 mutations in the *TCN2* gene have been found to cause transcobalamin deficiency. This condition impairs the transport of cobalamin from the bloodstream to cells throughout the body. Affected individuals have difficulty gaining weight and growing at the expected rate (failure to thrive), vomiting, diarrhea, a shortage of all types of blood cells, and neurological problems. Many *TCN2* gene mutations lead to a complete or near-complete lack (deficiency) of transcobalamin. Other *TCN2* gene mutations result in a transcobalamin protein that cannot bind to cobalamin or a protein that cannot bind to the receptor at the surface of cells. The resulting lack of cobalamin within cells interferes with the functioning of certain enzymes, which impacts many cell activities. As a result, a wide range of signs and symptoms characteristic of transcobalamin deficiency can develop.

Other Names for This Gene

- D22S676
- D22S750
- TC
- TC II
- TC-2
- TC2
- TCII
- transcobalamin II
- transcobalamin-2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TCN2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6948[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TCN2%5BTI%5D%29+OR+%28transcobalamin+II%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TRANSCOBALAMIN II; TCN2 (https://omim.org/entry/613441)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6948)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TCN2[gene])

References

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- Nissen PH, Nordwall M, Hoffmann-Lucke E, Sorensen BS, Nexo E.

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- Seetharam B, Li N. Transcobalamin II and its cell surface receptor. VitamHorm. 2000;59:337-66. doi: 10.1016/s0083-6729(00)59012-8. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10714245)

Genomic Location

The *TCN2* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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