

TCN2 Gene

Subjects: **Genetics & Heredity**

Contributor: Rui Liu

Transcobalamin 2: The *TCN2* gene provides instructions for making a protein called transcobalamin (formerly known as transcobalamin II).

genes

1. 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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

3. Other Names for This Gene

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

References

1. Nielsen MJ, Rasmussen MR, Andersen CB, Nexø E, Moestrup SK. Vitamin B12 transport from food to the body's cells--a sophisticated, multistep pathway. *Nat Rev Gastroenterol Hepatol*. 2012 May 1;9(6):345-54. doi: 10.1038/nrgastro.2012.76. Review.
2. Nissen PH, Nordwall M, Hoffmann-Lücke E, Sorensen BS, Nexo E. Transcobalamin deficiency caused by compound heterozygosity for two novel mutations in the TCN2 gene: a study of two affected siblings, their brother, and their parents. *J Inherit Metab Dis*. 2010 Dec;33 Suppl 3:S269-74. doi: 10.1007/s10545-010-9145-z.
3. Qian L, Quadros EV, Regec A, Zittoun J, Rothenberg SP. Congenital transcobalamin II deficiency due to errors in RNA editing. *Blood Cells Mol Dis*. 2002 Mar-Apr;28(2):134-42; discussion 143-5.
4. Seetharam B, Li N. Transcobalamin II and its cell surface receptor. *Vitam Horm*. 2000;59:337-66. Review.

Retrieved from <https://encyclopedia.pub/entry/history/show/12953>