CDAN1 gene

codanin 1

Normal Function

The *CDAN1* gene provides instructions for making a protein called codanin-1. Although this protein is active in cells throughout the body, very little is known about its function.

A recent study suggests that codanin-1 is associated with a form of DNA called heterochromatin. Heterochromatin is densely packed DNA that contains few functional genes, but it plays an important role in maintaining the structure of the nucleus (where most of the cell's DNA is located).

Researchers speculate that codanin-1 may be involved in the formation of red blood cells, a process called erythropoiesis. Specifically, this protein may play a key role in the organization of heterochromatin during the division of these developing cells.

Health Conditions Related to Genetic Changes

Congenital dyserythropoietic anemia

More than 30 mutations in the *CDAN1* gene have been identified in people with congenital dyserythropoietic anemia (CDA) type I. Most of these mutations change single protein building blocks (amino acids) in the codanin-1 protein. The *CDAN1* mutations that cause CDA type I likely reduce the function of codanin-1. However, researchers suspect that these mutations do not completely eliminate the function of the protein, which appears to be essential for life.

It is unclear how *CDAN1* mutations cause the characteristic features of CDA type I. A shortage of functional codanin-1 somehow disrupts the normal development of red blood cells. In people with CDA type I, immature red blood cells called erythroblasts are large, unusually shaped, and have an abnormally formed nucleus. These defective erythroblasts cannot develop into functional mature red blood cells. The resulting shortage of healthy red blood cells leads to the characteristic signs and symptoms of anemia, as well as complications including an enlarged liver and spleen (hepatosplenomegaly) and an abnormal buildup of iron that can damage the body's organs.

Other Names for This Gene

- CDA-I
- CDA1
- CDAI
- CDAN1_HUMAN
- codanin
- congenital dyserythropoietic anemia, type I
- DLT
- PRO1295

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CDAN1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=146059[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CDAN1%5BTIAB%5D%29+OR+%28codanin-1%5BTIAB%5D%29+OR+%28codanin+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CODANIN 1; CDAN1 (https://omim.org/entry/607465)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/146059)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CDAN1[gene])

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Genomic Location

The *CDAN1* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

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