CDAN1 gene

codanin 1

Normal Function

The *CDAN1* gene provides instructions for making a protein called codanin-1. Research suggests that codanin-1 plays an important role in maintaining the structure of the nucleus (where most of the cell's DNA is located). Specifically, codanin-1 is likely involved in putting together cell components called nucleosomes that help bundle DNA in a tight package. This tight packaging helps control gene expression.

Because codanin-1 has the ability to regulate gene activity, it is involved in many processes during development, including the growth of cells as they progress through the step-by-step process for replication (the cell cycle). Codanin-1 is also thought to be involved in the differentiation and development of red blood cells (erythropoiesis), though the protein's specific role in this process is unclear.

Health Conditions Related to Genetic Changes

Congenital dyserythropoietic anemia

Variants (also called mutations) in the *CDAN1* gene have been identified in people with congenital dyserythropoietic anemia (CDA) type I. This condition is characterized by a shortage of red blood cells that is caused by abnormal red blood cell formation (dyserythropoietic anemia). In people with CDA type I, immature red blood cells cannot develop into functional, mature cells. As a result, the number of mature and functioning red blood cells decreases, which leads to weakness, yellowing of the skin and eyes (jaundice), an enlarged liver and spleen (hepatosplenomegaly), the buildup of too much iron (iron overload), and skeletal problems in people with CDA type I.

Most *CDAN1* gene variants change single protein building blocks (amino acids) in the codanin-1 protein. The *CDAN1* gene variants that cause CDA type I do not completely eliminate the function of codanin-1, since codanin-1 appears to be essential for life. However, a shortage of functional codanin-1 likely disrupts the normal progression of the cell cycle, especially during the development of red blood cells.

In people with CDA type I, immature red blood cells called erythroblasts are abnormal, large, and have more than one nucleus. When viewed under a microscope, the inside of the nucleus of these cells can look speckled. These irregular 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 and the other features of CDA type I.

Other Names for This Gene

- CDA-I
- CDA1
- CDAN1_HUMAN
- codanin

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=((CDAN1%5BTIAB%5D)+OR+(codanin-1%5BTIAB%5D)+OR+(codanin+1%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3000+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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