# SEC23B gene

SEC23 homolog B, COPII component

## Normal Function

The *SEC23B* gene provides instructions for making one component of a large group of interacting proteins called coat protein complex II (COPII). COPII is involved in the formation of vesicles, which are small, sac-like structures that transport proteins and other materials within cells. Specifically, COPII triggers the formation of vesicles in a cellular structure called the endoplasmic reticulum (ER), which is involved in protein processing and transport. These COPII vesicles carry proteins that will be exported out of cells (secreted).

The SEC23B protein is very similar to the protein produced from a related gene, *SEC23A*. These proteins are both components of COPII, and they appear to have overlapping functions. In most types of cells, if one of these proteins is missing, the other may be able to compensate for the loss. However, research indicates that the SEC23B protein may have a unique function in developing red blood cells (erythroblasts) .

## Health Conditions Related to Genetic Changes

Congenital dyserythropoietic anemia

Variants (also called mutations) in the *SEC23B* gene have been identified in people with congenital dyserythropoietic anemia (CDA) type II.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 II, immature red blood cells are unusually shaped and 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), and the buildup of too much iron (iron overload) in people with CDA type II.

Most *SEC23B* gene variants change single protein building blocks (amino acids) in the SEC23B protein, likely disrupting its function. It is unclear how *SEC23B* gene variants cause the characteristic features of CDA type II. It is thought that the decrease in normal SEC23B protein affects erythroblasts because these cells do not have enough SEC23A protein to make up for the loss. This shortage of SEC23B protein leads to the

production of irregular erythroblasts that 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 II.

Cowden syndrome

MedlinePlus Genetics provides information about Cowden syndrome

## Other Names for This Gene

- HEMPAS
- SC23B_HUMAN
- transport protein SEC23B

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SEC23B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10483[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SEC23*%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SEC23 HOMOLOG B, COAT COMPLEX II COMPONENT; SEC23B (https://omim.org/entry/610512)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10483)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SEC23B[gene])

## References


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## Genomic Location

The *SEC23B* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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