SEC23B gene

SEC23 homolog B, COPII coat complex 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 are destined to 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

At least 20 mutations in the *SEC23B* gene have been identified in people with congenital dyserythropoietic anemia (CDA) type II. Most of these mutations change single protein building blocks (amino acids) in the SEC23B protein. Other mutations delete genetic material from the *SEC23B* gene or alter the way the gene's instructions are used to make the SEC23B protein. The mutations responsible for CDA type II likely disrupt the function of the SEC23B protein. 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 *SEC23B* mutations cause the characteristic features of CDA type II. The abnormal SEC23B protein leads to the production of erythroblasts that are unusually shaped and may have extra nuclei. 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.

Cowden syndrome

MedlinePlus Genetics provides information about Cowden syndrome

Other Names for This Gene

- CDA-II
- CDAII
- HEMPAS
- SC23B_HUMAN
- Sec23 homolog B
- Sec23 homolog B (S. cerevisiae)
- Sec23 homolog B, COPII coat complex component
- SEC23-like protein B
- SEC23-related protein B
- 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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