

CCBE1 gene

collagen and calcium binding EGF domains 1

Normal Function

The *CCBE1* gene provides instructions for making a protein that is found in the lattice of proteins and other molecules outside the cell (extracellular matrix). The CCBE1 protein is involved in the formation of the lymphatic system, which consists of a network of vessels that transport lymph fluid and immune cells throughout the body. Specifically, the CCBE1 protein helps guide maturation (differentiation) and movement (migration) of immature cells called lymphangioblasts that will eventually form the lining (epithelium) of lymphatic vessels.

Health Conditions Related to Genetic Changes

Hennekam syndrome

At least 13 mutations in the *CCBE1* gene have been found to cause Hennekam syndrome, an inherited disorder resulting from malformation of the lymphatic system. Most *CCBE1* gene mutations change single protein building blocks (amino acids) in the CCBE1 protein leading to a change in the three-dimensional shape of the protein. The abnormal protein cannot play its role in the formation of the lymphatic vessel epithelium. A poorly formed lymphatic system leads to lymphatic vessels that are abnormally expanded (lymphangiectasia) and are prone to break open (rupture), puffiness or swelling caused by a buildup of fluid (lymphedema), and other features of Hennekam syndrome. *CCBE1* gene mutations account for about 25 percent of all cases of Hennekam syndrome.

Other Names for This Gene

- collagen and calcium-binding EGF domain-containing protein 1
- FLJ30681
- full of fluid protein homolog
- KIAA1983

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CCBE1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=147372\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=147372[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CCBE1%5BTIAB%5D%29+OR+%28EGF+domains+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- COLLAGEN AND CALCIUM-BINDING EGF DOMAIN-CONTAINING PROTEIN 1; CCBE1 (<https://omim.org/entry/612753>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/147372>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CCBE1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CCBE1[gene]))

References

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Genomic Location

The *CCBE1* gene is found on chromosome 18 (<https://medlineplus.gov/genetics/chromosome/18/>).

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