

COLEC10 gene

collectin subfamily member 10

Normal Function

The *COLEC10* gene provides instructions for making a protein called collectin liver 1 (CL-L1). This protein is involved in a series of steps called the lectin complement pathway, which is thought to help direct the movement (migration) of cells during development before birth to form the organs and systems of the body. The lectin complement pathway appears to be particularly important in directing the migration of neural crest cells. These cells give rise to various tissues including many tissues in the face and skull, glands that produce hormones (endocrine glands), and portions of the nervous system. After birth, the lectin complement pathway is involved in the immune system.

Health Conditions Related to Genetic Changes

3MC syndrome

At least three *COLEC10* gene mutations have been identified in people with 3MC syndrome, a disorder characterized by unusual facial features and a variety of problems affecting other tissues and organs. The *COLEC10* gene mutations that cause 3MC syndrome impair or eliminate the function of the CL-L1 protein, resulting in faulty control of cell migration in early development and leading to the various abnormalities that occur in this disorder. Researchers suggest that similar pathways in the immune system can compensate for problems in the lectin complement pathway, which explains why immune system abnormalities are not part of 3MC syndrome.

Other Names for This Gene

- 3MC3
- CL-34
- CL-L1
- CLL1
- collectin liver 1
- collectin liver protein 1
- collectin sub-family member 10 (C-type lectin)

- collectin-10 isoform 1 precursor
- collectin-10 isoform 2
- collectin-34

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COLEC10 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10584\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10584[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28COLEC10%5BTIAB%5D%29+OR+%28collectin+subfamily+member+10%5BTIAB%5D%29%29+OR+%28%28CL-34%5BTIAB%5D%29+OR+%28CL-L1%5BTIAB%5D%29+OR+%28CLL1%5BTIAB%5D%29+OR+%28collectin+liver+1%5BTIAB%5D%29+OR+%28collectin+liver+protein+1%5BTIAB%5D%29+OR+%28collectin+sub-family+member+10%5BTIAB%5D%29+OR+%28collectin-10+isoform+1+precursor%5BTIAB%5D%29+OR+%28collectin-10+isoform+2%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>)

Catalog of Genes and Diseases from OMIM

- COLLECTIN 10; COLEC10 (<https://omim.org/entry/607620>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10584>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COLEC10\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COLEC10[gene]))

References

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Genomic Location

The *COLEC10* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

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