COLEC11 gene

collectin subfamily member 11

Normal Function

The *COLEC11* gene provides instructions for making a protein called collectin kidney 1 (CL-K1). 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 nine *COLEC11* 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 *COLEC11* gene mutations that cause 3MC syndrome impair or eliminate the function of the CL-K1 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

- 3MC2
- CL-K1
- CL-K1-I
- CL-K1-II
- CL-K1-IIa
- CL-K1-IIb
- CLK1

- collectin K1
- collectin kidney protein 1
- MGC3279

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COLEC11 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=78989[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28COLEC11%5BTIAB%5D%29+OR+%28collectin+sub-family+member+11%5BTIAB%5D%29%29+OR+%28%283MC2%5BTIAB%5D%29+OR+%28CL-K1%5BTIAB%5D%29+OR+%28CLK1%5BTIAB%5D%29+OR+%28Collectin+K1%5BTIAB%5D%29+OR+%28collectin+kidney+protein+1%5BTIAB%5D%29+OR+%28collectin-11+isoform+a+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+b%5BTIAB%5D%29+OR+%28collectin-11+isoform+c+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+d+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+e+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+f+precursor%5BTIAB%5D%29+OR+%28collectin-11+isoform+g%5BTIAB%5D%29+OR+%28collectin-11+isoform+h%5BTIAB%5D%29+OR+%28collectin-11+isoform+i%5BTIAB%5D%29+OR+%28collectin-11+isoform+j%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- COLLECTIN 11; COLEC11 (https://omim.org/entry/612502)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/78989)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=COLEC11[gene])

References

- Bayarri-Olmos R, Hansen S, Henriksen ML, Storm L, Thiel S, Garred P,Munthe-Fog L. Genetic variation of COLEC10 and COLEC11 and association with serumlevels of collectin liver 1 (CL-L1) and collectin kidney 1 (CL-K1). PLoS One.2015 Feb 24;10(2):e0114883. doi: 10.1371/journal.pone.0114883. eCollection 2015. Citation on

PubMed (https://pubmed.ncbi.nlm.nih.gov/25710878) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4339841/)

- Hansen SW, Ohtani K, Roy N, Wakamiya N. The collectins CL-L1, CL-K1 and CL-P1,and their roles in complement and innate immunity. Immunobiology. 2016Oct;221(10):1058-67. doi: 10.1016/j.imbio.2016.05.012. Epub 2016 Jun 2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27377710)
- Henriksen ML, Brandt J, Andrieu JP, Nielsen C, Jensen PH, Holmskov U,Jorgensen TJ, Palarasah Y, Thielens NM, Hansen S. Heteromeric complexes of nativecollectin kidney 1 and collectin liver 1 are found in the circulation with MASPsand activate the complement system. J Immunol. 2013 Dec 15;191(12):6117-27. doi:10.4049/jimmunol.1302121. Epub 2013 Oct 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24174618)
- Ohtani K, Suzuki Y, Wakamiya N. Biological functions of the novel collectinsCL-L1, CL-K1, and CL-P1. J Biomed Biotechnol. 2012;2012:493945. doi:10.1155/2012/493945. Epub 2012 Apr 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22570530) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3336186/)
- Rooryck C, Diaz-Font A, Osborn DP, Chabchoub E, Hernandez-Hernandez V, Shamseldin H, Kenny J, Waters A, Jenkins D, Kaissi AA, Leal GF, Dallapiccola B, Carnevale F, Bitner-Glindzicz M, Lees M, Hennekam R, Stanier P, Burns AJ, PeetersH, Alkuraya FS, Beales PL. Mutations in lectin complement pathway genes COLEC11and MASP1 cause 3MC syndrome. Nat Genet. 2011 Mar;43(3):197-203. doi:10.1038/ng.757. Epub 2011 Jan 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21258343) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3045628/)
- Selman L, Hansen S. Structure and function of collectin liver 1 (CL-L1) andcollectin 11 (CL-11, CL-K1). Immunobiology. 2012 Sep;217(9):851-63. doi:10.1016/j.imbio.2011.12.008. Epub 2012 Feb 4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22475410)
- Venkatraman Girija U, Furze CM, Gingras AR, Yoshizaki T, Ohtani K, MarshallJE, Wallis AK, Schwaeble WJ, El-Mezgueldi M, Mitchell DA, Moody PC, Wakamiya N, Wallis R. Molecular basis of sugar recognition by collectin-K1 and the effects ofmutations associated with 3MC syndrome. BMC Biol. 2015 Apr 17;13:27. doi:10.1186/s12915-015-0136-2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25912189) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4431178/)

Genomic Location

The *COLEC11* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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