MED13L gene

mediator complex subunit 13L

Normal Function

The *MED13L* gene provides instructions for making a protein that is one piece (subunit) of a group of proteins known as the mediator complex. This complex regulates the activity (transcription) of genes. Transcription is the first step in the process by which information stored in a gene's DNA is used to build proteins.

The mediator complex physically links the proteins that can turn genes on, called transcription factors, with the enzyme that carries out transcription, called RNA polymerase II. Once transcription factors are attached to RNA polymerase II, transcription begins.

Researchers believe that as part of the mediator complex, the MED13L protein is involved in many aspects of early development, including development of the heart, nerve cells (neurons) in the brain, and structures in the face. The mediator complex plays a role in several chemical signaling pathways within cells. These pathways help direct a broad range of cellular activities, such as cell growth, cell movement (migration), and the process by which cells mature to carry out specific functions (differentiation).

Health Conditions Related to Genetic Changes

MED13L syndrome

More than 50 mutations in the *MED13L* gene have been found to cause *MED13L* syndrome. This condition is characterized by moderate to severe developmental delay and intellectual disability and minor differences in facial features. Additionally, some people with *MED13L* syndrome have recurrent seizures (epilepsy) or heart abnormalities that are present from birth (congenital heart defects).

Some *MED13L* gene mutations insert or delete regions of DNA within the *MED13L* gene. These genetic changes lead to a reduction in the total amount of MED13L protein in cells. Other mutations change single protein building blocks (amino acids) in the MED13L protein. It is thought that the altered protein interferes with the function of the normal protein produced from the non-mutated copy of the *MED13L* gene (such mutations are described as "dominant-negative"). Because dominant negative mutations impair the function of proteins made from both the altered copy of the

MED13L gene and the normal copy, individuals with dominant negative mutations tend to have more severe signs than people with mutations that affect protein production from just the altered copy of the gene. While it is likely that mutations in the *MED13L* gene impair the control of gene activity by the mediator complex, it is unclear how these changes lead to the particular cognitive and physical features of the disorder.

Critical congenital heart disease

MedlinePlus Genetics provides information about Critical congenital heart disease

Other Names for This Gene

- KIAA1025
- MEDIATOR COMPLEX SUBUNIT 13-LIKE
- PROSIT240
- PROTEIN SIMILAR TO TRAP240
- THRAP2
- THYROID HORMONE RECEPTOR-ASSOCIATED PROTEIN 2
- TRAP240-LIKE PROTEIN
- TRAP240L

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MED13L (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23389[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MED13L%5BTIAB%5D%29+OR+%28mediator+complex+subunit+13+like%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

- TRANSPOSITION OF THE GREAT ARTERIES, DEXTRO-LOOPED; DTGA (https://omim.org/entry/608808)
- MEDIATOR COMPLEX SUBUNIT 13-LIKE; MED13L (https://omim.org/entry/608771)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23389)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MED13L[gene])

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Genomic Location

The *MED13L* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

Last updated May 1, 2019