CD40LG gene

CD40 ligand

Normal Function

The *CD40LG* gene provides instructions for making a protein called CD40 ligand, which is found on the surface of immune system cells known as T cells. CD40 ligand attaches like a key in a lock to its receptor protein, CD40, which is located on the surface of immune system cells known as B cells. B cells are involved in the production of proteins called antibodies or immunoglobulins that help protect the body against infection. There are several classes of antibodies, and each one has a different function in the immune system. B cells are able to mature into the cells that produce immunoglobulin M (IgM) without any signals from other cells. In order for B cells to mature into the cells that produce antibodies of a different class, the CD40 receptor must interact with CD40 ligand. When these two proteins are connected, they trigger a series of chemical signals that instruct the B cell to start making immunoglobulin G (IgG), immunoglobulin A (IgA), and immunoglobulin E (IgE).

CD40 ligand is also necessary for T cells to interact with other cells of the immune system, and it plays a key role in T cell differentiation (the process by which cells mature to carry out specific functions).

Health Conditions Related to Genetic Changes

X-linked hyper IgM syndrome

More than 150 mutations in the *CD40LG* gene have been found to cause X-linked hyper IgM syndrome. These mutations lead to the production of an abnormal CD40 ligand or prevent production of this protein. If CD40 ligand does not attach to its receptor on B cells, these cells cannot produce IgG, IgA, or IgE antibodies. Mutations in the *CD40LG* gene also impair the T cell's ability to differentiate and interact with immune system cells. People with X-linked hyper IgM syndrome are more susceptible to infections because they do not have a properly functioning immune system.

Other Names for This Gene

- CD154
- CD40 antigen ligand
- CD40L

- CD40L_HUMAN
- gp39
- hCD40L
- HIGM1
- IGM
- IMD3
- T-B cell-activating molecule
- T-BAM
- TNF-related activation protein
- TNFSF5
- TRAP
- tumor necrosis factor (ligand) superfamily member 5

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CD40LG (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=959[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CD40LG%5BTIAB%5D%29+OR+%28CD40+ligand%5BTIAB%5D%29%29+AND+%28%28tumor+necrosis+factor+ligand+superfamily+member+5%5BMAJR%5D%29+OR+%28cd40+ligand%5BMAJR%5D%29+OR+%28cd154%5BMAJR%5D%29+OR+%28tnf+superfamily,+member+5%5BMAJR%5D%29+OR+%28cd40l%5BMAJR%5D%29%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CD40 LIGAND; CD40LG (https://omim.org/entry/300386)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/959)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CD40LG[gene])

References

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Genomic Location

The *CD40LG* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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