ANTXR2 gene

ANTXR cell adhesion molecule 2

Normal Function

The *ANTXR2* gene provides instructions for making a protein that is found at the surface of many types of cells. The ANTXR2 protein is believed to interact with components of the extracellular matrix, which is the lattice of proteins and other molecules outside the cell. This matrix strengthens and supports connective tissues, such as skin, bone, cartilage, tendons, and ligaments.

The ANTXR2 protein is involved in the formation of tiny blood vessels (capillaries). It may also be important for maintaining the structure of basement membranes, which are thin, sheet-like extracellular matrix structures that separate and support cells in many connective tissues. Research suggests that the ANTXR2 protein aids in the breakdown of at least one type of extracellular matrix protein, ensuring the correct balance of proteins is maintained for normal functioning of muscles and connective tissues.

The ANTXR2 protein also acts as a receptor for the toxin that causes anthrax, allowing the toxin to attach to cells and trigger disease.

Health Conditions Related to Genetic Changes

Hyaline fibromatosis syndrome

More than 45 mutations in the *ANTXR2* gene have been found to cause hyaline fibromatosis syndrome, a painful condition characterized by accumulation of a clear (hyaline) substance in different tissues in the body. The nature of the hyaline substance is unknown, but it likely contains extracellular matrix proteins, among other materials. Buildup of this material can cause firm lumps of noncancerous tissue (nodules) under the skin and in internal organs, joint deformities called contractures that restrict movement, and overgrowth of the gums. The severity of the signs and symptoms falls along a spectrum. The most severely affected individuals have severe diarrhea and recurrent infections and usually do not survive beyond early childhood. Individuals at the milder end of the spectrum typically survive into adulthood.

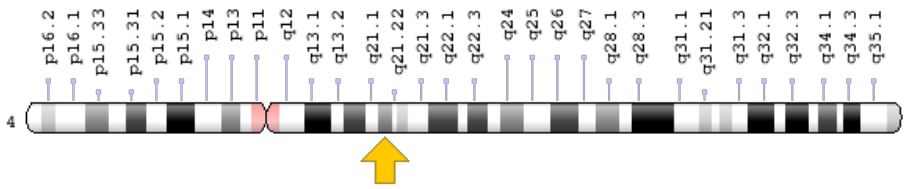
Some *ANTXR2* gene mutations reduce or eliminate the amount of ANTXR2 protein at the surface of cells. Others are thought to impair the protein's ability to interact with extracellular matrix components. It is unclear what effect these mutations have in cells and tissues. Researchers suspect that gene mutations disrupt the formation of basement membranes, allowing a hyaline substance to leak through and build up in various body tissues. Alternatively, the mutations could impair the breakdown of excess extracellular matrix proteins, which then accumulate in tissues and lead to the signs and symptoms of hyaline fibromatosis syndrome.

It is unclear why the severity of hyaline fibromatosis syndrome varies among affected individuals. Some studies have indicated that the severity of the condition may be linked to where in the gene the mutation occurs.

Chromosomal Location

Cytogenetic Location: 4q21.21, which is the long (q) arm of chromosome 4 at position 21.21

Molecular Location: base pairs 79,901,146 to 80,073,472 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- anthrax toxin receptor 2
- ANTR2_HUMAN
- capillary morphogenesis protein 2
- CMG-2
- CMG2
- FLJ31074
- ISH
- JHF
- MGC111533
- MGC45856

Additional Information & Resources

Clinical Information from GeneReviews

- Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ANTXR2%5BTIAB%5D%29+OR+%28anthrax+toxin+receptor+2%5BTIAB%5D%29%29+OR+%28%28CMG2%5BTIAB%5D%29+OR+%28CMG-2%5BTIAB%5D%29+OR+%28capillary+morphogenesis+protein+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+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ANTHRAX TOXIN RECEPTOR 2
  http://omim.org/entry/608041

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ANTXR2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ANTXR2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:21732
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:118429
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/118429
- UniProt
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