

FBN2 gene

fibrillin 2

Normal Function

The *FBN2* gene provides instructions for making a large protein called fibrillin-2. This protein is transported out of cells into the extracellular matrix, which is an intricate lattice of proteins and other molecules that forms in the spaces between cells. In this matrix, fibrillin-2 binds to other proteins to form threadlike filaments called microfibrils.

Microfibrils become part of elastic fibers which enable the skin, ligaments, and blood vessels to stretch. Researchers have suggested that fibrillin-2 plays a role in directing the assembly of elastic fibers during embryonic development. Microfibrils also contribute to more rigid tissues that support the lens of the eye, nerves, and muscles. Additionally, microfibrils hold certain growth factors called transforming growth factor-beta (TGF-beta) proteins, which keeps them inactive. When released from microfibrils, TGF-beta growth factors are activated and affect the growth and repair of tissues throughout the body.

Health Conditions Related to Genetic Changes

Congenital contractural arachnodactyly

More than 20 mutations in the *FBN2* gene have been found to cause congenital contractural arachnodactyly. Most of these mutations change one protein building block (amino acid) in the fibrillin-2 protein, usually replacing the amino acid cysteine with a different amino acid. The substitution of another amino acid for cysteine can alter the structure or function of fibrillin-2. Most other *FBN2* mutations disrupt the way the *FBN2* gene's instructions are used to make the fibrillin-2 protein. All of these mutations reduce the amount of fibrillin-2 available to form microfibrils. Decreased microfibril formation probably weakens the elastic fibers and causes overactivation of TGF-beta growth factors, which leads to the signs and symptoms of congenital contractural arachnodactyly.

Other Names for This Gene

- CCA
- DA9
- FBN2_HUMAN
- fibrillin 2 (congenital contractural arachnodactyly)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FBN2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2201\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2201[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FBN2%5BTIAB%5D%29+OR+%28fibrillin+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

- FIBRILLIN 2; FBN2 (<https://omim.org/entry/612570>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2201>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FBN2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FBN2[gene]))

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Genomic Location

The *FBN2* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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