



EFEMP2 gene

EGF containing fibulin extracellular matrix protein 2

Normal Function

The *EFEMP2* gene provides instructions for making a protein called EGF-containing fibulin extracellular matrix protein 2, which is also known as fibulin-4. This protein is part of a group of proteins called fibulins. Fibulins have a variety of functions in the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells.

Little is known about the function of fibulin-4 in the extracellular matrix. It appears to play a critical role in the assembly of elastic fibers, which are slender bundles of protein that provide strength and flexibility to connective tissue (tissue that supports the body's joints and organs). Fibulin-4 is found in tissues and organs that are rich in elastic fibers, including the blood vessels, heart valves, lungs, and skin.

Health Conditions Related to Genetic Changes

Cutis laxa

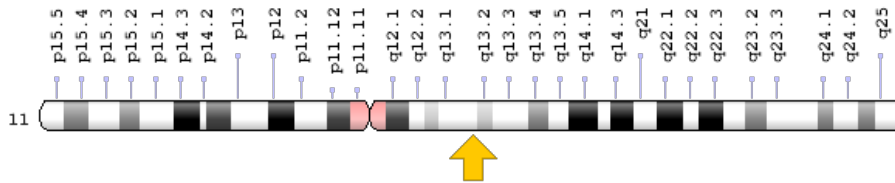
At least three mutations in the *EFEMP2* gene have been identified in people with cutis laxa. *EFEMP2* mutations cause a form of the disorder called autosomal recessive cutis laxa type I, which is characterized by loose and sagging skin, a lung disease called emphysema, and severe abnormalities involving the heart and blood vessels.

Mutations in the *EFEMP2* gene likely prevent cells from producing any functional fibulin-4. Without this protein, elastic fibers cannot be assembled normally in the extracellular matrix. A shortage of normal elastic fibers weakens connective tissue in the skin, blood vessels, lungs, and other organs. These defects in connective tissue underlie the major features of cutis laxa.

Chromosomal Location

Cytogenetic Location: 11q13.1, which is the long (q) arm of chromosome 11 at position 13.1

Molecular Location: base pairs 65,866,441 to 65,872,934 on chromosome 11 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EGF-containing fibulin-like extracellular matrix protein 2
- EGF containing fibulin like extracellular matrix protein 2
- EGF containing fibulin-like extracellular matrix protein 2
- FBLN4
- fibulin 4
- fibulin-like extracellular matrix protein
- MBP1
- mutant p53 binding protein 1
- UPH1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Fibulins
<https://www.ncbi.nlm.nih.gov/books/NBK6448/#A22215>

Clinical Information from GeneReviews

- EFEMP2-Related Cutis Laxa
<https://www.ncbi.nlm.nih.gov/books/NBK54467>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EFEMP2%5BTIAB%5D%29+OR+%28FBLN4%5BTIAB%5D%29+OR+%28fibulin-4%5BTIAB%5D%29+OR+%28MBP1%5BTIAB%5D%29%29+OR+%28fibulin+4%5BTIAB%5D%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

- EGF-CONTAINING FIBULIN-LIKE EXTRACELLULAR MATRIX PROTEIN 2
<http://omim.org/entry/604633>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_EFEMP2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=EFEMP2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3219
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:30008>
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