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 six variants (also known as mutations) in the *EFEMP2* gene have been identified in people with cutis laxa. *EFEMP2* variants cause a form of the disorder called autosomal recessive cutis laxa type 1B (ARCL1B), which is characterized by loose and sagging skin, a lung disease called emphysema, and severe abnormalities involving the heart and blood vessels.

Variants 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.

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

Tests Listed in the Genetic Testing Registry

- Tests of EFEMP2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=30008[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; EFEMP2 (https://omim.org/entry/604633)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/30008)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=EFEMP2[gene])

References

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Genomic Location

The *EFEMP2* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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