

LTBP4 gene

latent transforming growth factor beta binding protein 4

Normal Function

The *LTBP4* gene provides instructions for making a protein that has multiple functions in the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells. The LTBP4 protein controls the activity of a protein called transforming growth factor beta-1 (TGF β -1) by attaching it to the extracellular matrix. This growth factor is turned off (inactivated) when stored in the extracellular matrix and turned on (activated) when released. When turned on, the TGF β -1 protein triggers chemical signals that direct the functions of the body's cells during growth and development and that regulate the formation of the extracellular matrix.

The LTBP4 protein also plays a critical role in the formation of elastic fibers in the extracellular matrix. Elastic fibers are slender bundles of proteins that provide strength and flexibility to connective tissue, which supports organs and tissues such as the skin, lungs, heart, ligaments, and blood vessels.

Health Conditions Related to Genetic Changes

Cutis laxa

At least 20 variants (also known as mutations) in the *LTBP4* gene have been identified in people with cutis laxa. *LTBP4* gene variants cause a form of the disorder called autosomal recessive cutis laxa type 1C (ARCL1C) or Urban-Rifkin-Davis syndrome, which is characterized by loose and sagging skin, a lung disease called emphysema, and abnormalities involving the digestive and urinary systems.

Most of the *LTBP4* gene variants involved in cutis laxa change single DNA building blocks. These changes alter the blueprint for making proteins, and the abnormal blueprint is broken down in the cell before LTBP4 protein is made. A shortage of this protein prevents the assembly of elastic fibers, which weakens connective tissue in the skin, lungs, and other organs. These defects in connective tissue underlie the major features of ARCL1C.

Other Names for This Gene

- FLJ46318

- FLJ90018
- LTBP-4
- LTBP-4L

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LTBP4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8425\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8425[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=LTBP4>)

Catalog of Genes and Diseases from OMIM

- LATENT TRANSFORMING GROWTH FACTOR-BETA-BINDING PROTEIN 4; LTBP4 (<https://omim.org/entry/604710>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8425>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LTBP4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LTBP4[gene]))

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