FKBP14 gene

FKBP prolyl isomerase 14

Normal Function

The *FKBP14* gene provides instructions for making a protein called FKBP prolyl isomerase 14 (also known as FKBP22). This protein is found in a cell structure called the endoplasmic reticulum (ER), which is involved in protein processing and transport. Among its many functions, the endoplasmic reticulum folds and modifies newly formed proteins so they have the 3-dimensional shape they need to function properly. FKBP prolyl isomerase 14 is thought to assist with protein folding, particularly the folding of procollagens. Procollagens are the precursors of collagens, which are complex molecules found in the spaces between cells (the extracellular matrix) that add strength, support, and stretchiness (elasticity) to organs and tissues throughout the body. Studies suggest that FKBP prolyl isomerase 14 may also play a role in processing other components of the extracellular matrix.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

Mutations in the *FKBP14* gene are one cause of a rare form of Ehlers-Danlos syndrome called the kyphoscoliotic type (kEDS-FKBP14). Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues supporting the skin, bones, blood vessels, and many other organs and tissues. The kyphoscoliotic type is characterized by an unusually large range of joint movement (hypermobility); weak muscle tone (hypotonia); severe, progressive curvature of the spine (kyphoscoliosis) that can interfere with breathing; and fragile blood vessels that can tear (rupture), leading to internal bleeding. When the kyphoscoliotic type is caused by *FKBP14* gene mutations, affected individuals may also have muscle wasting (atrophy) and hearing loss that is present from birth.

At least four *FKBP14* gene mutations have been found to cause the kyphoscoliotic type of Ehlers-Danlos syndrome. These mutations, which affect both copies of the gene in each cell, abnormally copy (duplicate) or delete a small amount of DNA from the gene. The extra or missing genetic material prevents the gene from making functional FKBP prolyl isomerase 14. A loss of this protein disrupts the activities of the endoplasmic reticulum, including folding procollagens and processing other components of the extracellular matrix. As a result, the extracellular matrix becomes disorganized, which weakens connective tissues throughout the body and leads to the signs and symptoms

of the disorder.

Other Names for This Gene

- 22 kDa FK506-binding protein
- 22 kDa FKBP
- EDSKMH
- FK506 binding protein 14
- FK506 binding protein 14, 22 kDa
- FKBP-22
- FKBP22
- FLJ20731
- IPBP12
- peptidyl-prolyl cis-trans isomerase FKBP14 precursor
- PPIase FKBP14
- rotamase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FKBP14 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55033[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28FKBP14%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- FK506-BINDING PROTEIN 14; FKBP14 (https://omim.org/entry/614505)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55033)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FKBP14[gene])

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Genomic Location

The *FKBP14* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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