TNXB gene

tenascin XB

Normal Function

The *TNXB* gene provides instructions for making a protein called tenascin-X. This protein plays an important role in organizing and maintaining the structure of tissues that support the body's muscles, joints, organs, and skin (connective tissues). In particular, studies suggest that it helps to regulate the production and assembly of certain types of collagen. Collagens are a family of proteins that strengthen and support connective tissues throughout the body. Tenascin-X is also involved in regulating the structure and stability of elastic fibers, which provide flexibility and stretchiness (elasticity) to connective tissues.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

Mutations in the *TNXB* gene cause a very small percentage of all cases of a form of Ehlers-Danlos syndrome called the hypermobile type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. The hypermobile type is characterized by an unusually large range of joint movement (hypermobility). The mutations that cause this form of the disorder occur in one copy of the *TNXB* gene in each cell. These mutations reduce the amount of functional tenascin-X that cells produce, which decreases the ability of tenascin-X to interact with collagens and elastic fibers. These changes weaken connective tissues in many parts of the body, which results in the signs and symptoms of the hypermobile type of Ehlers-Danlos syndrome.

Some people with a condition called benign joint hypermobility syndrome (BJHS) also make a reduced amount of tenascin-X protein, although no *TNXB* gene mutations have been identified in these individuals. This condition causes hypermobility and chronic joint pain. The signs and symptoms of benign joint hypermobility syndrome overlap significantly with those of the hypermobile type of Ehlers-Danlos syndrome. Studies suggest that they may be forms of the same condition.

Some people with Ehlers-Danlos syndrome have mutations in two copies of the *TNXB* gene in each cell. These individuals have a form of the disorder that is often called the classical-like type. Its signs and symptoms are similar to the classical type of Ehlers-

Danlos syndrome, including hypermobility and skin that is soft, highly stretchy (elastic), and fragile. However, affected individuals do not have the unusual scarring that is characteristic of that type. Mutations that occur in both copies of the *TNXB* gene prevent production of any tenascin-X protein. A loss of this protein severely disrupts the organization of collagen fibrils and elastic fibers, which significantly weakens connective tissues.

Other Names for This Gene

- hexabrachion-like
- HXBL
- tenascin XB1
- tenascin XB2
- TENX
- TENX_HUMAN
- TNX
- TNXB1
- TNXB2
- TNXBS
- XB
- XBS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TNXB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7148[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TNXB%5BTIAB%5D%29+OR+%28tenascin+XB%5BTIAB%5D%29+OR+%28tenascin-X%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TENASCIN XB; TNXB (https://omim.org/entry/600985)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7148)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TNXB[gene])

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Genomic Location

The *TNXB* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

Last updated November 1, 2017