TGFB1 Gene

Subjects: Genetics
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Definition

transforming growth factor beta 1

1. Normal Function

The *TGFB1* gene provides instructions for producing a protein called transforming growth factor beta-1 (TGFβ-1). The TGFβ-1 protein triggers chemical signals that regulate various cell activities inside the cell, including the growth and division (proliferation) of cells, the maturation of cells to carry out specific functions (differentiation), cell movement (motility), and controlled cell death (apoptosis).

The TGFβ-1 protein is found throughout the body but is particularly abundant in tissues that make up the skeleton, where it helps regulate the formation and growth of bone and cartilage, a tough, flexible tissue that makes up much of the skeleton during early development. TGFβ-1 is also involved in the formation of blood vessels, development of muscle tissue and body fat, wound healing, inflammatory processes in the immune system, and prevention of tumor growth.

2. Health Conditions Related to Genetic Changes

2.1. Camurati-Engelmann disease

At least 12 mutations in the *TGFB1* gene have been found to cause Camurati-Engelmann disease. This condition is characterized by abnormally thick bones (hyperostosis) in the arms, legs, and skull. Hyperostosis can cause bone pain, muscle weakness, and increased pressure on the brain that results in neurological problems, including headaches and hearing and vision problems.

Most of the *TGFB1* gene mutations change single protein building blocks (amino acids) in the TGFβ-1 protein. Three mutations account for approximately 75 percent of cases of Camurati-Engelmann disease. Two of these mutations change the amino acid arginine at position 218 in the protein. One replaces arginine with the amino acid cysteine (written as Arg218Cys or R218C) and the other replaces arginine with the amino acid histidine (written as Arg218His R218H). The third mutation replaces the amino acid cysteine with the amino acid arginine at protein position 225 (written as Cys225Arg C225R).

All *TGFB1* gene mutations that cause Camurati-Engelmann disease result in the production of an overly active TGFβ-1 protein. This abnormal TGFβ-1 protein activity causes an increase in signal transduction, which leads to more bone formation. As a result, the bones in the arms, legs, and skull are thicker than normal, contributing to the movement and neurological problems often experienced by individuals with Camurati-Engelmann disease.

2.2. Cancers

Changes in the *TGFB1* gene have been reported to be associated with certain cancers. These variants are acquired during a person's lifetime and are present only in certain cells. Studies have shown that *TGFB1* gene variants are associated with breast, colorectal, lung, liver, and prostate cancer. The altered protein expression may increase several cell processes that promote cancer formation such as cell proliferation, cell motility, and the development of new blood vessels (angiogenesis) that nourish a growing tumor.

Idiopathic pulmonary fibrosis

3. Other Names for This Gene

- TGF-beta 1 protein
- TGF-beta-1
- TGFB
- TGFB1_HUMAN
- TGFbeta
- transforming growth factor, beta 1

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Keywords

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