TRIP11 Gene

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Thyroid hormone receptor interactor 11: The TRIP11 gene provides instructions for making a protein known as Golgi microtubule-associated protein 210 (GMAP-210).

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1. Normal Function

The *TRIP11* gene provides instructions for making a protein known as Golgi microtubule-associated protein 210 (GMAP-210). This protein is found in the Golgi apparatus, a cell structure in which newly produced proteins are modified so they can carry out their functions. Studies suggest that the GMAP-210 protein helps to maintain the structure of the Golgi apparatus, and it may also be involved in the transport of certain proteins out of cells.

Although the GMAP-210 protein is found throughout the body, researchers suspect that it may have a particularly important role in cells called chondrocytes in the developing skeleton. Chondrocytes give rise to cartilage, a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

2. Health Conditions Related to Genetic Changes

2.1. Achondrogenesis

At least nine mutations in the *TRIP11* gene have been found to cause a form of achondrogenesis known as type 1A or the Houston-Harris type. This rare disorder of bone development is characterized by extremely short limbs, a narrow chest, short ribs that fracture easily, and a lack of normal bone formation (ossification) in the skull, spine, and pelvis. Serious health problems result from these abnormalities, and infants with achondrogenesis usually die before or soon after birth.

The *TRIP11* gene mutations associated with achondrogenesis type 1A lead to the production of a nonfunctional version of the GMAP-210 protein or prevent the cell from producing any of this protein. Studies suggest that a shortage of GMAP-210 activity alters the structure and function of the Golgi apparatus, which impairs protein modification. Chondrocytes appear to be particularly sensitive to these changes, and malfunction of the Golgi apparatus in these cells likely underlies the problems with bone formation in achondrogenesis type 1A.

3. Other Names for This Gene

- ACG1A
- CEV14
- clonal evolution-related gene on chromosome 14 protein
- GMAP-210
- Golgi-associated microtubule-binding protein 210
- Golgi-microtubule-associated protein of 210 kDa
- thyroid receptor-interacting protein 11
- TR-interacting protein 11
- TRIP-11
- TRIP230
- TRIPB_HUMAN

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