COL6A2 Gene

Subjects: Genetics & Heredity
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collagen type VI alpha 2 chain

genes

1. Normal Function

The *COL6A2* gene provides instructions for making one component of type VI collagen, which is a flexible protein found in the space that surrounds cells. Specifically, the protein produced from the *COL6A2* gene is the alpha(α)2(VI) chain of type VI collagen. This chain combines with chains produced from other genes to produce a complete type VI collagen molecule.

Collagens are found in the extracellular matrix, which is an intricate lattice that forms in the space between cells and provides structural support. Type VI collagen is located in the extracellular matrix surrounding cells that make up the muscles used for movement (skeletal muscle cells) and cells that make up connective tissue, which provides strength and flexibility to structures throughout the body, including skin and joints. The extracellular matrix is necessary for cell stability and growth. Research suggests that type VI collagen links basement membranes, which are thin, sheet-like structures that are part of the extracellular matrix, to nearby cells.

2. Health Conditions Related to Genetic Changes

2.1. Collagen VI-Related Myopathy

Mutations in the *COL6A2* gene have been found to cause some cases of collagen VI-related myopathy, which is a group of disorders that vary in severity but generally result in muscle weakness and joint deformities called contractures. These mutations often change single protein building blocks (amino acids) in the α2(VI) chain. The most frequently affected amino acid is glycine; changes to this building block disrupt the structure of the α2(VI) chain. Other mutations can also disrupt the structure of the α2(VI) chain.

Mutations in the *COL6A2* gene affect type VI collagen in different ways. Some mutations lead to altered α2(VI) chains that can be incorporated into the mature type VI collagen molecule, although they disrupt the molecule's structure and function. Other mutations result in an altered chain that cannot be incorporated at all. Still other mutations prevent the production of any functional α2(VI) chain, which impedes formation of type VI collagen. All of these *COL6A2* gene mutations lead to a reduction or absence of functional collagen VI molecules. While it is

difficult to predict the severity of collagen VI-related myopathy based on the type of mutation, in general, lower amounts of type VI collagen lead to more severe signs and symptoms that begin earlier in life.

Changes in α2(VI) chain structure or production lead to an unstable extracellular matrix that is no longer attached to cells through the basement membrane. As a result, the stability of muscle cells and connective tissue progressively declines, which leads to the muscle weakness, contractures, and other signs and symptoms of collagen VI-related myopathy.

2.2. Limb-Girdle Muscular Dystrophy

Limb-girdle muscular dystrophy

3. Other Names for This Gene

- CO6A2_HUMAN
- collagen alpha-2(VI) chain
- collagen type VI alpha 2
- collagen VI, alpha-2 polypeptide
- collagen, type VI, alpha 2

References

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