COL17A1 Gene

Subjects: Genetics & Heredity
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collagen type XVII alpha 1 chain

genes

1. Normal Function

The *COL17A1* gene provides instructions for making a protein that is used to assemble type XVII collagen. Collagens are a family of proteins that strengthen and support connective tissues, such as skin, bone, tendons, and ligaments, throughout the body. In particular, type XVII collagen plays an essential role in strengthening and stabilizing the skin.

The protein produced from the *COL17A1* gene is known as a pro-α1(XVII) chain. Three identical pro-α1(XVII) chains twist together to form a triple-stranded, ropelike molecule known as a procollagen. Procollagen molecules are released from the cell and processed by enzymes to remove extra protein segments from the ends. Once these molecules are processed, they arrange themselves into long, thin bundles of mature type XVII collagen.

Type XVII collagen is a major component of hemidesmosomes, which are microscopic structures on the inner surface of the top layer of skin (the epidermis). These structures help to anchor the epidermis to underlying layers of skin. Type XVII collagen is critical for the stability of hemidesmosomes, and therefore it plays an important role in holding the layers of skin together.

2. Health Conditions Related to Genetic Changes

Junctional Epidermolysis Bullosa

More than 100 mutations in the *COL17A1* gene have been identified in people with junctional epidermolysis bullosa (JEB). Most of these mutations add or remove several DNA building blocks (nucleotides) in the *COL17A1* gene or create a premature stop signal in the instructions for making the pro-α1(XVII) chain. These changes reduce the amount of functional type XVII collagen in the skin. Without enough of this collagen, the epidermis is only weakly attached to underlying layers of skin. Friction or other minor trauma (such as rubbing or scratching) can cause the skin layers to separate, leading to the formation of blisters.

Most *COL17A1* gene mutations cause the milder form of junctional epidermolysis bullosa, known as JEB generalized intermediate. Affected individuals experience blistering, but it may be limited to the hands, feet, knees, and elbows and often improves after the newborn period. A few individuals with mutations in the *COL17A1* gene have had the more serious form of the disorder, JEB generalized severe. Infants with JEB generalized severe develop widespread blistering that causes life-threatening complications.

3. Other Names for This Gene

- alpha 1 type XVII collagen
- BA16H23.2
- BP180
- BPAG2
- bullous pemphigoid antigen 2 (180kD)
- COHA1_HUMAN
- collagen type XVII alpha 1
- collagen XVII, alpha-1 polypeptide
- collagen, type XVII, alpha 1
- FLJ60881
- KIAA0204
- LAD-1

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