KRT17 Gene

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Keratin 17

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1. Introduction

The *KRT17* gene provides instructions for making a protein called keratin 17 or K17. Keratins are a group of tough, fibrous proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, nails, and similar tissues. Keratin 17 is produced in the nails, the hair follicles, and the skin on the palms of the hands and soles of the feet. It is also found in the skin's sebaceous glands, which produce an oily substance called sebum that normally lubricates the skin and hair.

Keratin 17 partners with a similar protein, keratin 6b, to form molecules called keratin intermediate filaments. These filaments assemble into dense networks that provide strength and resilience to the skin, nails, and other tissues. Networks of keratin intermediate filaments protect these tissues from being damaged by friction and other everyday physical stresses. Keratin 17 is also among several keratins involved in wound healing.

2. Health Conditions Related to Genetic Changes

2.1. Pachyonychia Congenita

More than 20 mutations in the *KRT17* gene have been identified in people with pachyonychia congenita, a rare condition that primarily affects the nails and skin. In most cases, this condition becomes apparent within the first few months of life. Most of the *KRT17* gene mutations associated with pachyonychia congenita change single protein building blocks (amino acids) in keratin 17.

The *KRT17* gene mutations responsible for pachyonychia congenita change the structure of keratin 17, preventing it from interacting effectively with keratin 6b and interfering with the assembly of the keratin intermediate filament network. Without this network, skin cells become fragile and are easily damaged, making the skin less resistant to friction and minor trauma. Even normal activities such as walking can cause skin cells to break down, resulting in the formation of painful blisters and calluses. In the sebaceous glands, abnormal keratin filaments lead to the development of sebum-filled cysts called steatocystomas. Defective keratin 17 also disrupts the growth and function of other tissues, such as the hair follicles and nails, which explains why the signs and symptoms of pachyonychia congenita can also affect these other parts of the body.

2. Steatocystoma Multiplex

At least four mutations in the *KRT17* gene have been found to cause a skin disorder called hereditary steatocystoma multiplex. Researchers suggest that this condition may be a mild form (variant) of pachyonychia congenita (described above). Like pachyonychia congenita, hereditary steatocystoma multiplex involves the development of multiple sebaceous gland cysts called steatocystomas. Most people with hereditary steatocystoma multiplex do not have the other features of pachyonychia congenita, although mild nail and dental abnormalities are possible.

The *KRT17* gene mutations that cause hereditary steatocystoma multiplex change single amino acids in the keratin 17 protein, which interferes with the assembly of the keratin intermediate filament network. In the sebaceous glands, these keratin abnormalities lead to the development of steatocystomas. It is unclear why these sebum-containing cysts are typically the only feature of this disorder.

3. Other Names for This Gene

- CK-17
- cytokeratin-17
- K17
- K1C17_HUMAN
- keratin 17, type I
- keratin, type I cytoskeletal 17
- keratin-17

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