KRT6B Gene

Subjects: Genetics & Heredity

Contributor: Dean Liu

Keratin 6B

genes

1. Introduction

The *KRT6B* gene provides instructions for makalms 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.

Keratin 6b partners with a similar protein, keratin 17, to form molecules called keratin intermediate filaments. These filaments assemble into dense networks that provide strengthing a protein called keratin 6b or K6b. Keratins are a group of tough, fibrous proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, and nails. Keratin 6b is produced in the nails, the hair follicles, and the skin on the p 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 6b is also among several keratins involved in wound healing.

2. Health Conditions Related to Genetic Changes

2.1. Pachyonychia Congenita

At least four mutations in the *KRT6B* 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. These mutations either change single protein building blocks (amino acids) in keratin 6b or delete a small number of amino acids from the protein.

The *KRT6B* gene mutations responsible for pachyonychia congenita change the structure of keratin 6b, preventing it from interacting effectively with keratin 17 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 6b 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.

3. Other Names for This Gene

- CK 6B
- CK6B
- cytokeratin 6B
- cytokeratin-6B
- K2C6B_HUMAN
- K6B
- K6b keratin
- keratin 6B, type II
- keratin, epidermal, type II, K6B
- keratin, type II cytoskeletal 6B
- KRTL1

References

1. McLean WH, Hansen CD, Eliason MJ, Smith FJ. The phenotypic and moleculargenetic features of pachyonychia congenita. J Invest Dermatol. 2011May;131(5):1015-7. doi: 10.1038/jid.2011.59.
2. Sharma VM, Stein SL. A novel mutation in K6b in pachyonychia congenita type 2.J Invest Dermatol. 2007 Aug;127(8):2060-2.
3. Smith FJ, Jonkman MF, van Goor H, Coleman CM, Covello SP, Uitto J, McLean WH. A mutation in human keratin K6b produces a phenocopy of the K17 disorderpachyonychia congenita type 2. Hum Mol Genet. 1998 Jul;7(7):1143-8.
4. Wilson NJ, O'Toole EA, Milstone LM, Hansen CD, Shepherd AA, Al-Asadi E,Schwartz ME, McLean WH, Sprecher E, Smith FJ. The molecular genetic analysis ofthe expanding

pachyonychia congenita case collection. Br J Dermatol. 2014Aug;171(2):343-55. doi: 10.1111/bjd.12958.