

TCHH Gene

Subjects: Genetics & Heredity

Contributor: Rui Liu

Trichohyalin: The TCHH gene provides instructions for making a protein called trichohyalin. This protein is primarily found in hair follicles, which are specialized structures in the skin where hair growth occurs.

Keywords: genes

1. Normal Function

The *TCHH* gene provides instructions for making a protein called trichohyalin. This protein is primarily found in hair follicles, which are specialized structures in the skin where hair growth occurs. Trichohyalin can also be found in the hair strand (shaft). Once trichohyalin is produced, it is modified by other proteins so that it can attach (bind) to other trichohyalin proteins and to molecules called keratin intermediate filaments to create organized cross-links. These links form dense networks that give the hair shaft its cylindrical shape.

2. Health Conditions Related to Genetic Changes

2.1. Uncombable hair syndrome

At least one mutation in the *TCHH* gene has been found to cause uncombable hair syndrome. This condition is characterized by dry, frizzy, blond scalp hair that cannot be combed flat. This condition usually improves over time, and by adolescence individuals with uncombable hair syndrome have hair that lies flat and has normal or nearly normal texture.

The *TCHH* gene mutation that has been identified leads to a premature stop signal in the instructions used to make trichohyalin, resulting in an abnormally short protein with severely reduced activity. A shortage (deficiency) of functional protein reduces the cross-links that are formed between trichohyalin proteins and keratin intermediate filaments. As a result, the cross-section of the hair shaft becomes triangular, heart-shaped, or flat. These angular hair shafts result in frizzy hair that will not lie flat, which is typical of uncombable hair syndrome.

3. Other Names for This Gene

- AHF
 - THH
 - THL
 - TRHY
-

References

1. Calderon P, Otberg N, Shapiro J. Uncombable hair syndrome. *J Am Acad Dermatol*. 2009 Sep;61(3):512-5. doi: 10.1016/j.jaad.2009.01.006.
2. Takase T, Hirai Y. Identification of the C-terminal tail domain of AHF/trichohyalin as the critical site for modulation of the keratin filamentous meshwork in the keratinocyte. *J Dermatol Sci*. 2012 Feb;65(2):141-8. doi:10.1016/j.jdermsci.2011.12.014.
3. Ü Basmanav FB, Cau L, Tafazzoli A, Méchin MC, Wolf S, Romano MT, Valentin F, Wiegmann H, Huchénq A, Kandil R, Garcia Bartels N, Kilic A, George S, Ralser DJ, Bergner S, Ferguson DJP, Oprisoreanu AM, Wehner M, Thiele H, Altmüller J, Nürnberg P, Swan D, Houniet D, Büchner A, Weibel L, Wagner N, Grimalt R, Bygum A, Serre G, Blume-Peytavi U, Sprecher E, Schoch S, Oji V, Hamm H, Farrant P, Simon M, Betz RC. Mutations in Three Genes Encoding Proteins Involved in Hair Shaft Formation Cause Uncombable Hair Syndrome. *Am J Hum Genet*. 2016 Dec 1;99(6):1292-1304. doi: 10.1016/j.ajhg.2016.10.004.

