

TGM1 gene

transglutaminase 1

Normal Function

The *TGM1* gene provides instructions for making an enzyme called transglutaminase 1. This enzyme is found in cells that make up the outermost layer of the skin (the epidermis). Transglutaminase 1 is involved in the formation of the cornified cell envelope, which is a structure that surrounds skin cells and helps form a protective barrier between the body and its environment. Specifically, transglutaminase 1 forms strong bonds, called cross-links, between the structural proteins that make up the cornified cell envelope. This cross-linking provides strength and stability to the epidermis.

Health Conditions Related to Genetic Changes

Lamellar ichthyosis

Many mutations in the *TGM1* gene have been found to cause lamellar ichthyosis, which is a condition that causes scaly skin that covers much of the body, and other skin abnormalities. Some *TGM1* gene mutations that cause this condition change single DNA building blocks (nucleotides) in the transglutaminase 1 enzyme. The most frequently occurring mutation (written as 877-2A>G) affects the way the gene's instructions are pieced together to form the enzyme and results in an abnormally shortened, nonfunctional enzyme. Other *TGM1* gene mutations result in a transglutaminase 1 enzyme that cannot function normally, is abnormally short, or is not produced. A lack of functional transglutaminase 1 prevents the formation of the cornified cell envelope, causing the skin abnormalities of lamellar ichthyosis.

Other disorders

In addition to lamellar ichthyosis (described above), *TGM1* gene mutations have been found to cause other forms of ichthyosis. In one type, called self-healing collodion baby, affected infants are born with a tight, clear sheath covering their skin called a collodion membrane. This membrane usually dries and peels off during the first few weeks of life, and affected infants often show near normal skin within a few months. Another type of ichthyosis, called bathing suit ichthyosis, is characterized by scaly skin that is limited to the trunk.

Other Names for This Gene

- epidermal TGase
- ICR2
- protein-glutamine gamma-glutamyltransferase K
- TGASE
- TGase K
- TGase-1
- TGK
- TGM1_HUMAN
- transglutaminase 1 (K polypeptide epidermal type I, protein-glutamine-gamma-glutamyltransferase)
- transglutaminase K
- transglutaminase, keratinocyte
- transglutaminase-1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TGM1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7051\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7051[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TGM1%5BTIAB%5D%29+OR+%28transglutaminase+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- TRANSGLUTAMINASE 1; TGM1 (<https://omim.org/entry/190195>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7051>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TGM1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TGM1[gene]))

References

- Farasat S, Wei MH, Herman M, Liewehr DJ, Steinberg SM, Bale SJ, Fleckman P, Toro JR. Novel transglutaminase-1 mutations and genotype-phenotype investigation of 104 patients with autosomal recessive congenital ichthyosis in the USA. *J Med Genet*. 2009 Feb;46(2):103-11. doi: 10.1136/jmg.2008.060905. Epub 2008 Oct 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18948357>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3044481/>)
- Herman ML, Farasat S, Steinbach PJ, Wei MH, Toure O, Fleckman P, Blake P, Bale SJ, Toro JR. Transglutaminase-1 gene mutations in autosomal recessive congenital ichthyosis: summary of mutations (including 23 novel) and modeling of TGase-1. *Hum Mutat*. 2009 Apr;30(4):537-47. doi: 10.1002/humu.20952. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19241467>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3243309/>)
- Israeli S, Goldberg I, Fuchs-Telem D, Bergman R, Indelman M, Bitterman-Deutsch O, Harel A, Mashiach Y, Sarig O, Sprecher E. Non-syndromic autosomal recessive congenital ichthyosis in the Israeli population. *Clin Exp Dermatol*. 2013 Dec;38(8):911-6. doi: 10.1111/ced.12148. Epub 2013 Apr 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23621129>)
- Oji V, Tadini G, Akiyama M, Blanchet Bardon C, Bodemer C, Bourrat E, Coudiere P, DiGiovanna JJ, Elias P, Fischer J, Fleckman P, Gina M, Harper J, Hashimoto T, Hausser I, Hennies HC, Hohl D, Hovnanian A, Ishida-Yamamoto A, Jacyk WK, Leachman S, Leigh I, Mazereeuw-Hautier J, Milstone L, Morice-Picard F, Paller AS, Richard G, Schmuth M, Shimizu H, Sprecher E, Van Steensel M, Taieb A, Toro JR, Vabres P, Vahlquist A, Williams M, Traupe H. Revised nomenclature and classification of inherited ichthyoses: results of the First Ichthyosis Consensus Conference in Soreze 2009. *J Am Acad Dermatol*. 2010 Oct;63(4):607-41. doi:10.1016/j.jaad.2009.11.020. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20643494>)
- Rodriguez-Pazos L, Ginarte M, Vega A, Toribio J. Autosomal recessive congenital ichthyosis. *Actas Dermosifiliogr*. 2013 May;104(4):270-84. doi:10.1016/j.adengl.2011.11.021. Epub 2013 Apr 3. English, Spanish. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23562412>)
- Terrinoni A, Serra V, Codispoti A, Talamonti E, Bui L, Palombo R, Sette M, Campione E, Didona B, Annicchiarico-Petruzzelli M, Zambruno G, Melino G, Candi E. Novel transglutaminase 1 mutations in patients affected by lamellar ichthyosis. *Cell Death Dis*. 2012 Oct 25;3(10):e416. doi: 10.1038/cddis.2012.152. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23096117>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3481139/>)

Genomic Location

The *TGM1* gene is found on chromosome 14 (<https://medlineplus.gov/genetics/chromosome/14/>).

Last updated March 1, 2015