# TGM1 Gene

Subjects: Genetics
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## Definition

Transglutaminase 1: The TGM1 gene provides instructions for making an enzyme called transglutaminase 1.

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## 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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.

### 2.2. 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.

## 3. 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

## References


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## Keywords

genes