

LAMB3 Gene

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Laminin subunit beta 3

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1. Introduction

The *LAMB3* gene provides instructions for making one part (subunit) of a protein called laminin 332 (formerly known as laminin 5). This protein is made up of three subunits, called alpha, beta, and gamma. The *LAMB3* gene carries instructions for the beta subunit; the alpha and gamma subunits are produced from other genes.

Laminins are a group of proteins that regulate cell growth, cell movement (motility), and the attachment of cells to one another (adhesion). They are also involved in the formation and organization of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. Laminin 332 has a particularly important role in the basement membrane that underlies the top layer of skin (the epidermis). This membrane gives strength and resiliency to the skin and creates an additional barrier between the body and its surrounding environment. Laminin 332 is a major component of fibers called anchoring filaments, which connect the two layers of the basement membrane and help hold the skin together.

Studies suggest that laminin 332 also has several other functions. This protein appears to be important for wound healing. Additionally, researchers have proposed roles for laminin 332 in the clear outer covering of the eye (the cornea) and in the development of tooth enamel.

2. Health Conditions Related to Genetic Changes

2.1. Junctional Epidermolysis Bullosa

More than 100 mutations in the *LAMB3* gene have been identified in people with junctional epidermolysis bullosa (JEB). The more serious form of the disease, known as JEB generalized severe, usually results from mutations that prevent the production of functional laminin 332. Most of these mutations lead to a premature stop signal in the instructions for making the beta subunit of laminin 332, which prevents the assembly of this protein. Without laminin 332, the epidermis is only weakly connected to the underlying layers of skin. Friction or other minor trauma (such as rubbing or scratching) can cause the skin layers to separate, leading to the formation of blisters. Infants with JEB generalized severe develop widespread blistering that causes life-threatening complications.

Other *LAMB3* gene mutations cause the milder form of junctional epidermolysis bullosa, JEB generalized intermediate. Some of these mutations alter single protein building blocks (amino acids) in the beta subunit of laminin 332. Others add or delete a small number of amino acids in the beta subunit or change the way the gene's instructions are used to make the subunit. The genetic changes responsible for JEB generalized intermediate usually lead to the production of a laminin 332 protein that retains some of its function. Affected individuals experience blistering, but it may be limited to the hands, feet, knees, and elbows and often improves after the newborn period.

3. Other Names for This Gene

- BM600-125KDA
- FLJ99565
- kalinin B1 chain
- kalinin-140kDa

- LAM5, beta-3 subunit
- LAMB3_HUMAN
- laminin 5, beta-3 subunit
- laminin B1k chain
- laminin B3
- laminin S B3 chain
- laminin, beta 3
- laminin, beta 3 precursor
- laminin, beta-3
- LAMNB1
- nicein-125kDa

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