FERMT1 Gene

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Fermitin family member 1

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1. Normal Function

The *FERMT1* gene provides instructions for making a protein called kindlin-1. This protein is found in epithelial cells, which are the cells that line the surfaces and cavities of the body. In the skin, kindlin-1 plays a critical role in specialized cells called keratinocytes, which are the major component of the outer layer of the skin (the epidermis).

Kindlin-1 is part of cell structures called focal adhesions. These structures contain many different kinds of proteins, which are involved in linking the cell's internal framework (the cytoskeleton) to the intricate lattice of proteins and other molecules that surrounds cells (the extracellular matrix). This linking is known as cell-matrix adhesion. Kindlin-1 attaches (binds) to and turns on (activates) proteins called integrins, which directly connect the cytoskeleton with the extracellular matrix and help transmit chemical signals into the cell.

As part of focal adhesions, Kindlin-1 is involved in several important cell functions, including cell growth and division (proliferation) and the movement (migration) of cells.

2. Health Conditions Related to Genetic Changes

2.1 Kindler Syndrome

More than 70 mutations in the *FERMT1* gene have been identified in people with Kindler syndrome. This disorder is a rare type of epidermolysis bullosa, which is a group of genetic conditions that cause the skin to be very fragile and to blister easily. Kindler syndrome also affects the moist lining (mucosae) of the mouth, eyes, esophagus, intestines, genitals, and urinary system, causing these tissues to be very fragile. In addition, people with Kindler syndrome have an increased risk of developing a form of cancer called squamous cell carcinoma.

Most mutations in the *FERMT1* gene prevent the production of any functional kindlin-1. A lack of this protein disrupts many essential cell functions. For example, keratinocytes without kindlin-1 have an abnormal structure and cannot grow or divide normally. They are also less able to attach the epidermis to the underlying layer of skin (the dermis). These changes make the skin fragile and prone to blistering. Similarly, a lack of kindlin-1 in epithelial cells of the mucosae causes damage that makes these tissues extremely fragile. It is unclear how a shortage of kindlin-1 is related to squamous cell carcinoma in people with Kindler syndrome.

3. Other Names for This Gene

- C20orf42
- DTGCU2
- fermitin family homolog 1
- FLJ20116
- KIND1
- kindlerin
- kindlin 1

- kindlin syndrome protein
- unc-112-related protein 1
- UNC112 related protein 1
- UNC112A
- URP1

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