ATP2C1 Gene

Subjects: Genetics
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Definition

ATPase secretory pathway Ca2+ transporting 1

1. Normal Function

The *ATP2C1* gene provides instructions for making a protein called hSPCA1. This protein is an adenosine triphosphate (ATP)-powered calcium pump, which uses energy from ATP molecules to pump charged calcium atoms (calcium ions) across cell membranes. Specifically, the hSPCA1 protein transports calcium ions into a cell structure called the Golgi apparatus, where they are stored until needed. The appropriate storage and release of calcium is essential for many cell activities, including cell growth and division (proliferation), cell movement (migration), and attachment of cells to one another (cell adhesion).

The hSPCA1 protein also transports manganese ions into the Golgi apparatus. Manganese works with a variety of enzymes and is involved in processing newly formed proteins.

The hSPCA1 protein is present in cells throughout the body. It appears to be particularly important for the normal function of cells called keratinocytes, which are found in the outer layer of the skin (the epidermis). In addition to proliferation and adhesion, calcium regulation in these cells appears to play an important role in maintaining the skin's barrier function, helping to keep foreign invaders such as bacteria out of the body.

2. Health Conditions Related to Genetic Changes

Hailey-Hailey disease

More than 200 mutations in the *ATP2C1* gene have been found to cause Hailey-Hailey disease, a rare skin condition characterized by red, raw, and blistered areas of skin that can become infected. Mutations in this gene reduce the amount of functional hSPCA1 protein, which impairs the storage of calcium ions in the Golgi apparatus. For unknown reasons, this abnormal calcium storage affects keratinocytes more than other types of cells. Problems with calcium regulation impair many cell functions, including cell adhesion. As a result, keratinocytes do not stick tightly to one another, which causes the epidermis to become fragile and less resistant to minor trauma. Because the skin is easily damaged, it develops raw, blistered areas, particularly in skin folds where there is moisture and friction. In addition, abnormal calcium regulation disrupts the barrier function of the skin, making it more susceptible to infections. However, it is unclear how a reduction of hSPCA1 protein function affects the skin barrier, and how its impairment is involved in Hailey-Hailey disease.

Although *ATP2C1* gene mutations probably also affect the transport of manganese within cells, abnormal manganese regulation is not thought to contribute to the signs and symptoms of Hailey-Hailey disease.

3. Other Names for This Gene

- AT2C1_HUMAN
- ATP-dependent Ca(2+) pump PMR1
- ATP2C1A
- ATPase 2C1
- ATPase, Ca(2+)-sequestering
- ATPase, Ca++ transporting, type 2C, member 1
- BCPM

- calcium-transporting ATPase type 2C member 1
- HHD
- hSPCA1
- HUSSY-28
- KIAA1347
- PMR1
- secretory pathway Ca2+/Mn2+ ATPase 1
- SPCA1

References

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Keywords

genes