ITGA6 Gene

Subjects: Genetics & Heredity
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Integrin subunit alpha 6

genes

1. Introduction

The *ITGA6* gene provides instructions for making one part (the α6 subunit) of two proteins known as α6β4 integrin and α6β1 integrin. Integrins are a group of proteins that regulate the attachment of cells to one another (cell-cell adhesion) and to the surrounding network of proteins and other molecules (cell-matrix adhesion). Integrins also transmit chemical signals that regulate cell growth and the activity of certain genes.

The α6β4 integrin protein is found primarily in epithelial cells, which are cells that line the surfaces and cavities of the body. This protein plays a particularly important role in strengthening and stabilizing the skin. It is a component of hemidesmosomes, which are microscopic structures that anchor the outer layer of the skin (the epidermis) to underlying layers. As part of a complex network of proteins in hemidesmosomes, α6β4 integrin helps to hold the layers of skin together.

The other integrin made with the α6 subunit, α6β1 integrin, functions during the formation of organs and tissues before birth. The α6β1 integrin protein has not been as well studied as α6β4 integrin.

2. Health Conditions Related to Genetic Changes

2.1. Epidermolysis Bullosa with Pyloric Atresia

At least five mutations in the *ITGA6* gene have been found to cause epidermolysis bullosa with pyloric atresia (EB-PA). In addition to skin blistering, people with EB-PA are born with a life-threatening obstruction of the digestive tract called pyloric atresia. Mutations in the *ITGA6* gene account for about 5 percent of all cases of EB-PA.

The *ITGA6* gene mutations responsible for EB-PA lead to a loss of functional α6β4 integrin. These mutations alter the normal structure and function of the α6 integrin subunit or prevent cells from producing enough of this subunit. The resulting shortage of functional α6β4 integrin causes cells in the epidermis to be fragile and easily damaged. Friction or other minor trauma can cause the skin layers to separate, leading to the widespread formation of blisters. It is less clear how mutations in the *ITGA6* gene are related to pyloric atresia.

2.2. Cancers

Researchers believe that both α6β1 integrin and α6β4 integrin may play critical roles in the progression of cancerous tumors called carcinomas. These cancers arise in epithelial cells and can affect many tissues and organs, including the breast, lung, liver, prostate, and skin.

Changes in the location and activity of α6β1 integrin and α6β4 integrin within cancer cells are associated with the progression of carcinomas. The integrin proteins activate key signaling molecules, which trigger cancer cells to migrate through the body and invade other tissues. These signals also make cancer cells more resistant to self-destruction (apoptosis).

Recent studies suggest that, in addition to their roles in the progression of existing carcinomas, α6β1 integrin and α6β4 integrin may be involved in the initial formation of these tumors.

3. Other Names for This Gene

- CD49f
- CD49f Antigens
- Cluster of differentiation antigen 49f
- FLJ18737
- integrin alpha 6
- integrin alpha chain, alpha 6
- Integrin alpha6
- integrin, alpha 6
- integrin, alpha-6
- ITA6_HUMAN
- Lymphocyte antigen CD49F
- VLA-6

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