ITGA6 gene
integrin subunit alpha 6

Normal Function

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.

Health Conditions Related to Genetic Changes

**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.

**Prostate cancer**
Researchers believe that both $\alpha_6\beta_1$ integrin and $\alpha_6\beta_4$ 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 $\alpha_6\beta_1$ integrin and $\alpha_6\beta_4$ 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, $\alpha_6\beta_1$ integrin and $\alpha_6\beta_4$ integrin may be involved in the initial formation of these tumors.

**Chromosomal Location**

Cytogenetic Location: 2q31.1, which is the long (q) arm of chromosome 2 at position 31.1

Molecular Location: base pairs 172,427,336 to 172,506,459 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

**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

Additional Information & Resources

Educational Resources
• Madame Curie Bioscience Database: Integrins in Cancer Cell Invasion
  https://www.ncbi.nlm.nih.gov/books/NBK6070/
• Madame Curie Bioscience Database: Integrins: An Overview of Structural and
  Functional Aspects
  https://www.ncbi.nlm.nih.gov/books/NBK6259/
• Molecular Biology of the Cell (fourth edition, 2002): Integrins
  https://www.ncbi.nlm.nih.gov/books/NBK26867/

Clinical Information from GeneReviews
• Epidermolysis Bullosa with Pyloric Atresia
  https://www.ncbi.nlm.nih.gov/books/NBK1157

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ITGA6%5BTIAB%5D%29+OR+%28integrin+%5Bti%5D+AND+alpha+6+%5Btiab%5D%29+OR+%28integrin+%5Bti%5D+AND+alpha6+%5Btiab%5D%29%29+AND+%28%28Genes%5BMH%5D+OR+Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• INTEGRIN, ALPHA-6
  http://omim.org/entry/147556

Research Resources
• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ITGA6ID41007ch2q31.html
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ITGA6%5Bgene%5D
• HGNC Gene Symbol Report
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14675179

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11251584

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15179031

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15256436

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9804362

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