ITGB2 Gene

Subjects: Genetics & Heredity
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Integrin subunit beta 2

genes

1. Introduction

The *ITGB2* gene provides instructions for making one part (the β2 subunit) of at least four different proteins known as β2 integrins. The other subunit can be one of a variety of alpha (α) subunits that are produced from different genes. 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 signals that regulate cell growth and the activity of certain genes.

Integrins that contain the β2 subunit are found embedded in the membrane that surrounds white blood cells (leukocytes). β2 integrins help leukocytes gather at sites of infection or injury, where they are needed to contribute to the immune response. β2 integrins recognize signs of inflammation and attach (bind) to proteins called ligands on the lining of blood vessels. This binding leads to linkage (adhesion) of the leukocyte to the blood vessel wall. Signaling through the β2 integrins triggers the transport of the attached leukocyte across the blood vessel wall to the site of infection or injury.

2. Health Conditions Related to Genetic Changes

2.1. Leukocyte Adhesion Deficiency Type 1

At least 90 mutations in the *ITGB2* gene have been found to cause leukocyte adhesion deficiency type 1. This condition is characterized by severe, recurrent infections and delayed healing after injury. Many of the mutations that cause this condition change single protein building blocks (amino acids) in the β2 subunit and lead to the production of a β2 subunit that cannot bind with other subunits to form integrins. Rarely, defective β2 subunits are able to form integrins, but the integrins cannot bind ligands. Leukocytes that lack these integrins cannot attach to the blood vessel wall or cross the vessel wall to contribute to the immune response. As a result, there is a decreased response to injury and foreign invaders, such as bacteria and fungi, resulting in frequent infections, delayed wound healing, and other signs and symptoms of leukocyte adhesion deficiency type 1.

3. Other Names for This Gene

- CD11b/ CD18
- CD11c/CD18
- CD11d/CD18
- CD18
- complement receptor C3 beta-subunit
- complement receptor C3 subunit beta
- CR3
- integrin beta 2
- integrin beta chain, beta 2
- integrin beta-2
- integrin, beta 2 (complement component 3 receptor 3 and 4 subunit)
- LFA-1 (αLβ2)
- Mac-1 (αMβ2)
- p150/95 (αXβ2)
- αDβ2

References

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