IL23R Gene

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Interleukin 23 receptor

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1. Introduction

The *IL23R* gene provides instructions for making a protein called the interleukin 23 (IL-23) receptor. This protein is embedded in the outer membrane of several types of immune system cells, including T cells, natural killer (NK) cells, monocytes, and dendritic cells. These cells identify foreign substances and defend the body against infection and disease.

At the cell surface, the IL-23 receptor interacts with a protein called IL-23. These two proteins fit together like a lock and key. IL-23 is a cytokine, which is a type of protein that regulates the activity of immune cells. When IL-23 binds to its receptor, it triggers a series of chemical signals inside the cell. These signals promote inflammation and help coordinate the immune system's response to foreign invaders such as bacteria and viruses.

2. Health Conditions Related to Genetic Changes

2.1. Ankylosing Spondylitis

Several variations (polymorphisms) in the *IL23R* gene have been found to influence the risk of ankylosing spondylitis. One of these variations appears to reduce the likelihood of developing this disorder. This genetic change alters a single protein building block (amino acid) in the IL-23 receptor, replacing the amino acid arginine with the amino acid glutamine at protein position 381 (written as Arg381Gln or R381Q). Other *IL23R* variations appear to increase the risk of developing ankylosing spondylitis. It is not clear how these changes are related to a person's risk of developing this disorder, but studies suggest that the effects of *IL23R* variations are likely related to the IL-23 receptor's role in inflammation. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing ankylosing spondylitis.

2.2. Crohn Disease

Several variations in or near the *IL23R* gene have been found to influence the risk of developing Crohn disease. These associations have been found primarily in people of northern European ancestry. For example, Arg381Gln, which is a protective factor for ankylosing spondylitis (described above), also appears to reduce the risk of developing Crohn disease. Although it is unclear how this change protects against Crohn disease, researchers believe that the receptor's role in triggering inflammation in the intestinal walls may underlie its connection with this disorder.

2.3. Other Disorders

Variations in the *IL23R* gene have also been associated with the risk of several other immune system-related conditions, including a skin disorder called psoriasis. People with this chronic inflammatory condition have patches of red, irritated skin that are often covered by flaky white scales. Psoriasis likely results from a malfunction of the immune system in which the body's immune response turns against itself, attacking healthy skin cells by mistake.

Each of the known *IL23R* variations changes a single amino acid in the IL-23 receptor. One of these variations, Arg381Gln, appears to reduce the risk of developing psoriasis. (This variation has also been shown to protect against ankylosing spondylitis and Crohn disease, described above.) Other *IL23R* variations may increase the risk of developing psoriasis. Researchers suggest that changes in the *IL23R* gene may contribute to general problems with regulation of the immune system, which may help explain why these variations are related to several different disorders characterized by immune system dysfunction.

3. Other Names for This Gene

- IL-23R
- IL23R_HUMAN
- interleukin-23 receptor

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