IL23R gene

interleukin 23 receptor

Normal Function

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.

Health Conditions Related to Genetic Changes

Ankylosing spondylitis

Several variations (polymorphisms) in the *IL23R* gene have been found to influence the risk of ankylosing spondylitis. This condition is a form of painful, ongoing joint inflammation (chronic inflammatory arthritis) that primarily affects the spine.

One of these *IL23R* gene polymorphisms 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 fully known 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.

Crohn's disease

Several variants in or near the *IL23R* gene have been found to influence the risk of

developing Crohn's disease. These associations have been reported 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's disease. Although it is unclear how this change protects against Crohn's disease, researchers believe that the receptor's role in triggering inflammation in the intestinal walls may underlie its connection with this disorder.

Psoriatic arthritis

MedlinePlus Genetics provides information about Psoriatic arthritis

Ulcerative colitis

MedlinePlus Genetics provides information about Ulcerative colitis

Other disorders

Variants in the *IL23R* gene have also been associated with the risk of several other immune system-related conditions, including a skin disorder called psoriasis. In people with light skin, this chronic inflammatory condition causes patches of red, irritated skin that are often covered by flaky white scales, but these patches are darker looking in people with darker skin. 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' s 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.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IL23R (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=149233[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28IL23R%5BTI%5D%29+OR+%28interleukin+23+receptor%5BTI%5D%29%29+OR+%28IL-23R%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- INTERLEUKIN 23 RECEPTOR; IL23R (https://omim.org/entry/607562)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/149233)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IL23R[gene])

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Genomic Location

The *IL23R* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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