

## IRF5 gene

interferon regulatory factor 5

### Normal Function

The protein produced from the *IRF5* gene, called interferon regulatory factor 5 (IRF5), acts as a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of certain genes. When a virus is recognized in the cell, the *IRF5* gene is turned on (activated), which leads to the production of IRF5 protein. The protein binds to specific regions of DNA that regulate the activity of genes that produce interferons and other cytokines. Cytokines are proteins that help fight infection by promoting inflammation and regulating the activity of immune system cells. In particular, interferons control the activity of genes that help block the replication of viruses, and they stimulate the activity of certain immune system cells known as natural killer cells.

### Health Conditions Related to Genetic Changes

#### Rheumatoid arthritis

Studies have associated normal variations in the *IRF5* gene with an increased risk of rheumatoid arthritis. This condition causes chronic abnormal inflammation, primarily affecting the joints. Rheumatoid arthritis is an autoimmune disorders, which occur when the immune system malfunctions and attacks the body's tissues and organs.

There is some evidence that certain variations of the *IRF5* gene are associated with increased activity of the gene and elevated cytokines. However, it is unknown what role, if any, these effects play in the increased risk of rheumatoid arthritis. Researchers believe that a combination of genetic and environmental factors may contribute to the development of these conditions.

#### Systemic lupus erythematosus

Studies have associated normal variations in the *IRF5* gene with an increased risk of systemic lupus erythematosus. This is a chronic disease that causes inflammation in connective tissues, such as cartilage and the lining of blood vessels, which provide strength and flexibility to structures throughout the body. Systemic lupus erythematosus is an autoimmune disorders, which occur when the immune system malfunctions and attacks the body's tissues and organs.

There is some evidence that certain variations of the *IRF5* gene are associated with increased activity of the gene and elevated cytokines. However, it is unknown what role, if any, these effects play in the increased risk of systemic lupus erythematosus. Researchers believe that a combination of genetic and environmental factors may contribute to the development of these conditions.

### Systemic scleroderma

Several normal variations in the *IRF5* gene have been associated with an increased risk of developing systemic scleroderma, which is an autoimmune disorder characterized by the buildup of scar tissue (fibrosis) in the skin and internal organs. Although the *IRF5* gene is known to stimulate the immune system in response to viruses, it is unknown how the gene variations contribute to the increased risk of systemic scleroderma. Researchers believe that a combination of genetic and environmental factors may play a role in development of the condition.

### Ulcerative colitis

MedlinePlus Genetics provides information about Ulcerative colitis

## **Other Names for This Gene**

- IRF-5
- IRF5\_HUMAN
- SLEB10

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of IRF5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3663\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3663[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28IRF5%5BTIAB%5D%29+OR+%28interferon+regulatory+factor+5%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- SJOGREN SYNDROME (<https://omim.org/entry/270150>)
- RHEUMATOID ARTHRITIS; RA (<https://omim.org/entry/180300>)
- INTERFERON REGULATORY FACTOR 5; IRF5 (<https://omim.org/entry/607218>)

- SYSTEMIC LUPUS ERYTHEMATOSUS, SUSCEPTIBILITY TO, 10; SLEB10 (<https://omim.org/entry/612251>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3663>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=IRF5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=IRF5[gene]))

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## Genomic Location

The *IRF5* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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