

STAT4 gene

signal transducer and activator of transcription 4

Normal Function

The *STAT4* gene provides instructions for a protein that acts as a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of certain genes. The STAT4 protein is turned on (activated) by immune system proteins called cytokines, which are part of the inflammatory response to fight infection. When activated, the STAT4 protein increases the activity of genes that help immune cells called T-cells mature into specialized T-cells. These specialized T-cells, called Th1 cells, produce specific cytokines and stimulate other immune cells to get rid of foreign invaders (pathogens) in the cell.

Health Conditions Related to Genetic Changes

Systemic scleroderma

A normal variation in the *STAT4* gene has 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 *STAT4* gene is known to stimulate the immune system in response to pathogens, it is unknown how the gene variation contributes 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.

Juvenile idiopathic arthritis

MedlinePlus Genetics provides information about Juvenile idiopathic arthritis

Rheumatoid arthritis

Studies have associated a normal variation in the *STAT4* gene with an increased risk of rheumatoid arthritis. This condition is an autoimmune disorder, which occurs when the immune system malfunctions and attacks the body's tissues and organs.

The variant associated with increased risk of rheumatoid arthritis changes a single DNA building block (nucleotide) in the *STAT4* gene. It is unknown how the gene variation contributes to increased risk of this condition. Researchers believe that a combination of

genetic and environmental factors may play a role in development of autoimmunity.

Systemic lupus erythematosus

Studies have associated a normal variation in the *STAT4* gene with an increased risk of an autoimmune disorder called system lupus erythematosus.

The variant associated with increased risk of system lupus erythematosus changes a single nucleotide in the *STAT4* gene. It is unknown how the gene variation contributes to increased risk of system lupus erythematosus. Researchers believe that a combination of genetic and environmental factors are likely involved in development of autoimmunity.

Autoimmune disorders

Studies have associated a normal variation in the *STAT4* gene with an increased risk of an autoimmune disorder called Sjögren syndrome.

The variant associated with increased risk of this disorder changes a single nucleotide in the *STAT4* gene. It is unknown how the gene variation contributes to increased risk of this condition. Researchers believe that a combination of genetic and environmental factors may play a role in disease development.

Other Names for This Gene

- SLEB11
- STAT4_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of STAT4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6775\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6775[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28STAT4%5BTIAB%5D%29+OR+%28signal+transducer+and+activator+of+transcription+4%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+360+days%22%5Bdp%5D%29%29>)

Catalog of Genes and Diseases from OMIM

- SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 4; STAT4 (<https://omim.org/entry/600558>)

- SJOGREN SYNDROME (<https://omim.org/entry/270150>)
- RHEUMATOID ARTHRITIS; RA (<https://omim.org/entry/180300>)
- RHEUMATOID ARTHRITIS, SYSTEMIC JUVENILE (<https://omim.org/entry/604302>)
- SYSTEMIC LUPUS ERYTHEMATOSUS, SUSCEPTIBILITY TO, 11; SLEB11 (<https://omim.org/entry/612253>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6775>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=STAT4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=STAT4[gene]))

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Genomic Location

The *STAT4* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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