

# STAT4 Gene

Subjects: Genetics & Heredity

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Signal transducer and activator of transcription 4: 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.

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## 1. 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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.

### 2.2. Autoimmune disorders

Studies have associated a normal variation in the *STAT4* gene with an increased risk of several autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include systemic lupus erythematosus, rheumatoid arthritis, and Sjögren syndrome.

The variant associated with increased risk of autoimmune disorders changes a single DNA building block (nucleotide) in the *STAT4* gene. It is unknown how the gene variation contributes to increased risk of these conditions. Researchers believe that a combination of genetic and environmental factors may play a role in development of autoimmunity.

### 2.3. Other related diseases

- Juvenile idiopathic arthritis
- Rheumatoid arthritis
- Systemic lupus erythematosus

## 3. Other Names for This Gene

- SLEB11
  - STAT4\_HUMAN
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