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

Rheumatoid arthritis

Systemic lupus erythematosus

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.

Chromosomal Location

Cytogenetic Location: 2q32.2-q32.3, which is the long (q) arm of chromosome 2 between positions 32.2 and 32.3

Molecular Location: base pairs 191,029,576 to 191,172,684 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Other Names for This Gene

• SLEB11
• STAT4_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28STAT4%5BTIAB%5D%29+OR+%28signal+transducer+and+activator+of+transcription+4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22+AND+Fdb%5D

Catalog of Genes and Diseases from OMIM

• RHEUMATOID ARTHRITIS
  http://omim.org/entry/180300
• RHEUMATOID ARTHRITIS, SYSTEMIC JUVENILE
  http://omim.org/entry/604302
• SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 4
  http://omim.org/entry/600558
• SJOGREN SYNDROME
  http://omim.org/entry/270150
• SYSTEMIC LUPUS ERYTHEMATOSUS, SUSCEPTIBILITY TO, 11
  http://omim.org/entry/612253

Research Resources
• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_STAT4.html
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=STAT4%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6775
• NCBI Gene
• UniProt
  https://www.uniprot.org/uniprot/Q14765

Sources for This Summary
• OMIM: SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 4
  http://omim.org/entry/600558

  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2768040/


Reprinted from Genetics Home Reference:

Reviewed: September 2011
Published: June 25, 2019

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