

STING1 gene

stimulator of interferon response cGAMP interactor 1

Normal Function

The *STING1* gene provides instructions for making a protein that is involved in immune system function. STING helps produce beta-interferon, a member of a class of proteins called cytokines that promote inflammation. Inflammation normally occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight microbial invaders and help with tissue repair.

Health Conditions Related to Genetic Changes

STING-associated vasculopathy with onset in infancy

At least three *STING1* gene mutations have been identified in individuals with STING-associated vasculopathy with onset in infancy (SAVI), a potentially life-threatening disorder that damages the skin, lungs, blood vessels, and other tissues. The mutations that cause SAVI are described as "gain-of-function" mutations because they enhance the activity of the STING protein, leading to overproduction of beta-interferon. Abnormally high beta-interferon levels cause excessive inflammation, resulting in damage to the body's own cells and tissues and leading to the signs and symptoms of SAVI. Disorders such as SAVI that result from abnormally increased inflammation are known as autoinflammatory diseases.

Other Names for This Gene

- endoplasmic reticulum IFN stimulator
- endoplasmic reticulum interferon stimulator
- ERIS
- FLJ38577
- hMITA
- hSTING
- mediator of IRF3 activation
- MITA
- mitochondrial mediator of IRF3 activation

- MPYS
- N-terminal methionine-proline-tyrosine-serine plasma membrane tetraspanner
- NET23
- SAVI
- stimulator of interferon genes protein
- STING
- TMEM173
- transmembrane protein 173

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of STING1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=340061\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=340061[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28TMEM173%5BTIAB%5D%29+OR+%28%28ERIS%5BTIAB%5D%29+OR+%28MITA%5BTIAB%5D%29+OR+%28MPYS%5BTIAB%5D%29+OR+%28SAVI%5BTIAB%5D%29+OR+%28STING%5BTIAB%5D%29+OR+%28hSTING%5BTIAB%5D%29+OR+%28mediator+of+IRF3+activation%5BTIAB%5D%29+OR+%28endoplasmic+reticulum+IFN+stimulator%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>)

Catalog of Genes and Diseases from OMIM

- STIMULATOR OF INTERFERON RESPONSE cGAMP INTERACTOR 1; STING1 (<https://omim.org/entry/612374>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/340061>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=STING1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=STING1[gene]))

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Genomic Location

The *STING1* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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