

## **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%29>)

### 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]))

## References

- Burdette DL, Monroe KM, Sotelo-Troha K, Iwig JS, Eckert B, Hyodo M, Hayakawa Y, Vance RE. STING is a direct innate immune sensor of cyclic di-GMP. *Nature*. 2011 Sep 25;478(7370):515-8. doi: 10.1038/nature10429. Citation on PubMed (<https://pub>)

med.ncbi.nlm.nih.gov/21947006) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3203314/>)

- Chen H, Sun H, You F, Sun W, Zhou X, Chen L, Yang J, Wang Y, Tang H, Guan Y, Xia W, Gu J, Ishikawa H, Gutman D, Barber G, Qin Z, Jiang Z. Activation of STAT6 by STING is critical for antiviral innate immunity. *Cell*. 2011 Oct 14;147(2):436-46. doi: 10.1016/j.cell.2011.09.022. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22000020>)
- Chiliveru S, Rahbek SH, Jensen SK, Jorgensen SE, Nissen SK, Christiansen SH, Mogensen TH, Jakobsen MR, Iversen L, Johansen C, Paludan SR. Inflammatory cytokines break down intrinsic immunological tolerance of human primary keratinocytes to cytosolic DNA. *J Immunol*. 2014 Mar 1;192(5):2395-404. doi: 10.4049/jimmunol.1302120. Epub 2014 Jan 31. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24489095>)
- Ishikawa H, Barber GN. STING is an endoplasmic reticulum adaptor that facilitates innate immune signalling. *Nature*. 2008 Oct 2;455(7213):674-8. doi:10.1038/nature07317. Epub 2008 Aug 24. Erratum In: *Nature*. 2008 Nov 13;456(7219):274. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18724357>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2804933/>)
- Ishikawa H, Barber GN. The STING pathway and regulation of innate immune signaling in response to DNA pathogens. *Cell Mol Life Sci*. 2011 Apr;68(7):1157-65. doi: 10.1007/s00018-010-0605-2. Epub 2010 Dec 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21161320>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3056141/>)
- Ishikawa H, Ma Z, Barber GN. STING regulates intracellular DNA-mediated, type I interferon-dependent innate immunity. *Nature*. 2009 Oct 8;461(7265):788-92. doi:10.1038/nature08476. Epub 2009 Sep 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19776740>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4664154/>)
- Liu Y, Jesus AA, Marrero B, Yang D, Ramsey SE, Sanchez GAM, Tenbrock K, Wittkowski H, Jones OY, Kuehn HS, Lee CR, DiMattia MA, Cowen EW, Gonzalez B, Palmer I, DiGiovanna JJ, Biancotto A, Kim H, Tsai WL, Trier AM, Huang Y, Stone DL, Hill S, Kim HJ, St Hilaire C, Gurprasad S, Plass N, Chapelle D, Horkayne-Szakaly I, Foell D, Barysenka A, Candotti F, Holland SM, Hughes JD, Mehmet H, Issekutz AC, Raffeld M, McElwee J, Fontana JR, Minniti CP, Moir S, Kastner DL, Gadina M, Steven AC, Wingfield PT, Brooks SR, Rosenzweig SD, Fleisher TA, Deng Z, Boehm M, Paller AS, Goldbach-Mansky R. Activated STING in a vascular and pulmonary syndrome. *N Engl J Med*. 2014 Aug 7;371(6):507-518. doi:10.1056/NEJMoa1312625. Epub 2014 Jul 16. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25029335>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4174543/>)
- Paludan SR, Bowie AG. Immune sensing of DNA. *Immunity*. 2013 May 23;38(5):870-80. doi: 10.1016/j.immuni.2013.05.004. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23706668>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3683625/>)
- Xiao TS, Fitzgerald KA. The cGAS-STING pathway for DNA sensing. *Mol Cell*.

2013Jul 25;51(2):135-9. doi: 10.1016/j.molcel.2013.07.004. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23870141>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3782533/>)

## **Genomic Location**

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

**Last updated October 1, 2014**