

SAMHD1 gene

SAM and HD domain containing deoxynucleoside triphosphate triphosphohydrolase 1

Normal Function

The *SAMHD1* gene provides instructions for making an enzyme that helps cut (cleave) molecules called deoxynucleoside triphosphates (dNTPs) into their deoxynucleoside and triphosphate components. The dNTP molecules are needed for the replication and maintenance of the genetic material (DNA) in the body's cells, and the amount of available dNTPs must be tightly controlled for these functions to proceed normally. Invading viruses such as the human immunodeficiency virus (HIV) also need dNTPs in order to replicate themselves. The SAMHD1 enzyme helps regulate the amount of available dNTPs to both meet the needs of the body's cells and control viral infections.

Health Conditions Related to Genetic Changes

Aicardi-Goutières syndrome

At least 78 mutations in the *SAMHD1* gene have been identified in people with Aicardi-Goutières syndrome, a disorder that involves severe brain dysfunction (encephalopathy), skin lesions, and other health problems. Research suggests that mutations in this gene may lead to production of a SAMHD1 protein that does not properly regulate the amount of available dNTPs. As a result, DNA maintenance is impaired, allowing DNA damage to accumulate in cells. Research suggests that this DNA damage may lead to cell signaling that inappropriately activates an immune response. As a result, the immune system attacks the body's own tissues and organs, causing inflammatory damage to the brain, skin, and other body systems that lead to the characteristic features of Aicardi-Goutières syndrome.

Other Names for This Gene

- AGS5
- DCIP
- dendritic cell-derived IFNG-induced protein
- HDDC1
- Mg11
- monocyte protein 5

- MOP-5
- SAM domain and HD domain 1
- SAM domain and HD domain-containing protein 1
- SAMH1_HUMAN
- SBBI88

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SAMHD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=25939\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=25939[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SAMHD1%5BTIAB%5D%29+OR+%28%28MOP-5%5BTIAB%5D%29+OR+%28AGS5%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

- SAM DOMAIN- AND HD DOMAIN-CONTAINING PROTEIN 1; SAMHD1 (<https://omim.org/entry/606754>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/25939>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SAMHD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SAMHD1[gene]))

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Genomic Location

The *SAMHD1* gene is found on chromosome 20 (<https://medlineplus.gov/genetics/chromosome/20/>).

Last updated November 1, 2017