SAA1 gene

serum amyloid A1

Normal Function

The *SAA1* gene provides instructions for making a protein called serum amyloid A1. This protein is made primarily in the liver and circulates in low levels in the blood. Although its function is not fully understood, serum amyloid A1 appears to play a role in the immune system. Serum amyloid A1 may help repair damaged tissues, act as an antibacterial agent, and signal the migration of germ-fighting cells to sites of infection.

Levels of this protein increase in the blood and other tissues under conditions of inflammation. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight microbial invaders and facilitate tissue repair. When this has been accomplished, the body stops the inflammatory response to prevent damage to its own cells and tissues.

There are three versions of the serum amyloid A1 protein, known as alpha, beta, and gamma, which differ by one or two protein building blocks (amino acids). The frequency of these versions differs across populations. In white populations, for example, the alpha version predominates and gamma is rare. In the Japanese population, however, the three versions appear almost equally.

Health Conditions Related to Genetic Changes

Familial Mediterranean fever

Several studies of people with familial Mediterranean fever indicate that having the alpha version of the serum amyloid A1 protein increases the risk of a serious complication called amyloidosis. Amyloidosis involves the buildup of protein deposits that can lead to kidney failure if left untreated. Studies indicate that individuals with familial Mediterranean fever who also have the alpha version of the protein are two to seven times more likely to develop amyloidosis than are people with the beta or gamma version.

More serum amyloid A1 is produced in the body during episodes of inflammation such as those that occur in familial Mediterranean fever. This protein and related compounds may form abnormal clumps in the body's organs and tissues. It remains unclear, however, how the alpha version of serum amyloid A1 increases the susceptibility to

amyloidosis (or alternatively, how the beta and gamma versions may protect against this complication) in people with this disorder.

Other Names for This Gene

- PIG4
- SAA
- SAA_HUMAN
- TP53I4
- tumor protein p53 inducible protein 4

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SAA1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6288[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SAA1%5BTIAB%5D%29+OR+%28serum+amyloid+A1%5BTIAB%5D%29%29+OR+%28PIG4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SERUM AMYLOID A1; SAA1 (https://omim.org/entry/104750)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6288)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SAA1[gene])

References

- Bakkaloglu A, Duzova A, Ozen S, Balci B, Besbas N, Topaloglu R, Ozaltin F,Yilmaz E. Influence of Serum Amyloid A (SAA1) and SAA2 gene polymorphisms onrenal amyloidosis, and on SAA/C-reactive protein values in patients with familialmediterranean fever in the Turkish population. J Rheumatol. 2004Jun;31(6): 1139-42. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15170927)
- Ben-Chetrit E. Familial Mediterranean fever (FMF) and renal AAamyloidosis--

phenotype-genotype correlation, treatment and prognosis. J Nephrol.2003 May-Jun;16(3): 431-4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12832747)

- Booth DR, Booth SE, Gillmore JD, Hawkins PN, Pepys MB. SAA1 alleles as riskfactors in reactive systemic AA amyloidosis. Amyloid. 1998 Dec;5(4):262-5. doi: 10.3109/13506129809007299. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov /10036584)
- Cazeneuve C, Ajrapetyan H, Papin S, Roudot-Thoraval F, Genevieve D, MndjoyanE, Papazian M, Sarkisian A, Babloyan A, Boissier B, Duquesnoy P, Kouyoumdjian JC,Girodon-Boulandet E, Grateau G, Sarkisian T, Amselem S. Identification ofMEFV-independent modifying genetic factors for familial Mediterranean fever. Am JHum Genet. 2000 Nov;67(5):1136-43. doi: 10.1016/ S0002-9297(07)62944-9. Epub 2000Oct 3. Citation on PubMed (https://pubmed.ncbi .nlm.nih.gov/11017802) or Free article on PubMed Central (https://www.ncbi.nlm.nih. gov/pmc/articles/PMC1288556/)
- Delibas A, Oner A, Balci B, Demircin G, Bulbul M, Bek K, Erdogan O, Baysun S, Yilmaz E. Genetic risk factors of amyloidogenesis in familial Mediterraneanfever. Am J Nephrol. 2005 Sep-Oct;25(5):434-40. doi: 10.1159/000087824. Epub 2005Aug 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16118480)
- Gershoni-Baruch R, Brik R, Zacks N, Shinawi M, Lidar M, Livneh A. Thecontribution of genotypes at the MEFV and SAA1 loci to amyloidosis and diseaseseverity in patients with familial Mediterranean fever. Arthritis Rheum. 2003Apr;48(4):1149-55. doi: 10.1002/art.10944. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12687 559)
- Kelkitli E, Bilgici B, Tokgoz B, Dilek M, Bedir A, Akpolat I, Utas C, AkpolatT. SAA1 alpha/alpha alleles in amyloidosis. J Nephrol. 2006 Mar-Apr;19(2):189-91. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16736418)
- Medlej-Hashim M, Delague V, Chouery E, Salem N, Rawashdeh M, Lefranc G, Loiselet J, Megarbane A. Amyloidosis in familial Mediterranean fever patients: correlation with MEFV genotype and SAA1 and MICA polymorphisms effects. BMC MedGenet. 2004 Feb 10;5:4. doi: 10.1186/1471-2350-5-4. Citation on PubMed (http s://pubmed.ncbi.nlm.nih.gov/15018633) or Free article on PubMed Central (https://w ww.ncbi.nlm.nih.gov/pmc/articles/PMC356915/)
- Moriguchi M, Kaneko H, Terai C, Koseki Y, Kajiyama H, Inada S, Kitamura Y, Kamatani N. Relative transcriptional activities of SAA1 promoters polymorphic atposition -13(T/C): potential association between increased transcription andamyloidosis. Amyloid. 2005 Mar;12(1):26-32. doi: 10.1080/13506120500032394. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16076608)
- Ray A, Shakya A, Kumar D, Benson MD, Ray BK. Inflammation-responsivetranscription factor SAF-1 activity is linked to the development of amyloid Aamyloidosis. J Immunol. 2006 Aug 15;177(4):2601-9. doi:10.4049/jimmunol.177.4. 2601. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16888022)
- Stevens FJ. Hypothetical structure of human serum amyloid A protein. Amyloid. 2004 Jun;11(2):71-80. doi: 10.1080/13506120412331272296. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15478462)

- Thorn CF, Lu ZY, Whitehead AS. Tissue-specific regulation of the humanacute-phase serum amyloid A genes, SAA1 and SAA2, by glucocorticoids in hepaticand epithelial cells. Eur J Immunol. 2003 Sep;33(9):2630-9. doi:10.1002/eji.200323985. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12938239)
- Yamada T. Serum amyloid A (SAA): a concise review of biology, assay methodsand clinical usefulness. Clin Chem Lab Med. 1999 Apr;37(4):381-8. doi:10.1515/CCLM.1999.063. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10369107)

Genomic Location

The *SAA1* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

Last updated August 11, 2021