# SAA1 Gene

Subjects: Genetics & Heredity
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serum amyloid A1

Keywords: genes

## 1. 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 variants differs across populations. In white populations, the alpha version predominates and gamma is rare. In the Japanese population, however, the three versions appear almost equally.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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.

### 2.2. Other disorders

Among people with certain other inflammatory disorders, studies indicate that variants of the serum amyloid A1 protein also modify the risk of amyloidosis. For example, in the Japanese population, the gamma version of the protein appears to increase the risk of amyloidosis among adults with rheumatoid arthritis. Among white people with juvenile chronic arthritis, the alpha version indicates a high risk of developing amyloidosis.

More serum amyloid A1 is produced in the body during chronic inflammation such as occurs in these disorders. This protein and related compounds may form abnormal clumps in the body's organs and tissues. It remains unclear, however, how certain versions of serum amyloid A1 increase the susceptibility to amyloidosis.

## 3. Other Names for This Gene

- PIG4

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

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