

FAS gene

Fas cell surface death receptor

Normal Function

The *FAS* gene provides instructions for making a protein that is involved in cell signaling. Three FAS proteins group together to form a structure called a trimer, which then interacts with other molecules to perform its signaling function. This signaling initiates a process called a caspase cascade. The caspase cascade is a series of steps that results in the self-destruction of cells (apoptosis) when they are not needed.

Health Conditions Related to Genetic Changes

Autoimmune lymphoproliferative syndrome

At least 115 mutations in the *FAS* gene have been identified in people with a disorder of the immune system called autoimmune lymphoproliferative syndrome (ALPS). ALPS is characterized by the production of an abnormally large number of immune system cells (lymphocytes), resulting in enlargement of the lymph nodes (lymphadenopathy), the liver (hepatomegaly), and the spleen (splenomegaly). Autoimmune disorders, in which the immune system malfunctions and attacks the body's own tissues and organs, are also common in ALPS. People with ALPS have an increased risk of developing cancer of the immune system cells (lymphoma).

When the immune system is activated to fight an infection, large numbers of lymphocytes are produced. Normally, these lymphocytes undergo apoptosis when they are no longer required. *FAS* gene mutations lead to an abnormal trimer that interferes with the initiation of apoptosis. As a result, excess lymphocytes accumulate in the body's tissues and organs and often begin attacking them, leading to autoimmune disorders. Interference with apoptosis allows cells to multiply without control, leading to the lymphomas that occur in people with this disorder.

Juvenile idiopathic arthritis

MedlinePlus Genetics provides information about Juvenile idiopathic arthritis

Cancers

Studies have associated certain *FAS* gene variations with increased risk of developing

cancer, including cancers of the lung, breast, and esophagus. Researchers believe that these variations may affect the signaling that initiates apoptosis, increasing the risk that cells will multiply out of control and result in cancer.

Other Names for This Gene

- APO-1
- apo-1 antigen
- APO-1 cell surface antigen
- apoptosis antigen 1
- apoptosis-mediating surface antigen FAS
- APT1
- CD95
- CD95 antigen
- Fas (TNF receptor superfamily, member 6)
- Fas AMA
- Fas antigen
- FAS1
- FASLG receptor
- TNFRSF6
- TNR6_HUMAN
- tumor necrosis factor receptor superfamily member 6

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FAS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=355\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=355[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FAS%5BTI%5D%29+OR+%28Fas%5BTI%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+360+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- FAS CELL SURFACE DEATH RECEPTOR; FAS (<https://omim.org/entry/134637>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/355>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FAS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FAS[gene]))

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Genomic Location

The *FAS* gene is found on chromosome 10 (<https://medlineplus.gov/genetics/chromosome/10/>).

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