JAG1 gene

jagged canonical Notch ligand 1

Normal Function

The *JAG1* gene provides instructions for making a protein called Jagged-1, which is involved in an important pathway by which cells can signal to each other. The Jagged-1 protein is inserted into the membranes of certain cells. It connects with other proteins called Notch receptors, which are bound to the membranes of adjacent cells. These proteins fit together like a lock and its key. When a connection is made between the Jagged-1 and Notch proteins, it launches a series of signaling reactions (Notch signaling) affecting cell functions. Notch signaling controls how certain types of cells develop in a growing embryo, especially cells destined to be part of the heart, liver, eyes, ears, and spinal column. The Jagged-1 protein continues to play a role throughout life in the development of new blood cells.

Health Conditions Related to Genetic Changes

Alagille syndrome

At least 226 mutations in the *JAG1* gene have been identified in people with Alagille syndrome. Most of these mutations result in an abnormally short Jagged-1 protein that is missing the segment that normally spans the cell membrane (the transmembrane domain). Other mutations interfere with proper transport (trafficking) of the protein within the cell, preventing it from reaching the cell membrane.

The loss of Jagged-1 protein at the cell membrane precludes its interaction with Notch proteins and prevents cell signaling. The lack of Notch signaling causes errors in development that result in missing or narrowed bile ducts in the liver, heart defects, distinctive facial features, and changes in other parts of the body. People with *JAG1* gene mutations may have one or more of these problems. In particular, some affected individuals have a particular combination of heart defects known as tetralogy of Fallot without other signs or symptoms of Alagille syndrome. The type and severity of problems associated with Alagille syndrome may differ even within the same family.

Critical congenital heart disease

MedlinePlus Genetics provides information about Critical congenital heart disease

Cancers

Increased activity (expression) of the *JAG1* gene has been linked to certain cancers, including breast cancer and head and neck tumors. The increased expression of the *JAG1* gene may promote the development of new blood vessels that nourish a growing tumor. The altered gene expression may also enhance other cancer-related events such as cell division (proliferation) and the inflammatory response.

Other Names for This Gene

- AGS
- AHD
- AWS
- CD339
- CD339 antigen
- HJ1
- JAG1_HUMAN
- jagged 1 (Alagille syndrome)
- jagged 1 precursor
- JAGL1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of JAG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=182[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28JAG1%5BTIAB%5D%29+OR+%28jagged+1%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- JAGGED 1; JAG1 (https://omim.org/entry/601920)
- TETRALOGY OF FALLOT; TOF (https://omim.org/entry/187500)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/182)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=JAG1[gene])

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Genomic Location

The *JAG1* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

Last updated April 1, 2010