

## HEPACAM gene

hepatic and glial cell adhesion molecule

### Normal Function

The *HEPACAM* gene provides instructions for making a protein called GlialCAM. This protein is found in liver cells and certain brain cells known as glial cells. In liver cells, GlialCAM plays a role in the attachment of cells to one another (adhesion) and cell movement. In glial cells, GlialCAM attaches (binds) to other GlialCAM proteins or to other proteins called MLC1 and CIC-2. GlialCAM ensures that these proteins are transported to junctions that connect neighboring glial cells. The function of GlialCAM at the cell junction is unclear.

### Health Conditions Related to Genetic Changes

#### Megalencephalic leukoencephalopathy with subcortical cysts

At least 20 mutations in the *HEPACAM* gene have been found to cause megalencephalic leukoencephalopathy with subcortical cysts.

This condition affects brain development and function, resulting in problems with movement and recurrent seizures. *HEPACAM* gene mutations lead to a protein that is unable to correctly transport proteins to the cell junction. It is unknown how a lack of functional GlialCAM protein (or certain other proteins) at neuronal cell junctions impairs brain development and function, causing the signs and symptoms of megalencephalic leukoencephalopathy with subcortical cysts.

Mutations in the *HEPACAM* gene cause two types of megalencephalic leukoencephalopathy with subcortical cysts, type 2A and type 2B; together, these types account for 20 percent of all cases. A major difference between the two types is that the signs and symptoms of type 2B improve over time. The reasons for this improvement are unclear.

### Other Names for This Gene

- FLJ25530
- glial cell adhesion molecule
- GlialCAM
- HECAM\_HUMAN

- hepatocyte and glial cell adhesion molecule
- hepatocyte cell adhesion molecule

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of HEPACAM ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=220296\[genedj\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=220296[genedj]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28HEPACAM%5BTIAB%5D%29+OR+%28GlialCAM%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+3600+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- HEPATOCYTE CELL ADHESION MOLECULE; HEPACAM (<https://omim.org/entry/611642>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/220296>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HEPACAM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HEPACAM[gene]))

## **References**

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## Genomic Location

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

**Last updated March 1, 2015**