MLC1 gene
megalecephalic leukoencephalopathy with subcortical cysts

Normal Function

The MLC1 gene provides instructions for making a protein that is found primarily in the brain but also in the spleen and white blood cells (leukocytes). Within the brain, the MLC1 protein is found in astroglial cells, which are a specialized form of brain cells called glial cells. Glial cells protect and maintain other nerve cells (neurons). The MLC1 protein functions at junctions that connect neighboring astroglial cells. The role of the MLC1 protein at the cell junction is unknown, but research suggests that it may control the flow of fluids into cells or the strength of cells' attachment to one another (cell adhesion). Studies indicate that the MLC1 protein may be involved in transporting molecules across the blood-brain barrier and the brain-cerebrospinal fluid barrier. These barriers protect the brain's delicate nerve tissue by allowing only certain substances to pass into the brain.

Health Conditions Related to Genetic Changes

Megalecephalic leukoencephalopathy with subcortical cysts

More than 80 mutations in the MLC1 gene have been found to cause megalencephalic leukoencephalopathy with subcortical cysts type 1; this type accounts for 75 percent of all cases. This condition affects brain development and function, resulting in problems with movement and recurrent seizures. Most of the MLC1 gene mutations that cause this condition change single protein building blocks (amino acids) in the MLC1 protein. These changes alter the structure of the MLC1 protein or prevent the cell from producing any protein. It is unknown how a lack of MLC1 protein at astroglial cell junctions impairs brain development and function, causing the signs and symptoms of megalencephalic leukoencephalopathy with subcortical cysts type 1.
Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 50,059,391 to 50,085,929 on chromosome 22 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Other Names for This Gene

• KIAA0027
• LVM
• megalencephalic leukoencephalopathy with subcortical cysts 1 gene product
• MLC
• MLC1_HUMAN
• VL

Additional Information & Resources

Educational Resources

• Basic Neurochemistry (sixth edition, 1999): The Blood-Brain Barrier Undergoes Development
  https://www.ncbi.nlm.nih.gov/books/NBK28180/#A2335

• Basic Neurochemistry (sixth edition, 1999): Virtually Nothing Can Enter or Leave the Central Nervous System Parenchyma Without Passing Through an Astrocytic Interphase
  https://www.ncbi.nlm.nih.gov/books/NBK28217/#A32

  https://www.ncbi.nlm.nih.gov/books/NBK26857/

Clinical Information from GeneReviews

• Megalencephalic Leukoencephalopathy with Subcortical Cysts
  https://www.ncbi.nlm.nih.gov/books/NBK1535
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MLC1%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

Catalog of Genes and Diseases from OMIM

- MLC1 GENE
  http://omim.org/entry/605908

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MLC1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:23209
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q15049

Sources for This Summary

OMIM: MLC1 GENE
http://omim.org/entry/605908

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