CASK-related disorders

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→

Genes

→

CASK gene

CASK gene

calcium/calmodulin dependent serine protein kinase

Normal Function

The CASK gene provides instructions for making a protein called calcium/calmodulin-dependent serine protein kinase (CASK). The CASK protein is primarily found in nerve cells (neurons) in the brain, where it helps control the activity (expression) of other genes that are involved in brain development. It also helps regulate the movement of chemicals called neurotransmitters and of charged atoms (ions), which are necessary for signaling between neurons. Research suggests that the CASK protein may also interact with the protein produced from another gene, FRMD7, to promote development of the nerves that control eye movement (the oculomotor neural network).

Health Conditions Related to Genetic Changes

CASK-related intellectual disability

More than 35 CASK gene mutations have been identified in people with CASK-related intellectual disability. This disorder affects brain development and has two main forms: a severe form called microcephaly with pontine and cerebellar hypoplasia (MICPCH), and a milder form called X-linked intellectual disability (XL-ID) with or without nystagmus.The mutations that cause CASK-related intellectual disability affect the role of the CASK protein in brain development and function. MICPCH is caused by mutations that eliminate CASK function, while mutations that impair the function of this protein cause XL-ID with or without nystagmus. Nystagmus refers to rapid, involuntary back-and-forth eye movements. Affected individuals with nystagmus may have CASK gene mutations that disrupt the interaction between the CASK protein and the protein produced from the FRMD7 gene, leading to problems with the development of the oculomotor neural network and resulting in abnormal eye movements.

More About This Health Condition

FG syndrome

MedlinePlus Genetics provides information about FG syndrome

More About This Health Condition

Other Names for This Gene

calcium/calmodulin-dependent serine protein kinase (MAGUK family) CAMGUK CMG

CSKP_HUMAN hCASK LIN2 protein lin-2 homolog TNRC8

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of CASK

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE; CASK

Gene and Variant Databases

NCBI Gene

ClinVar

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Genomic LocationThe CASK gene is found on the X chromosome.

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