

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.

FG syndrome

MedlinePlus Genetics provides information about FG syndrome

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 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8573\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8573[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28CASK%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+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE; CASK (<https://omim.org/entry/300172>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8573>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=CASK\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=CASK[gene]))

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Genomic Location

The CASK gene is found on the X chromosome (<https://medlineplus.gov/genetics/chrom>)

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