MAP2K2 gene

mitogen-activated protein kinase kinase 2

Normal Function

The *MAP2K2* gene provides instructions for making a protein known as MEK2 protein kinase. This protein is part of a signaling pathway called the RAS/MAPK pathway, which transmits chemical signals from outside the cell to the cell's nucleus. RAS/MAPK signaling helps control the growth and division (proliferation) of cells, the process by which cells mature to carry out specific functions (differentiation), cell movement, and the self-destruction of cells (apoptosis).

The *MAP2K2* gene is very similar to a gene called *MAP2K1*, which provides instructions for making a protein known as MEK1 protein kinase. Like MEK2 protein kinase, this protein functions as part of the RAS/MAPK signaling pathway. Together, the MEK1 and MEK2 protein kinases appear to be essential for normal development before birth and for survival after birth.

Health Conditions Related to Genetic Changes

Cardiofaciocutaneous syndrome

At least 13 mutations in the *MAP2K2* gene have been identified in people with cardiofaciocutaneous syndrome. Most of these mutations change single protein building blocks (amino acids) in MEK2 protein kinase, although one mutation deletes several amino acids from the protein. These genetic changes abnormally activate MEK2 kinase, which disrupts the tightly regulated RAS/MAPK signaling pathway in cells throughout the body. The altered signaling interferes with the normal development of many organs and tissues, resulting in the characteristic features of cardiofaciocutaneous syndrome.

Other Names for This Gene

- dual specificity mitogen-activated protein kinase kinase 2
- ERK activator kinase 2
- MAP kinase kinase 2
- MAPK-ERK Kinase 2
- MAPK/ERK kinase 2
- MAPKK2

- MEK2
- mitogen-activated protein kinase kinase 2, p45
- MKK2
- MP2K2_HUMAN
- PRKMK2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MAP2K2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5605[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MAP2K2%5BTIAB%5D%29+OR+%28mitogen-activated+protein+kinase+kinase+2%5BTIAB%5D%29%29+OR+%28%28MAPKK2%5BTIAB%5D%29+OR+%28MEK2%5BTIAB%5D%29+OR+%28MKK2%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

- MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2; MAP2K2 (https://omim.org/entry/601263)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5605)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MAP2K2[gene])

References

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Genomic Location

The *MAP2K2* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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