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.
Chromosomal Location

Cytogenetic Location: 19p13.3, which is the short (p) arm of chromosome 19 at position 13.3

Molecular Location: base pairs 4,090,321 to 4,124,184 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

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

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK9870/#A2252

Clinical Information from GeneReviews

- Cardiofaciocutaneous Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1186
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MAP2K2%5BTIAB%5D %29+OR+%28mitogen-activated+protein+kinase+kinase+2%5BTIAB%5D %29%29+OR+%28%28MAPKK2%5BTIAB%5D%29%29+OR+%28MEK2%5BTIAB %5D%29+OR+%28MKK2%5BTIAB%5D%29%29+AND+%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
  http://omim.org/entry/601263

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MAP2K2.html

- ClinVar

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5605

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P36507

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16569817

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17366577


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