KIF21A gene

kinesin family member 21A

Normal Function

The *KIF21A* gene provides instructions for making a protein called kinesin family member 21A. Proteins in the kinesin family are essential for the transport of materials within cells. Kinesin proteins function like freight trains that transport cargo, and their structure is suited for this cargo-carrying function. One end of the protein, called the motor domain, provides power to move the protein and its cargo along a track-like system made from structures called microtubules. The other end of the protein attaches (binds) to specific cargo, such as groups of proteins, for transport. The two ends of each kinesin are connected by a flexible region known as the stalk.

Kinesin family member 21A is found in developing nerve cells (neurons). Researchers believe that this protein carries cargo that is needed for the normal development and function of nerves in the head and face. In particular, this kinesin plays a critical role in the development of cranial nerve III, which emerges from the brain and controls several of the muscles that surround the eyes (extraocular muscles). These muscles direct eye movement and determine the position of the eyes.

Health Conditions Related to Genetic Changes

Congenital fibrosis of the extraocular muscles

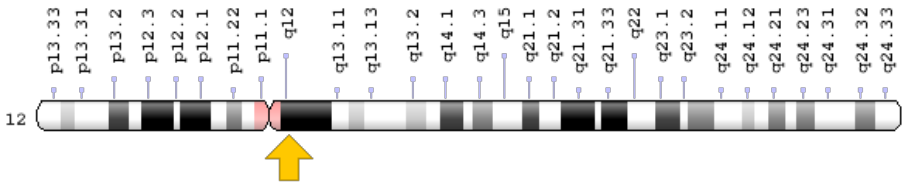
At least 11 mutations in the *KIF21A* gene have been identified in people with congenital fibrosis of the extraocular muscles. These mutations cause the most common form of the disorder, CFEOM1, and are a rare cause of another form of the condition called CFEOM3.

Each of the known *KIF21A* mutations changes a single protein building block (amino acid) in kinesin family member 21A. Most of these changes occur in the stalk region of the protein. These mutations alter the protein's structure, which likely interferes with its ability to transport cargo within neurons. As a result, several cranial nerves and the extraocular muscles they control do not develop normally. Abnormal development and function of these muscles leads to the characteristic features of congenital fibrosis of the extraocular muscles, including restricted eye movement and related problems with vision.

Chromosomal Location

Cytogenetic Location: 12q12, which is the long (q) arm of chromosome 12 at position 12

Molecular Location: base pairs 39,293,228 to 39,443,390 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CFEOM
- CFEOM1
- DKFZp779C159
- FEOM
- FEOM1
- Fibrosis of extraocular muscles, congenital, 1, autosomal dominant
- fibrosis of the extraocular muscles, congenital, 1
- FLJ20052
- KI21A_HUMAN
- KIAA1708
- KIF2
- KIF21A variant protein
- Kinesin-like protein KIF2
- Kinesin-like protein KIF21A
- NY-REN-62 antigen
- Renal carcinoma antigen NY-REN-62

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): There Are Two Types of Microtubule Motor Proteins: Kinesins and Dyneins
  https://www.ncbi.nlm.nih.gov/books/NBK26888/#A3047
- Neuroscience (2nd edition, 2001): The Actions and Innervation of Extraocular Muscles
  https://www.ncbi.nlm.nih.gov/books/NBK10793/
- The Engle Laboratory, Boston Children's Hospital: KIF21A
  http://www.childrenshospital.org/Research/Labs/engle-laboratory/neurodevelopmental-research/kif21a

Clinical Information from GeneReviews

- Congenital Fibrosis of the Extraocular Muscles
  https://www.ncbi.nlm.nih.gov/books/NBK1348

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KIF21A%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

- KINESIN FAMILY MEMBER 21A
  http://omim.org/entry/608283

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KIF21A%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:19349
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:55605
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/55605
- UniProt
  https://www.uniprot.org/uniprot/Q7Z4S6

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