KIF21A gene

kinesin family member 21A

Normal Function

The *KIF21A* gene provides instructions for making a protein that is part of the kinesin family. Many proteins in the kinesin family are essential for the transport of materials within cells. Kinesin proteins function like freight trains that transport cargo along a track-like system made from structures called microtubules. Some kinesins also help maintain microtubules. As well as functioning like a track, microtubules make up the structural framework of cells and help cells move.

The KIF21A protein is found in nerve cells (neurons) and many other cell types. Researchers believe that this protein plays an important role in neuron development by helping control the growth of microtubules. By blocking microtubule growth at critical times, the KIF21A protein may help direct the path of neuron extensions known as axons so they can reach their correct locations. Once in the right position, axons relay messages to and from the brain to control muscle movement and detect sensations such as touch, pain, and heat.

For proper neuron development, the KIF21A protein must be turned on and off at particular times. When a segment of the protein known as the regulatory region interacts with another segment of the protein known as the motor domain, the protein is turned off (which is known as autoinhibition).

Health Conditions Related to Genetic Changes

Congenital fibrosis of the extraocular muscles

At least 12 mutations in the *KIF21A* gene have been identified in people with congenital fibrosis of the extraocular muscles (CFEOM). These mutations cause the most common form of the disorder, CFEOM1, and are a rare cause of another form of the condition called CFEOM3. Individuals with CFEOM are unable to move their eyes normally. They have difficulty looking upward or, less commonly, side-to-side, and most also have droopy eyelids (ptosis). In addition, people with CFEOM3 can have intellectual disability or other neurological problems.

Each of the known *KIF21A* gene mutations changes a single protein building block (amino acid) in the KIF21A protein. Most of these changes occur in the regulatory region

of the protein. These mutations alter the protein's structure, which interferes with its ability to turn itself off. As a result, the KIF21A protein is always on (constitutively active) and cannot regulate microtubule growth. Without proper control of microtubule elongation, the axons of nerves develop abnormally and do not reach the muscles they control. Nerves in the head and face (cranial nerves) that control muscles that surround the eyes (extraocular muscles) are particularly affected. Problems with cranial nerve development impair extraocular muscle function, resulting in the characteristic features of CFEOM such as restricted eye movement and droopy eyelids.

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

Tests Listed in the Genetic Testing Registry

- Tests of KIF21A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55605[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; KIF21A (https://omim.org/entry/608283)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55605)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KIF21A[gene])

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Genomic Location

The *KIF21A* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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