

KIF7 gene

kinesin family member 7

Normal Function

The *KIF7* gene provides instructions for making a protein that is associated with structures called primary cilia. These microscopic, finger-like projections stick out from the surface of cells and are involved in signaling pathways that transmit information into cells. Studies suggest that the KIF7 protein helps to maintain the proper length and stability of cilia.

Through its association with primary cilia, the KIF7 protein helps regulate a signaling pathway known as Sonic Hedgehog. This pathway is essential for early development. It plays roles in cell growth, cell specialization, and the normal shaping (patterning) of many parts of the body, including the brain and limbs.

Health Conditions Related to Genetic Changes

Acrocallosal syndrome

At least 20 mutations in the *KIF7* gene have been identified in people with acrocallosal syndrome. This rare condition is characterized by certain brain abnormalities, the presence of extra fingers and toes (polydactyly), and distinctive facial features, including widely spaced eyes (hypertelorism) and a prominent forehead. Most of the *KIF7* gene mutations that cause acrocallosal syndrome lead to the production of an abnormally short, nonfunctional version of the KIF7 protein or prevent any protein from being produced from the gene. Little is known about the effects of these mutations, although they likely disrupt Sonic Hedgehog signaling during early development. It is unclear how these changes impair the development of the brain, limbs, and other parts of the body in people with acrocallosal syndrome.

Joubert syndrome

MedlinePlus Genetics provides information about Joubert syndrome

Other Names for This Gene

- ACLS
- AGBK

- EQYK340
- HLS2
- JBTS12
- kinesin-like protein KIF7
- UNQ340

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KIF7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=374654\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=374654[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28KIF7%5BTIAB%5D%29+OR+%28kinesin+family+member+7%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- KINESIN FAMILY MEMBER 7; KIF7 (<https://omim.org/entry/611254>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/374654>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KIF7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KIF7[gene]))

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Genomic Location

The *KIF7* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

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