IFT80 gene

intraflagellar transport 80

Normal Function

The *IFT80* gene provides instructions for making a protein that is part of a group (complex) called IFT complex B. This complex is found in cell structures known as cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells. IFT complex B is involved in a process called intraflagellar transport (IFT), by which materials are carried within cilia. Specifically, this complex helps transport materials from the base of cilia to the tip.

IFT is essential for the assembly and maintenance of cilia. These cell structures play central roles in many different chemical signaling pathways, including a series of reactions called the Sonic Hedgehog pathway. These pathways are important for the growth and division (proliferation) and maturation (differentiation) of cells. In particular, Sonic Hedgehog appears to be essential for the proliferation and differentiation of cells that ultimately give rise to cartilage and bone.

Health Conditions Related to Genetic Changes

Asphyxiating thoracic dystrophy

Mutations in the *IFT80* gene were the first genetic changes found to cause asphyxiating thoracic dystrophy, an inherited disorder of bone growth characterized by a small chest, short ribs, and shortened bones in the arms and legs. At least six mutations in the *IFT80* gene have since been associated with this disorder. Most of these mutations change single protein building blocks (amino acids) in the IFT80 protein. IFT complex B made with the altered protein cannot function normally, which disrupts the transport of materials within cilia. Researchers speculate that these changes in IFT alter certain signaling pathways, including the Sonic Hedgehog pathway, which may underlie the abnormalities of bone growth characteristic of asphyxiating thoracic dystrophy.

In some affected individuals, asphyxiating thoracic dystrophy is also associated with abnormalities of the kidneys, liver, retinas, and other tissues. However, when the disorder results from *IFT80* gene mutations, its features are usually limited to problems with bone growth. The reasons for this difference are unknown.

Other Names for This Gene

- ATD2
- IFT80_HUMAN
- intraflagellar transport 80 homolog (Chlamydomonas)
- KIAA1374
- MGC126543
- WD repeat domain 56
- WD repeat-containing protein 56
- WDR56

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IFT80 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57560[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28IFT80%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- INTRAFLAGELLAR TRANSPORT 80; IFT80 (https://omim.org/entry/611177)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/57560)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IFT80[gene])

References

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Genomic Location

The *IFT80* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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