IFT140 gene

intraflagellar transport 140

Normal Function

The *IFT140* gene provides instructions for making a protein that is involved in the formation and maintenance of cilia, which are microscopic, finger-like projections that stick out from the surface of cells and participate in signaling pathways that transmit information within and between cells. Cilia are important for the structure and function of many types of cells, including cells in the kidneys, liver, and brain. Light-sensing cells (photoreceptors) in the retina also contain cilia, which are essential for normal vision. Cilia also play a role in the development of the bones, although the mechanism is not well understood.

The movement of substances within cilia and similar structures called flagella is known as intraflagellar transport. This process is essential for the assembly and maintenance of these cell structures. During intraflagellar transport, cells use molecules called IFT particles to carry materials to and from the tips of cilia. IFT particles are made of proteins produced from related genes that belong to the IFT gene family. Each IFT particle is made up of two groups of IFT proteins: complex A, which includes at least 6 proteins, and complex B, which includes at least 15 proteins. The protein produced from the *IFT140* gene forms part of IFT complex A (IFT-A).

Health Conditions Related to Genetic Changes

Mainzer-Saldino syndrome

At least nine *IFT140* gene mutations have been identified in people with Mainzer-Saldino syndrome, a disorder characterized by kidney disease, eye problems, and skeletal abnormalities. Mutations in the *IFT140* gene that cause Mainzer-Saldino syndrome may change the shape of the IFT140 protein or its interactions with other IFT proteins, likely impairing the assembly of IFT-A and the development or maintenance of cilia. As a result, fewer cilia may be present or functional, affecting many organs and tissues in the body and resulting in the signs and symptoms of Mainzer-Saldino syndrome. Disorders such as Mainzer-Saldino syndrome that are caused by problems with cilia and involve bone abnormalities are called skeletal ciliopathies.

Asphyxiating thoracic dystrophy

MedlinePlus Genetics provides information about Asphyxiating thoracic dystrophy

Other Names for This Gene

- c305C8.4
- c380F5.1
- gs114
- IF140_HUMAN
- intraflagellar transport 140 homolog (Chlamydomonas)
- intraflagellar transport protein 140 homolog
- KIAA0590
- MZSDS
- WD and tetratricopeptide repeats protein 2
- WDTC2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IFT140 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9742[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28IFT140%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- INTRAFLAGELLAR TRANSPORT 140; IFT140 (https://omim.org/entry/614620)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9742)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IFT140[gene])

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Genomic Location

The *IFT140* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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