IFT122 gene

intraflagellar transport 122

Normal Function

The *IFT122* 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. Cilia participate in signaling pathways that transmit information within and between cells and are important for the development and function of many types of cells and tissues, including cells in the kidneys and liver and the light-sensitive tissue at the back of the eye (the retina). 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. Each IFT particle is made up of two groups of IFT proteins: complex A and complex B. The protein produced from the *IFT122* gene forms part of IFT complex A (IFT-A). During intraflagellar transport, this complex carries materials from the tip to the base of cilia.

The IFT-A complex is essential for proper regulation of the Sonic Hedgehog signaling pathway, which is important for the growth and maturation (differentiation) of cells and the normal shaping (patterning) of many parts of the body, especially during embryonic development. The exact role of the complex in this pathway is unclear.

Health Conditions Related to Genetic Changes

Cranioectodermal dysplasia

Mutations in the *IFT122* gene can cause cranioectodermal dysplasia. This condition is characterized by an elongated head (dolichocephaly) with a prominent forehead and other distinctive facial features; short bones; and abnormalities of certain tissues known as ectodermal tissues, which include the teeth, hair, nails, and skin.

At least six *IFT122* gene mutations have been found in people with cranioectodermal dysplasia. These mutations reduce the amount or function of the IFT122 protein. A shortage or reduction in activity of this component of the IFT-A complex impairs the function of the entire complex, disrupting transport of proteins and materials from the

tips of cilia. As a result, assembly and maintenance of cilia is impaired, which leads to a smaller number of cilia and abnormalities in their shape and structure. Although the mechanism is unclear, a loss of normal cilia impedes proper development of bone and other tissues, leading to the features of cranioectodermal dysplasia. Some researchers suggest that disrupted intraflagellar transport prevents signaling through the Sonic Hedgehog pathway, which could impact cell growth and other functions in several tissues throughout the body.

Other Names for This Gene

- CED
- CED1
- IF122_HUMAN
- intraflagellar transport 122 homolog (Chlamydomonas)
- intraflagellar transport protein 122 homolog
- SPG
- WD repeat domain 10
- WD repeat-containing protein 10
- WD repeat-containing protein 140
- WDR10
- WDR10p
- WDR140

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IFT122 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55764[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28IFT122%5BTIAB%5D%29+OR+%28%28WDR10%5BTIAB%5D%29+OR+%28WDR10p%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- INTRAFLAGELLAR TRANSPORT 122; IFT122 (https://omim.org/entry/606045)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55764)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IFT122[gene])

References

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Genomic Location

The *IFT122* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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