FGF10 gene

fibroblast growth factor 10

Normal Function

The *FGF10* gene provides instructions for making a protein called fibroblast growth factor 10 (FGF10). This protein is part of a family of proteins called fibroblast growth factors that are involved in important processes such as cell division, regulation of cell growth and maturation, formation of blood vessels, wound healing, and development before birth. By attaching to another protein known as a receptor, the FGF10 protein triggers a cascade of chemical reactions inside the cell that signals the cell to undergo certain changes, such as maturing to take on specialized functions. During development before birth, the signals triggered by the FGF10 protein appear to stimulate cells to form the structures that make up the ears, skeleton, organs, and glands in the eyes and mouth.

Health Conditions Related to Genetic Changes

Lacrimo-auriculo-dento-digital syndrome

At least three mutations in the *FGF10* gene have been found to cause lacrimo-auriculo-dento-digital (LADD) syndrome. This disorder affects the formation of the lacrimal system (the system in the eyes that produces and secretes tears), the ears, the salivary glands (the glands in the mouth that produce saliva), the teeth, the hands, and sometimes, other parts of the body. The main features of LADD syndrome are abnormal tear production, malformed ears with hearing loss, decreased saliva production, small teeth, and hand deformities.

The *FGF10* gene mutations that cause LADD syndrome reduce the amount and activity of FGF10 protein. Less growth factor is available to bind to receptors, which decreases signaling within cells. A decrease in cell signaling disrupts cell maturation and development, which results in abnormal formation of the ears, skeleton, and glands in the eyes and mouth in people with LADD syndrome.

Other disorders

Mutations in the *FGF10* gene have also been found to cause a condition similar to LADD syndrome called aplasia of lacrimal and salivary glands (ALSG). Individuals with this condition have absent (aplastic) or small (hypoplastic) lacrimal glands, which

secrete tears in the eyes, and aplastic or hypoplastic salivary glands. This condition often causes eye irritability, chronic tearing (epiphora), dry mouth (xerostomia), and a greater susceptibility to cavities. Sometimes, other structures that are involved in tear production are affected. The mutations that cause ALSG are thought to be less disruptive to cell signaling than the *FGF10* gene mutations that cause LADD syndrome, although in rare cases, the same mutation can cause either condition. Because there is overlap between the features of ALSG and LADD syndrome and because they are caused by mutations in the same gene, it is difficult to discern whether they are distinct disorders or part of the same disease spectrum.

Other Names for This Gene

- FGF-10
- FGF10_HUMAN
- keratinocyte growth factor 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FGF10 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2255[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FGF10%5BTIAB%5D%29+OR+%28fibroblast+growth+factor+10%5BTIAB%5D%29%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

- APLASIA OF LACRIMAL AND SALIVARY GLANDS; ALSG (https://omim.org/entry/180920)
- FIBROBLAST GROWTH FACTOR 10; FGF10 (https://omim.org/entry/602115)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2255)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FGF10[gene])

References

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- Shams I, Rohmann E, Eswarakumar VP, Lew ED, Yuzawa S, Wollnik B, SchlessingerJ, Lax I. Lacrimo-auriculo-dento-digital syndrome is caused by reduced activityof the fibroblast growth factor 10 (FGF10)-FGF receptor 2 signaling pathway. MolCell Biol. 2007 Oct;27(19):6903-12. doi: 10.1128/MCB.00544-07. Epub 2007 Aug 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17682060) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2099236/)

Genomic Location

The *FGF10* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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