GDF6 gene

growth differentiation factor 6

Normal Function

The *GDF6* gene provides instructions for making a protein that is part of the transforming growth factor beta (TGFβ) superfamily, which is a group of proteins that help control the growth and development of tissues throughout the body. Within the TGFβ superfamily, the GDF6 protein belongs to the bone morphogenetic protein family, which helps regulate the growth and maturation (differentiation) of bone and cartilage. Cartilage is a tough but flexible tissue that makes up much of the skeleton during early development. The proteins in this family help regulate cell growth and differentiation both before and after birth. The GDF6 protein is necessary for the formation of bones and joints in the limbs, skull, spine, chest, and ribs.

The GDF6 protein is also involved in the development of the eyes, specifically the specialized light-sensitive tissue that lines the back of the eye called the retina. The GDF6 protein likely plays a role in the survival of specialized cells within the retina that detect light and color (photoreceptor cells).

Health Conditions Related to Genetic Changes

Klippel-Feil syndrome

Variants (also called mutations) in the *GDF6* gene have been found to cause Klippel-Feil syndrome, a condition that is characterized by the abnormal joining (fusion) of two or more spinal bones in the neck (cervical vertebrae) and a variety of other features that affect many parts of the body. Most of the *GDF6* gene variants that cause Klippel-Feil syndrome result in the substitution of one protein building block (amino acid) for another in the GDF6 protein. This reduces the amount of normal GDF6 protein in cells. Although the GDF6 protein is involved in bone growth and the formation of vertebrae, it is unclear exactly how a shortage of this protein leads to the incomplete separation and fusion of the cervical vertebrae seen in people with Klippel-Feil syndrome.

Anophthalmia/Microphthalmia

MedlinePlus Genetics provides information about Anophthalmia/Microphthalmia

Coloboma

MedlinePlus Genetics provides information about Coloboma

Leber congenital amaurosis

MedlinePlus Genetics provides information about Leber congenital amaurosis

Nonsyndromic hearing loss

MedlinePlus Genetics provides information about Nonsyndromic hearing loss

Other disorders

Variants in the *GDF6* gene can also cause multiple synostoses syndrome 4, a condition that is characterized by hearing loss and the fusion of the bones in the hands and feet. The *GDF6* variants that cause this condition typically lead to the substitution of one amino acid for another in the GDF6 protein. It is unclear how changes in the *GDF6* gene can affect different parts of the body.

Other Names for This Gene

- BMP13
- CDMP2
- GDF-6
- growth/differentiation factor 6
- KFS
- KFS1
- SCDO4
- SGM1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GDF6 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=392255[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GDF6%5BTIAB%5D%29+OR+%28growth+differentiation+factor+6%5BTIAB%5D%29%29+OR+%28BMP13%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GROWTH/DIFFERENTIATION FACTOR 6; GDF6 (https://omim.org/entry/601147)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/392255)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GDF6[gene])

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Genomic Location

The *GDF6* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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