

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 (TGF β) superfamily, which is a group of proteins that help control the growth and development of tissues throughout the body. Within the TGF β superfamily, the GDF6 protein belongs to the bone morphogenetic protein family, which is involved in regulating 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 are regulators of 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 protein is involved in setting up boundaries between bones during skeletal development.

The GDF6 protein has also been found to be 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

At least 10 mutations in the *GDF6* gene have been found to cause Klippel-Feil syndrome, a condition characterized by the abnormal joining (fusion) of two or more spinal bones in the neck (cervical vertebrae) and a variety of other features affecting many parts of the body. Most *GDF6* gene mutations that cause Klippel-Feil syndrome replace single protein building blocks (amino acids) in the GDF6 protein. These mutations likely lead to a reduction in functional protein. Although the GDF6 protein is involved in bone growth and the formation of vertebrae, it is unclear how a shortage of this protein leads to incomplete separation of the cervical vertebrae in people with Klippel-Feil syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

MedlinePlus Genetics provides information about Microphthalmia

- BMP13
- CDMP2
- GDF-6
- GDF6_HUMAN
- growth/differentiation factor 6
- KFS
- KFS1
- SCDO4
- SGM1

Tests Listed in the Genetic Testing Registry

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

- 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>)

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

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

References

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Genomic Location

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

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