GDF3 Gene

Subjects: Genetics & Heredity
Contributor: Vivi Li

Growth differentiation factor 3

Keywords: genes

1. Normal Function

The *GDF3* 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 GDF3 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. While the GDF3 protein is known to be involved in bone and cartilage development, its exact role is unclear.

The GDF3 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.

2. Health Conditions Related to Genetic Changes

2.1 Klippel-Feil Syndrome

At least four mutations in the *GDF3* 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. *GDF3* gene mutations that cause Klippel-Feil syndrome replace single protein building blocks (amino acids) in the GDF3 protein. These mutations likely lead to a reduction in functional protein. Although the GDF3 protein is involved in bone growth, it is unclear how a shortage of this protein leads to incomplete separation of the cervical vertebrae in people with Klippel-Feil syndrome.

2.2 Coloboma

2.3 Microphthalmia

3. Other Names for This Gene

- GDF-3
- GDF3_HUMAN
- growth/differentiation factor 3

References

1. Levine AJ, Brivanlou AH. GDF3 at the crossroads of TGF-beta signaling. CellCycle. 2006 May;5(10):1069-73.
2. Levine AJ, Levine ZJ, Brivanlou AH. GDF3 is a BMP inhibitor that can activate Nodal signaling only at very high doses. Dev Biol. 2009 Jan 1;325(1):43-8. doi:10.1016/j.ydbio.2008.09.006.
3. Ye M, Berry-Wynne KM, Asai-Coakwell M, Sundaresan P, Footz T, French CR,Abitbol M, Fleisch VC, Corbett N, Allison WT, Drummond G, Walter MA, UnderhillTM, Waskiewicz AJ, Lehmann OJ. Mutation of the bone morphogenetic protein GDF3causes ocular and skeletal anomalies. Hum Mol Genet. 2010 Jan 15;19(2):287-98.doi: 10.1093/hmg/ddp496.