GH1 Gene

Subjects: Genetics & Heredity
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Growth hormone 1

Keywords: genes

1. Normal Function

The *GH1* gene provides instructions for making the growth hormone protein. Growth hormone is produced in the growth-stimulating somatotropic cells of the pituitary gland, which is located at the base of the brain. Growth hormone is necessary for the normal growth of the body's bones and tissues. The production of growth hormone is triggered when two other hormones are turned on (activated): ghrelin, which is produced in the stomach; and growth hormone releasing hormone, which is produced in a part of the brain called the hypothalamus. Ghrelin and growth hormone releasing hormone also stimulate the release of growth hormone from the pituitary gland. The release of growth hormone into the body peaks during puberty and reaches a low point at about age 55.

Cells in the liver respond to growth hormone and trigger the production of a protein called insulin-like growth factor-I (IGF-I). This protein stimulates cell growth and cell maturation (differentiation) in many different tissues, including bone. The production of IGF-I by the actions of growth hormone is a major contributor to the promotion of growth.

Growth hormone also plays a role in many chemical reactions (metabolic processes) in the body. By acting on specific tissues, growth hormone is involved in protein production and the breakdown (metabolism) of fats and carbohydrates.

2. Health Conditions Related to Genetic Changes

2.1 Isolated Growth Hormone Deficiency

More than 70 mutations in the *GH1* gene have been found to cause isolated growth hormone deficiency, a condition characterized by slow growth and short stature. Mutations that prevent the production of growth hormone, such as a deletion of a large section of the gene, result in isolated growth hormone deficiency type IA and severe growth failure by age 6 months. *GH1* gene mutations that cause type IB are found throughout the gene and allow some growth hormone to be produced.

Most *GH1* gene mutations that cause isolated growth hormone deficiency type II occur in a part of the gene called intron 3. These mutations result in the production of growth hormone that is shorter than normal. This short version of growth hormone is not released from cells and is not available to work in the body. In addition, this shorter growth hormone interferes with the function of normal growth hormone, so there is very little functional growth hormone available for use. A decrease in usable growth hormone results in the signs and symptoms of isolated growth hormone deficiency type II.

3. Other Names for This Gene

- GH
- GH-N
- GHN
- hGH-N
- pituitary growth hormone
- SOMA_HUMAN

- somatotrophin (ST)
- somatotropin
- somatotropin (ST)

References

1. Alatzoglou KS, Dattani MT. Genetic causes and treatment of isolated growthhormone deficiency-an update. Nat Rev En docrinol. 2010 Oct;6(10):562-76. doi:10.1038/nrendo.2010.147. Review.
2. Alatzoglou KS, Turton JP, Kelberman D, Clayton PE, Mehta A, Buchanan C, AylwinS, Crowne EC, Christesen HT, Hert el NT, Trainer PJ, Savage MO, Raza J, BanerjeeK, Sinha SK, Ten S, Mushtaq T, Brauner R, Cheetham TD, Hindmarsh PC, Mullis PE,Dattani MT. Expanding the spectrum of mutations in GH1 and GHRHR: geneticscreening in a large coho rt of patients with congenital isolated growth hormonedeficiency. J Clin Endocrinol Metab. 2009 Sep;94(9):3191-9. doi:1 0.1210/jc.2008-2783.
3. Hamid R, Phillips JA 3rd, Holladay C, Cogan JD, Austin ED, Backeljauw PF,Travers SH, Patton JG. A molecular basis f or variation in clinical severity ofisolated growth hormone deficiency type II. J Clin Endocrinol Metab. 2009Dec;94(12):4 728-34. doi: 10.1210/jc.2009-0746.
4. Mullis PE. Genetic control of growth. Eur J Endocrinol. 2005 Jan;152(1):11-31.Review.
5. Mullis PE. Genetics of isolated growth hormone deficiency. J Clin Res Pediatr Endocrinol. 2010;2(2):52-62. doi: 10.427 4/jcrpe.v2i2.52.