

GH1 gene

growth hormone 1

Normal Function

The *GH1* gene provides instructions for making the growth hormone protein. Growth hormone is produced in the growth-stimulating somatotrophic 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.

Health Conditions Related to Genetic Changes

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.

Other Names for This Gene

- GH
- GH-N
- GHN
- hGH-N
- pituitary growth hormone
- SOMA_HUMAN
- somatotrophin (ST)
- somatotropin
- somatotropin (ST)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GH1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2688\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2688[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GH1%5BTIAB%5D%29+OR+%28growth+hormone+1%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

- GROWTH HORMONE 1; GH1 (<https://omim.org/entry/139250>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2688>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GH1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GH1[gene]))

References

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Genomic Location

The *GH1* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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