

GHRHR gene

growth hormone releasing hormone receptor

Normal Function

The *GHRHR* gene provides instructions for making the growth hormone releasing hormone receptor. This receptor is active (expressed) on the growth-stimulating somatotrophic cells in the pituitary gland. This gland is located at the base of the brain and produces many hormones, including growth hormone. Growth hormone is necessary for the normal growth of the body's bones and tissues. The GHRHR receptor attaches (binds) to a molecule called growth hormone releasing hormone. This binding, along with the actions of other molecules, triggers the production of growth hormone and its release from the pituitary gland.

Health Conditions Related to Genetic Changes

Isolated growth hormone deficiency

More than 20 mutations in the *GHRHR* gene have been found to cause isolated growth hormone deficiency, a condition characterized by slow growth and short stature. Mutations in the *GHRHR* gene cause some cases of isolated growth hormone deficiency type IB. Most of these mutations replace single protein building blocks (amino acids) in the receptor protein, preventing the production of a functional receptor. A nonfunctional receptor cannot effectively trigger the production of growth hormone or signal its release from the pituitary gland. As a result, little growth hormone is produced or available in the body in people with *GHRHR* gene mutations, leading to isolated growth hormone deficiency type IB.

Other Names for This Gene

- GHRFR
- GHRH receptor
- GHRHR_HUMAN
- GRF receptor
- GRFR
- growth hormone-releasing factor receptor
- growth hormone-releasing hormone receptor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GHRHR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2692\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2692[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GHRHR%5BTIAB%5D%29+OR+%28growth+hormone+releasing+hormone+receptor%5BTIAB%5D%29+OR+%28GHRH+receptor%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-RELEASING HORMONE RECEPTOR; GHRHR (<https://omim.org/entry/139191>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2692>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GHRHR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GHRHR[gene]))

References

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Genomic Location

The *GHRHR* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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