

LEPR gene

leptin receptor

Normal Function

The *LEPR* gene provides instructions for making a protein called the leptin receptor, which is involved in the regulation of body weight. The leptin receptor protein is found on the surface of cells in many organs and tissues of the body, including a part of the brain called the hypothalamus. The hypothalamus controls hunger and thirst as well as other functions such as sleep, moods, and body temperature. It also regulates the release of many hormones that have functions throughout the body.

The leptin receptor is turned on (activated) by a hormone called leptin that attaches (binds) to the receptor, fitting into it like a key into a lock. Normally, the body's fat cells release leptin in proportion to their size. As fat cells become larger, they produce more leptin. This rise in leptin indicates that fat stores are increasing. In the hypothalamus, the binding of leptin to its receptor triggers a series of chemical signals that affect hunger and help produce a feeling of fullness (satiety).

Health Conditions Related to Genetic Changes

Leptin receptor deficiency

At least 18 *LEPR* gene mutations that cause leptin receptor deficiency have been identified; this disorder is associated with excessive hunger, massive weight gain, and reduced production of hormones that direct sexual development (hypogonadotropic hypogonadism). Some of the mutations result in less receptor protein getting to the cell surface where leptin binding takes place. The receptors that get to the cell surface may bind to leptin, but their signaling function is impaired. The resulting shortage of leptin signaling disrupts normal feelings of hunger and satiety, leading to extreme weight gain.

Because hypogonadotropic hypogonadism occurs in leptin receptor deficiency, researchers suggest that leptin receptor signaling is also involved in regulating the body's response to hormones that control sexual development, and that this response is affected by *LEPR* gene mutations. However, the mechanism of this effect is unknown.

Other Names for This Gene

- LEP-R

- LEPR_HUMAN
- OB receptor
- OB-R
- OBR

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LEPR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3953\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3953[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LEPR%5BTI%5D%29+OR+%28leptin+receptor%5BTI%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+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- LEPTIN RECEPTOR; LEPR (<https://omim.org/entry/601007>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3953>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LEPR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LEPR[gene]))

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Genomic Location

The *LEPR* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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