LEP gene

leptin

Normal Function

The *LEP* gene provides instructions for making a hormone called leptin, which is involved in the regulation of body weight. Normally, the body's fat cells release leptin in proportion to their size. As fat accumulates in cells, more leptin is produced. This rise in leptin indicates that fat stores are increasing.

Leptin attaches (binds) to and activates a protein called the leptin receptor, fitting into the receptor like a key into a lock. 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. 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

Congenital leptin deficiency

At least seven *LEP* gene mutations that cause congenital leptin 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). The *LEP* gene mutations that cause congenital leptin deficiency lead to an absence of leptin. As a result, the signaling that triggers feelings of satiety does not occur, leading to the excessive hunger and weight gain associated with this disorder. Because hypogonadotropic hypogonadism occurs in congenital leptin deficiency, researchers suggest that leptin signaling is also involved in regulating the hormones that control sexual development. However, the specifics of this involvement and how it may be altered in congenital leptin deficiency are unknown.

Other Names for This Gene

- LEP_HUMAN
- LEPD
- leptin (murine obesity homolog)

- leptin (obesity homolog, mouse)
- OB
- obese protein
- obese, mouse, homolog of
- obesity factor
- OBS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LEP (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3952[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LEP%5BTI%5D%29+OR+%28leptin%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+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- LEPTIN; LEP (https://omim.org/entry/164160)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3952)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=LEP[gene])

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Genomic Location

The *LEP* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosom

e/7/).

Last updated December 1, 2013