LIFR gene

LIF receptor alpha

Normal Function

The *LIFR* gene provides instructions for making the leukemia inhibitory factor receptor (LIFR) protein. This receptor spans the cell membrane, which allows it to attach (bind) to other proteins, called ligands, outside the cell and send signals inside the cell that help the cell respond to its environment. Ligands and receptors fit together like keys into locks.

LIFR acts as a receptor for a molecule known as leukemia inhibitory factor (LIF) as well as other ligands. LIFR signaling can control several cellular processes, including growth and division (proliferation), maturation (differentiation), and survival. First found to be important in blocking (inhibiting) growth of blood cancer (leukemia) cells, this signaling is also involved in the formation of bone and the development of nerve cells. It appears to play an important role in normal development and functioning of the autonomic nervous system, which controls involuntary body processes such as the regulation of breathing rate and body temperature.

Health Conditions Related to Genetic Changes

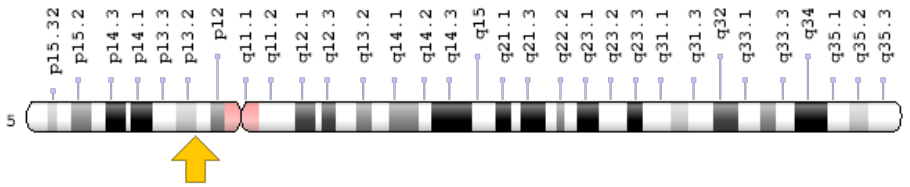
Stüve-Wiedemann syndrome

At least 27 mutations in the *LIFR* gene have been found to cause Stüve-Wiedemann syndrome. This severe condition, characterized by abnormally curved (bowed) legs, breathing problems, and episodes of dangerously high body temperature (hyperthermia), is often life-threatening in infancy. Most *LIFR* gene mutations involved in Stüve-Wiedemann syndrome prevent the production of any LIFR protein. Other mutations lead to production of an altered protein that likely cannot function. Without functional LIFR, signaling is impaired. The lack of LIFR signaling disrupts normal bone growth, leading to low bone mineral density (osteopenia), bowed legs, and other skeletal problems common in Stüve-Wiedemann syndrome. In addition, development of nerve cells, particularly those involved in the autonomic nervous system, is abnormal, leading to the problems with breathing, feeding, and regulating body temperature characteristic of this condition.

Chromosomal Location

Cytogenetic Location: 5p13.1, which is the short (p) arm of chromosome 5 at position 13.1

Molecular Location: base pairs 38,474,668 to 38,606,290 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CD118
- CD118 antigen
- leukemia inhibitory factor receptor alpha
- leukemia inhibitory factor receptor precursor
- LIF-R
- LIF receptor
- SJS2
- STWS
- SWS

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LIFR%5BTIAB%5D%29+OR+%28leukemia+inhibitory+factor+receptor+alpha%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

- LEUKEMIA INHIBITORY FACTOR RECEPTOR
  http://omim.org/entry/151443

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/LIFRID410ch5p13.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LIFR%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6597
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3977
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3977
- UniProt
  https://www.uniprot.org/uniprot/P42702

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