Genetics
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Your Guide to Understanding Genetic Conditions

CSF1R gene
colony stimulating factor 1 receptor

Normal Function
The *CSF1R* gene provides instructions for making a protein called the colony stimulating factor 1 receptor (CSF-1 receptor). This protein is found in the outer membrane of certain cell types. When a specific protein called colony stimulating factor 1 attaches (binds) to it, the receptor turns on (activates) a series of proteins inside the cell that are part of multiple signaling pathways. The signaling pathways stimulated by the CSF-1 receptor control many important cellular processes such as cell growth and division (proliferation) and maturation of cells to take on specific functions (differentiation).

In the brain, the CSF-1 receptor is abundant in the membrane of specialized cells called glial cells. These cells protect and maintain nerve cells (neurons). The CSF-1 receptor is thought to be involved in the proliferation and differentiation of glial cells, but its exact role in the brain is unclear.

Health Conditions Related to Genetic Changes
Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia

More than a dozen mutations in the *CSF1R* gene have been found in people with adult-onset leukoencephalopathy with axonal spheroids and pigmented glia (ALSP). ALSP is a severe neurological disorder characterized by damage to a type of brain tissue called white matter. Symptoms of this condition typically begin in adulthood and progress to severe cognitive and movement problems. Most *CSF1R* gene mutations in ALSP change single protein building blocks (amino acids) in the CSF-1 receptor. Other mutations change the sequence of amino acids in other ways. The mutations all occur in the region of the receptor that activates other proteins (called the kinase domain). It is likely that the altered receptor is unable to stimulate cell signaling pathways. However, it is unclear how the gene mutations lead to white matter damage or cognitive and movement problems in people with ALSP.
Chromosomal Location

Cytogenetic Location: 5q32, which is the long (q) arm of chromosome 5 at position 32
Molecular Location: base pairs 150,053,291 to 150,113,372 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• C-FMS
• CD115
• CD115 antigen
• CSF-1 receptor
• CSF-1R
• CSF1R_HUMAN
• CSFR
• FIM2
• FMS
• FMS proto-oncogene
• M-CSF-R
• macrophage colony-stimulating factor 1 receptor
• macrophage colony stimulating factor I receptor
• McDonough feline sarcoma viral (v-fms) oncogene homolog
• proto-oncogene c-Fms

Additional Information & Resources

Educational Resources

• Madame Curie Bioscience Database (2000): CSF-1
  https://www.ncbi.nlm.nih.gov/books/NBK6056/#A31637
Clinical Information from GeneReviews

- CSF1R-Related Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia
  https://www.ncbi.nlm.nih.gov/books/NBK100239

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CSF1R%5BTIAB%5D%29+OR+%28colony+stimulating+factor+1+receptor%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- COLONY-STIMULATING FACTOR 1 RECEPTOR
  http://omim.org/entry/164770

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/CSF1RID40161ch5q32.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CSF1R%5Bgene%5D
- HGNC Gene Family: CD molecules
  https://www.genenames.org/cgi-bin/genefamilies/set/471
- HGNC Gene Family: Immunoglobulin like domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/594
- HGNC Gene Family: Receptor tyrosine kinases
  https://www.genenames.org/cgi-bin/genefamilies/set/321
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2433
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1436
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P07333
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15062647

- OMIM: COLONY-STIMULATING FACTOR 1 RECEPTOR
  http://omim.org/entry/164770

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23052599

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23038421

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22197934 
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3267847/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/2153029

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