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.

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 I receptor
- macrophage colony-stimulating factor 1 receptor
- McDonough feline sarcoma viral (v-fms) oncogene homolog
- proto-oncogene c-Fms

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CSF1R (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1436[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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%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; CSF1R (https://omim.org/entry/164770)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1436)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CSF1R[gene])

References

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Genomic Location

The *CSF1R* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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