OSMR gene

oncostatin M receptor

Normal Function

The *OSMR* gene provides instructions for making a protein called oncostatin M receptor beta subunit (OSMRβ). This protein is one piece (subunit) of both the oncostatin M (OSM) receptor type II and the interleukin-31 (IL-31) receptor. These receptors are embedded in the cell membrane of many types of cells throughout the body. Each attaches to a particular protein, fitting together like a lock and its key. This attachment triggers a series of chemical signals inside the cell that directs certain cell functions.

OSM receptor type II interacts with a protein called oncostatin M (OSM). Signaling triggered by OSM was first recognized to block the growth of cancerous cells and appears to play a role in many other body processes, including the development of blood cells, the maturation of cells to become certain cell types, and an immune system response called inflammation. The signaling may also block the self-destruction (apoptosis) of cells.

The IL-31 receptor interacts with a protein called IL-31. Signaling triggered by IL-31 is involved in inflammation and stimulating itching (pruritus), although its role is not completely understood.

Health Conditions Related to Genetic Changes

Primary localized cutaneous amyloidosis

At least 13 mutations in the *OSMR* gene have been found to cause primary localized cutaneous amyloidosis (PLCA) type 1, an itchy skin condition in which clumps of abnormal proteins called amyloids build up in the skin. These mutations change single protein building blocks (amino acids) in OSMRβ. Most alter a region of the protein thought to interact with the other subunit of the OSM receptor type II or the IL-31 receptor and may impair formation of these receptors.

OSMR gene mutations reduce the chemical signals triggered by OSM and IL-31. Researchers speculate that this reduced signaling may make cells more likely to undergo apoptosis. Some studies suggest that apoptosis of skin cells releases abnormal proteins that form amyloids. It has been suggested that apoptosis is triggered by scratching the itchy skin, but the role of *OSMR* gene changes in skin itching is not

clear.

Other Names for This Gene

- IL-31 receptor subunit beta
- IL-31R subunit beta
- IL-31R-beta
- IL-31RB
- interleukin-31 receptor subunit beta
- oncostatin-M specific receptor beta subunit
- OSMRB
- PLCA1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OSMR (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9180[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28OSMR%5BTIAB%5D%29+OR+%28oncostatin+M+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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ONCOSTATIN M RECEPTOR; OSMR (https://omim.org/entry/601743)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9180)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=OSMR[gene])

References

- Arita K, South AP, Hans-Filho G, Sakuma TH, Lai-Cheong J, Clements S, OdashiroM, Odashiro DN, Hans-Neto G, Hans NR, Holder MV, Bhogal BS, Hartshorne ST,Akiyama M, Shimizu H, McGrath JA. Oncostatin M receptor-beta mutations underliefamilial primary localized cutaneous amyloidosis. Am J Hum

Genet. 2008Jan;82(1):73-80. doi: 10.1016/j.ajhg.2007.09.002. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18179886) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2253984/)

- Chang YT, Wong CK, Chow KC, Tsai CH. Apoptosis in primary cutaneousamyloidosis. Br J Dermatol. 1999 Feb;140(2):210-5. doi:10.1111/j.1365-2133.1999.02651.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10733268)
- Hermanns HM. Oncostatin M and interleukin-31: Cytokines, receptors, signaltransduction and physiology. Cytokine Growth Factor Rev. 2015 Oct;26(5):545-58.doi: 10.1016/j.cytogfr.2015.07.006. Epub 2015 Jul 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26198770)
- Lin MW, Lee DD, Liu TT, Lin YF, Chen SY, Huang CC, Weng HY, Liu YF, Tanaka A, Arita K, Lai-Cheong J, Palisson F, Chang YT, Wong CK, Matsuura I, McGrath JA, Tsai SF. Novel IL31RA gene mutation and ancestral OSMR mutant allele in familialprimary cutaneous amyloidosis. Eur J Hum Genet. 2010 Jan;18(1):26-32. doi: 10.1038/ejhg.2009.135. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19690585) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2987153/)
- Wali A, Liu L, Takeichi T, Jelani M, Rahman OU, Heng YK, Thng S, Lee J,Akiyama M, McGrath JA, Betz RC. Familial primary localized cutaneous amyloidosisresults from either dominant or recessive mutations in OSMR. Acta Derm Venereol.2015 Nov;95(8):1005-7. doi: 10.2340/00015555-2104. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25792357)

Genomic Location

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

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