



NLRP12 gene

NLR family pyrin domain containing 12

Normal Function

The *NLRP12* gene provides instructions for making a protein called monarch-1. Monarch-1 is a member of a family of proteins called nucleotide-binding domain and leucine-rich repeat containing (NLR) proteins, which are found in the fluid inside cells (cytoplasm). Monarch-1 is found mainly in certain types of white blood cells.

NLR proteins are involved in the immune system, helping to regulate the immune system's response to injury, toxins, or invasion by microorganisms. Unlike most NLR proteins that promote increased activity by the immune system, monarch-1 stops (inhibits) the release of certain molecules that are involved in the process of inflammation.

Inflammation occurs when the immune system sends signaling molecules as well as white blood cells to a site of injury or disease to fight microbial invaders and facilitate tissue repair. When this has been accomplished, stopping the inflammatory response helps to prevent damage to the body's own cells and tissues.

Health Conditions Related to Genetic Changes

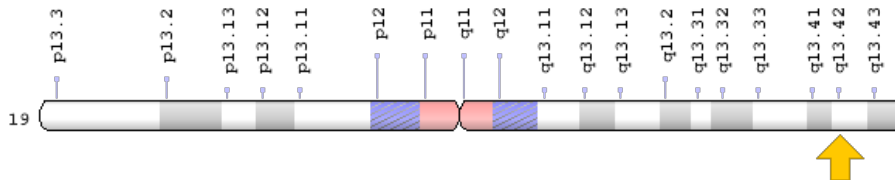
Familial cold autoinflammatory syndrome

Two mutations in the *NLRP12* gene have been identified in families with familial cold autoinflammatory syndrome from the Caribbean archipelago of Guadeloupe. These mutations appear to reduce the ability of the monarch-1 protein to inhibit the inflammatory response, resulting in the episodes of fever and inflammation seen in this disorder.

Chromosomal Location

Cytogenetic Location: 19q13.42, which is the long (q) arm of chromosome 19 at position 13.42

Molecular Location: base pairs 53,793,584 to 53,824,403 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CLR19.3
- FCAS2
- monarch 1
- Monarch1
- NACHT, leucine rich repeat and PYD containing 12
- NACHT, LRR and PYD containing protein 12
- NALP12
- NLR family, pyrin domain containing 12
- nucleotide-binding oligomerization domain, leucine rich repeat and pyrin domain containing 12
- PAN6
- PYPAF7
- PYRIN-containing APAF1-like protein 7
- regulated by nitric oxide
- RNO
- RNO2

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Innate Immunity
<https://www.ncbi.nlm.nih.gov/books/NBK10769/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NLRP12%5BTIAB%5D%29+OR+%28%28RNO%5BTIAB%5D%29+OR+%28PAN6%5BTIAB%5D%29+OR+%28RNO2%5BTIAB%5D%29+OR+%28NALP12%5BTIAB%5D%29+OR+%28PYPAF7%5BTIAB%5D%29+OR+%28monarch+1%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+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- NLR FAMILY, PYRIN DOMAIN-CONTAINING 12
<http://omim.org/entry/609648>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NLRP12.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NLRP12%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:22938
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:91662>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/91662>
- UniProt
<https://www.uniprot.org/uniprot/P59046>

Sources for This Summary

- Jéru I, Duquesnoy P, Fernandes-Alnemri T, Cochet E, Yu JW, Lackmy-Port-Lis M, Grimprel E, Landman-Parker J, Hentgen V, Marlin S, McElreavey K, Sarkisian T, Grateau G, Alnemri ES, Amselem S. Mutations in NALP12 cause hereditary periodic fever syndromes. *Proc Natl Acad Sci U S A*. 2008 Feb 5;105(5):1614-9. doi: 10.1073/pnas.0708616105. Epub 2008 Jan 29.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18230725>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2234193/>
- Macaluso F, Nothnagel M, Parwez Q, Petrasch-Parwez E, Bechara FG, Epplen JT, Hoffjan S. Polymorphisms in NACHT-LRR (NLR) genes in atopic dermatitis. *Exp Dermatol*. 2007 Aug;16(8):692-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17620097>
- OMIM: NLR FAMILY, PYRIN DOMAIN-CONTAINING 12
<http://omim.org/entry/609648>
- Touitou I, Lesage S, McDermott M, Cuisset L, Hoffman H, Dode C, Shoham N, Aganna E, Hugot JP, Wise C, Waterham H, Pugnere D, Demaille J, Sarrauste de Menthier C. Infefers: an evolving mutation database for auto-inflammatory syndromes. *Hum Mutat*. 2004 Sep;24(3):194-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15300846>
- Williams KL, Taxman DJ, Linhoff MW, Reed W, Ting JP. Cutting edge: Monarch-1: a pyrin/nucleotide-binding domain/leucine-rich repeat protein that controls classical and nonclassical MHC class I genes. *J Immunol*. 2003 Jun 1;170(11):5354-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12759408>
- Ye Z, Lich JD, Moore CB, Duncan JA, Williams KL, Ting JP. ATP binding by monarch-1/NLRP12 is critical for its inhibitory function. *Mol Cell Biol*. 2008 Mar;28(5):1841-50. Epub 2007 Dec 26.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18160710>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2258772/>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/NLRP12>

Reviewed: September 2008
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services