

NLRP12 Gene

Subjects: Genetics & Heredity

Contributor: Lily Guo

NLR family pyrin domain containing 12

Keywords: genes

1. Introduction

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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

3. 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

References

1. 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.
2. Macaluso F, Nothnagel M, Parwez Q, Petrasch-Parwez E, Bechara FG, Epplen JT, Hoffjan S. Polymorphisms in NACH-T-LRR (NLR) genes in atopic dermatitis. *Exp Dermatol*. 2007 Aug;16(8):692-8.
3. 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.
4. 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.
5. 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.

Retrieved from <https://encyclopedia.pub/entry/history/show/12699>