

## NLRP1 gene

NLR family pyrin domain containing 1

### Normal Function

The *NLRP1* gene provides instructions for making a member of a family of proteins called nucleotide-binding domain and leucine-rich repeat containing (NLR) proteins. These proteins are involved in the immune system, helping to regulate the process of inflammation. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight microbial invaders and facilitate tissue repair. The body then stops (inhibits) the inflammatory response to prevent damage to its own cells and tissues.

The NLRP1 protein is involved in the assembly of a molecular complex called an inflammasome, which helps trigger the inflammatory process in response to the presence of bacteria or viruses. Researchers believe that the NLRP1 protein may also play a role in the self-destruction of cells (apoptosis).

### Health Conditions Related to Genetic Changes

#### Vitiligo

Studies have associated variations in the *NLRP1* gene with an increased risk of vitiligo, an autoimmune condition that results in patchy changes in skin coloring (pigmentation). Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs.

One of the *NLRP1* gene variation associated with vitiligos, written as Leu155His or L155H, changes the protein building block (amino acid) leucine to the amino acid histidine at a particular location in the NLRP1 protein sequence. This and other variations likely affect the activity of the NLRP1 protein, making it more difficult for the body to control inflammation and prevent the immune system from attacking its own tissues. While the pigment loss associated with vitiligo results from the immune system attacking pigment-producing cells (melanocytes) in the skin, it is unclear what specific circumstances trigger the immune system to do so. The condition probably results from a combination of genetic and environmental factors, most of which have not been identified.

#### Autoimmune Addison disease

*NLRP1* gene variations have been associated with an increased risk of another autoimmune disorder called Addison disease, which occurs when the immune system attacks and damages the small hormone-producing glands on top of each kidney (adrenal glands). Certain *NLRP1* gene variations seem to make affected individuals more prone to overactivity of the immune system, resulting in damage to the body's own tissues and organs.

### Autoimmune disorders

*NLRP1* gene variations have been associated with an increased risk of type 1 diabetes, an autoimmune disorder in which insulin-producing cells in the pancreas are destroyed. Certain *NLRP1* gene variations seem to make affected individuals more prone to overactivity of the immune system, resulting in damage to the body's own tissues and organs.

### **Other Names for This Gene**

- CARD7
- CLR17.1
- DEFCAP
- DKFZp586O1822
- KIAA0926
- NAC
- NALP1
- NALP1\_HUMAN
- NLR family, pyrin domain containing 1
- SLEV1
- VAMAS1

### **Additional Information & Resources**

#### Tests Listed in the Genetic Testing Registry

- Tests of NLRP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=22861\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=22861[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NLRP1%5BTIAB%5D%29+OR+%28CARD7%5BTIAB%5D%29+OR+%28NALP1%5BTIAB%5D%29+OR+%28SLEV1%5BTIAB%5D%29+OR+%28DEFCAP%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>)

## Catalog of Genes and Diseases from OMIM

- NLR FAMILY, PYRIN DOMAIN-CONTAINING 1; NLRP1 (<https://omim.org/entry/606636>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/22861>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NLRP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NLRP1[gene]))

## **References**

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## Genomic Location

The *NLRP1* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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