

NLRP1 Gene

Subjects: Genetics & Heredity

Contributor: Lily Guo

NLR family pyrin domain containing 1

Keywords: genes

1. Introduction

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).

2. Health Conditions Related to Genetic Changes

2.1. 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).

One of the *NLRP1* gene variations associated with vitiligo changes the protein building block (amino acid) leucine to the amino acid histidine at position 155 in the NLRP1 protein sequence, written as Leu155His or L155H. 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.

2.2. Autoimmune disorders

NLRP1 gene variations have been associated with an increased risk of autoimmune disorders. Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs. These disorders include type 1 diabetes, in which insulin-producing cells in the pancreas are destroyed, and Addison disease, which is caused by autoimmune damage to 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.

Other Names for This Gene

- CARD7
- CLR17.1
- DEFCAP
- DKFZp586O1822

- KIAA0926
- NAC
- NALP1
- NALP1_HUMAN
- NLR family, pyrin domain containing 1
- SLEV1
- VAMAS1

References

1. Jin Y, Birlea SA, Fain PR, Spritz RA. Genetic variations in NALP1 are associated with generalized vitiligo in a Romanian population. *J Invest Dermatol*. 2007 Nov;127(11):2558-62.
2. Jin Y, Mailloux CM, Gowan K, Riccardi SL, LaBerge G, Bennett DC, Fain PR, Spritz RA. NALP1 in vitiligo-associated multiple autoimmune disease. *N Engl J Med*. 2007 Mar 22;356(12):1216-25.
3. Jin Y, Riccardi SL, Gowan K, Fain PR, Spritz RA. Fine-mapping of vitiligo susceptibility loci on chromosomes 7 and 9 and interactions with NLRP1 (NALP1). *J Invest Dermatol*. 2010 Mar;130(3):774-83. doi: 10.1038/jid.2009.273.
4. Pontillo A, Brandao L, Guimaraes R, Segat L, Araujo J, Crovella S. Two SNPs in NLRP3 gene are involved in the predisposition to type-1 diabetes and celiac disease in a pediatric population from northeast Brazil. *Autoimmunity*. 2010 Dec;43(8):583-9. doi: 10.3109/08916930903540432.
5. Smith AG, Sturm RA. Multiple genes and locus interactions in susceptibility to vitiligo. *J Invest Dermatol*. 2010 Mar;130(3):643-5. doi: 10.1038/jid.2009.403.
6. Spritz RA. The genetics of generalized vitiligo and associated autoimmune diseases. *Pigment Cell Res*. 2007 Aug;20(4):271-8. Review.
7. Taïeb A. NALP1 and the inflammasomes: challenging our perception of vitiligo and vitiligo-related autoimmune disorders. *Pigment Cell Res*. 2007 Aug;20(4):260-2.
8. Zurawek M, Fichna M, Januszkiewicz-Lewandowska D, Gryczyńska M, Fichna P, Nowak J. A coding variant in NLRP1 is associated with autoimmune Addison's disease. *Hum Immunol*. 2010 May;71(5):530-4. doi: 10.1016/j.humimm.2010.02.004.

Retrieved from <https://encyclopedia.pub/entry/history/show/12698>