NLRP3 Gene

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NLR family pyrin domain containing 3

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1. Introduction

The *NLRP3* gene (also known as *CIAS1*) provides instructions for making a protein called cryopyrin. Cryopyrin 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). Cryopyrin is found mainly in white blood cells and in cartilage-forming cells (chondrocytes).

NLR proteins are involved in the immune system, helping to start and regulate the immune system's response to injury, toxins, or invasion by microorganisms. These proteins recognize specific molecules, become activated, and respond by helping to engage components of the immune system. Cryopyrin recognizes bacterial particles; chemicals such as asbestos, silica, and uric acid crystals; and compounds released by injured cells.

Once activated, groups of cryopyrin molecules assemble themselves along with other proteins into structures called inflammasomes, which 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.

2. Health Conditions Related to Genetic Changes

2.1. Familial cold autoinflammatory syndrome

Several mutations in the *NLRP3* gene have been identified in people with familial cold autoinflammatory syndrome. These mutations are in a region of the gene known as exon 3. Researchers believe that the mutations cause cryopyrin to be hyperactive, leading to episodes of fever and inflammation that are usually triggered by exposure to cold.

2.2. Muckle-Wells syndrome

At least 10 mutations in exon 3 of the *NLRP3* gene have been identified in people with Muckle-Wells syndrome. These mutations are believed to cause hyperactive cryopyrin, resulting in episodes of fever and inflammation, as well as the hearing loss and kidney problems that occur in Muckle-Wells syndrome.

2.3. Neonatal onset multisystem inflammatory disease

About 30 mutations in the *NLRP3* gene have been identified in people with neonatal onset multisystem inflammatory disease (NOMID). Almost all of these mutations are found in exon 3. The mutations likely cause cryopyrin to be hyperactive, leading to an inappropriate inflammatory response that results in episodes of fever and widespread inflammatory damage to the body's cells and tissues. It is unclear why some mutations in exon 3 cause the severe symptoms of NOMID, some cause the less serious familial cold autoinflammatory syndrome, and others cause Muckle-Wells syndrome, which is intermediate in severity.

3. Other Names for This Gene

- AGTAVPRL
- AII

- AII/AVP
- AII/AVP receptor-like
- angiotensin/vasopressin receptor AII/AVP-like
- AVP
- C1orf7
- CIAS1
- CLR1.1
- cryopyrin
- FCAS
- FCU
- FLJ95925
- MWS
- NACHT domain-, leucine-rich repeat-, and PYD-containing protein 3
- NACHT, LRR and PYD containing protein 3
- NALP3
- NALP3_HUMAN
- NLR family, pyrin domain containing 3
- nucleotide-binding oligomerization domain, leucine rich repeat and pyrin domain containing 3
- PYPAF1
- PYRIN-containing APAF1-like protein 1

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