NLRP7 gene

NLR family pyrin domain containing 7

Normal Function

The *NLRP7* gene provides instructions for making a protein whose role is not known. The NLRP7 protein is thought to be involved in regulating gene activity (expression) through a phenomenon known as genomic imprinting. Through genomic imprinting, certain genes are turned off (inactivated) based on which parent the copy of the gene came from. For most genes, both copies of the gene (one copy inherited from each parent) are active in all cells. However, for a small subset of genes, only one of the two copies is active and the other is turned off. For some of these genes, the copy from the father is normally active, while for others, the copy from the mother is normally active.

Research suggests that the NLRP7 protein also plays a role in egg cell (oocyte) and embryonic development as well as inflammation and other immune responses by regulating the release of an immune protein called interleukin-1 beta.

Health Conditions Related to Genetic Changes

Recurrent hydatidiform mole

More than 75 mutations in the *NLRP7* gene have been found to cause a pregnancy-related condition called recurrent hydatidiform mole. A hydatidiform mole is a mass that forms early in pregnancy and is made up of cells from an abnormally developed embryo and placenta. The placenta, a structure in the uterus that normally provides nutrients to a growing fetus, is dysfunctional and appears as numerous small sacs, often described as resembling a bunch of grapes. When a hydatidiform mole occurs more than once, the condition is known as recurrent hydatidiform mole. *NLRP7* gene mutations account for recurrent hydatidiform mole in about 55 percent of women with this condition.

The *NLRP7* gene mutations that cause recurrent hydatidiform mole lead to production of a protein with reduced function or prevent production of any protein at all. As a result, oocytes do not develop normally. A pregnancy that results from an abnormal oocyte cannot develop properly, resulting in recurrent hydatidiform mole. *NLRP7* gene mutations can also prevent proper imprinting of multiple genes that contribute to a developing embryo, leading to abnormal gene activity (expression). It is not clear if problems with imprinting also contribute to the development of a hydatidiform mole. In women with *NLRP7* gene mutations, a hydatidiform mole will develop in every

pregnancy that occurs with her egg cells. Additionally, *NLRP7* gene mutations result in slowed release of interleukin-1 beta. A shortage of this protein disrupts the normal immune response that would remove the hydatidiform mole from the body. Instead, the hydatidiform mole must be removed surgically.

Other disorders

Some rare and common variations (polymorphisms) in the *NLRP7* gene are associated with an increased risk for early pregnancy loss, such as miscarriages and nonrecurrent (sporadic) hydatidiform mole. The polymorphisms are found in one copy of the *NLRP7* gene in each cell. Unlike women with recurrent hydatidiform mole (described above), women with these polymorphisms are able to have normal pregnancies.

Other Names for This Gene

- CLR19.4
- NACHT, leucine rich repeat and PYD containing 7
- NACHT, LRR and PYD containing protein 7
- NACHT, LRR and PYD domains-containing protein 7
- NALP7
- NLR family, pyrin domain containing 7
- NOD12
- nucleotide-binding oligomerization domain protein 12
- nucleotide-binding oligomerization domain, leucine rich repeat and pyrin domain containing 7
- PAN7
- PYPAF3
- PYRIN-containing Apaf1-like protein 3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NLRP7 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=199713[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28NLRP7%5BTIAB%5D%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 7; NLRP7 (https://omim.org/entry/609661)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/199713)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NLRP7[gene])

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Genomic Location

The *NLRP7* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

Last updated December 1, 2018