PRF1 gene

perforin 1

Normal Function

The *PRF1* gene provides instructions for making a protein called perforin. This protein is found in immune cells (lymphocytes) called T cells and natural killer (NK) cells, which destroy other cells. Perforin is involved in the process of cell destruction (cytolysis) and the regulation of the immune system.

Perforin is a major component of structures called cytolytic granules within T cells and NK cells. One of the main ways in which T cells and NK cells destroy other cells is to transport and secrete these cytolytic granules, which contain cell-killing proteins, onto the membranes of the target cells. Perforin helps create a channel through the membrane, allowing cytolytic proteins to enter the cell and trigger it to self-destruct.

This cytolytic mechanism also helps regulate the immune system by destroying unneeded T cells. Controlling the number of T cells prevents the overproduction of immune proteins called cytokines that lead to inflammation and which, in excess, cause tissue damage.

Health Conditions Related to Genetic Changes

Familial hemophagocytic lymphohistiocytosis

More than 90 *PRF1* gene mutations have been identified in people with familial hemophagocytic lymphohistiocytosis. These mutations result in the production of a defective perforin protein or prevent the production of perforin. The resulting shortage of functional perforin prevents it from carrying out its role in cell destruction and immune system regulation, leading to the exaggerated immune response characteristic of familial hemophagocytic lymphohistiocytosis.

Cancers

People with *PRF1* gene mutations are at increased risk of developing cancers of blood-forming cells (leukemia and lymphoma). Some of these individuals also have familial hemophagocytic lymphohistiocytosis (described above). *PRF1* gene mutations impair the immune system's ability to destroy abnormal cells, allowing them to grow and divide in an uncontrolled way and leading to the development of cancer.

Other Names for This Gene

- cytolysin
- FLH2
- HPLH2
- lymphocyte pore forming protein
- lymphocyte pore-forming protein
- MGC65093
- OTTHUMP00000019759
- P1
- PERF_HUMAN
- perforin 1 (pore forming protein)
- perforin-1
- perforin-1 precursor
- PFN1
- PFP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PRF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5551[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PRF1%5BTIAB%5D%29+OR+%28perforin+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PERFORIN 1; PRF1 (https://omim.org/entry/170280)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5551)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PRF1[gene])

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Genomic Location

The *PRF1* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromos

ome/10/).

Last updated January 1, 2011