NF1 gene

neurofibromin 1

Normal Function

The *NF1* gene provides instructions for making a protein called neurofibromin. This protein is produced in many types of cells, including nerve cells and specialized cells called oligodendrocytes and Schwann cells that surround nerves. These specialized cells form myelin sheaths, which are the fatty coverings that insulate and protect certain nerve cells.

Neurofibromin acts as a tumor suppressor protein. Tumor suppressors normally prevent cells from growing and dividing too rapidly or in an uncontrolled way. This protein appears to prevent cell overgrowth by turning off another protein (called ras) that stimulates cell growth and division. Other potential functions for neurofibromin are under investigation.

Health Conditions Related to Genetic Changes

Neurofibromatosis type 1

More than 1,000 *NF1* mutations that cause neurofibromatosis type 1 have been identified. Most of these mutations are unique to a particular family. Many *NF1* mutations result in the production of an extremely short version of neurofibromin. This shortened protein cannot perform its normal job of inhibiting cell division. When mutations occur in both copies of the *NF1* gene in Schwann cells, the resulting loss of neurofibromin allows noncancerous tumors called neurofibromas to form. Research indicates that the formation of neurofibromas requires the interaction of Schwann cells with other cells, including mast cells. Mast cells are normally involved in wound healing and tissue repair.

Cholangiocarcinoma

MedlinePlus Genetics provides information about Cholangiocarcinoma

Lung cancer

MedlinePlus Genetics provides information about Lung cancer

Cancers

In rare cases, inactivation of one copy of the *NF1* gene in each cell increases the risk of developing juvenile myelomonocytic leukemia (JMML). Juvenile myelomonocytic leukemia is cancer of blood-forming tissue that usually occurs in children younger than 2. This condition causes the bone marrow to make an excessive number of immature white blood cells that cannot carry out their normal infection-fighting functions. These abnormal cells can build up in the blood and bone marrow, leaving less room for healthy white blood cells, red blood cells, and platelets. Children affected by this disorder may experience fatigue, fever, and easy bleeding or bruising.

Other Names for This Gene

- Neurofibromatosis Type 1 Protein
- Neurofibromatosis-related protein NF-1
- neurofibromin 1 (neurofibromatosis, von Recklinghausen disease, Watson disease)
- NF1 GRP
- NF1 Protein
- NF1-GAP-Related Protein
- NF1_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4763[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28NF1%5BTI%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML (https://omim.org/entry/607785)
- NEUROFIBROMIN 1; NF1 (https://omim.org/entry/613113)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4763)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NF1[gene])

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Genomic Location

The *NF1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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