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[Neurofibromatosis Type 1](#) [1]

What is neurofibromatosis type 1

Neurofibromatosis type 1 (NF1) is a hereditary condition commonly associated with multiple café-au-lait spots on the skin. Café-au-lait spots are light brown in color, like the color of “coffee with milk.” About 10% to 25% of the general population has café-au-lait spots; NF1 is suspected when a person has six or more. People with NF1 also tend to develop varying numbers of neurofibromas, meaning benign (noncancerous) tumors on the covering of the nerves. Neurofibromas are often seen as raised bumps on the skin and can occur anywhere on the body. While these skin changes do not have serious medical consequences, they can affect a person’s appearance. Plexiform neurofibromas, which form under the skin or deeper in the body, are also benign tumors. However, these can grow quite large and can cause significant medical problems, and can affect the structure of nearby bone, skin, and muscle.

Other benign and cancerous tumors that can occur in people with NF1 include:

- Benign eye tumors, called Lisch nodules, that grow on the iris of the eye and cancerous [eye tumors](#) [2], called glioma’s, that grow in the optic nerve
- [Brain tumors](#) [3]
- [Adrenal gland tumors](#) [4]
- [Muscle tumors](#) [5]

- Spinal cord tumors
- Malignant [peripheral nerve sheath tumors](#) [5] (MPNST), which is a type of sarcoma (cancer) that grows from the cells around nerve endings

Some other features of NF1 include:

- High blood pressure
- Learning disabilities which can occur in about 50% of people with NF1
- Childhood leukemia
- Bone changes
- Scoliosis, meaning curving of the spine
- Short stature (height)

Multiple features have been associated with NF1, but the overall cancer risk is low, less than 7% over a person's lifetime. The number of features present and how severe the symptoms are can vary among people with NF1, even within the same family. Sometimes NF1 is "segmental," meaning that it affects only one portion of the body, such as one leg or one arm.

What causes NF1?

NF1 is a genetic condition. This means that the cancer risk and other features of NF1 can be passed from generation to generation in a family. The gene commonly associated with NF1 is also called *NF1*. The gene is noted in italics to help distinguish the gene from the condition. A mutation (alteration) in the *NF1* gene gives a person an increased risk of developing the various symptoms of NF1, including cancerous and benign tumors. Most people with NF1 have a mutation in the *NF1* gene. It is now believed that mutations in other genes may also produce multiple nerve tumors. In particular, a different tumor suppressor gene called *INI 1* may cause multiple schwannomas, a type of tumor of the nervous system, on nerves in some patients. This is a different disease from NF1 since the main identifying feature of NF1 is neurofibroma rather than schwannomas. Both are tumors that grow on nerves, but they have different causes. Research is ongoing to learn more about NF1 and other diseases causing nerve tumors.

How is NF1 inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. NF1 follows an autosomal dominant inheritance pattern, in which a mutation happens in only one copy of the gene. This means that a parent with a gene mutation may pass along a copy of their normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having the same mutation.

Options exist for couples interested in having a child when they know that one of them carries a gene mutation that increases the risk for this hereditary cancer syndrome. Preimplantation genetic diagnosis (PGD) is a medical procedure done in conjunction with in-vitro fertilization (IVF). It allows people who carry a specific known genetic mutation to have children who do not carry the mutation. A woman's eggs are removed and fertilized in a laboratory. When the embryos reach a certain size, one cell is removed and is tested for the hereditary condition in question. The parents can then choose to transfer embryos that do not have the mutation. PGD has been in use for over a decade, and more recently has been used for several hereditary cancer predisposition syndromes. However, this is a complex procedure with financial, physical, and emotional factors for couples to consider before starting. For more information, talk with an assisted reproduction specialist at a fertility clinic.

How common is NF1?

NF1 is one of the most common genetic conditions. It is estimated that as many as one in 3,000 people has NF1. About 50% of people with NF1 do not have any family history of the condition. They have a de novo (new) mutation in the *NF1* gene. However, the children of anyone with NF1 have a higher risk of inheriting the condition.

How is NF1 diagnosed?

NF1 is diagnosed when a person meets the official diagnostic criteria for NF1 that was developed at a National Institutes of Health Consensus Conference in 1987. Based on these criteria, a person who has at least two of the following features is considered to have NF1:

- Six or more café-au-lait spots. These spots must be more than five millimeters (mm) in diameter in young children and more than 15 mm in diameter after puberty.
- Two or more neurofibromas or one plexiform neurofibroma
- Freckling around the armpits or groin

- Optic glioma, which is a tumor on the optic nerve in the brain which effects vision
- Two or more Lisch nodules, which are tumors on the iris of the eye
- Specific bone changes, including sphenoid dysplasia, which is an abnormality of one of the bones forming the skull, or thinning of the long bones
- A parent, sibling (brother or sister), or child with NF1

If a person has a family history of NF1, they are suspected of having NF1 if they have any features of the condition. Genetic testing for mutations in the *NF1* gene is available for people diagnosed with NF1.

What are the estimated cancer risks associated with NF1?

The lifetime risk of cancer developing in a person with NF1 is estimated to be about 7%. This generally happens when a benign (non-cancerous) neurofibroma turns into a cancerous form called sarcoma, which grows much faster, invades nearby structures and sometimes metastasizes (spreads) to other areas of the body.

What are the screening options for NF1?

Suggested screenings for people with NF1 or at risk for NF1 include:

- Yearly examination by a doctor who is experienced with NF1
- Yearly eye examinations, beginning in childhood. This may become less frequent in adulthood
- Yearly blood pressure monitoring
- Developmental assessments in childhood, as needed
- Other evaluations, such as imaging scans, like [CT scan](#) [6] or [MRI](#) [7], as needed for symptoms

Screening recommendations may change over time as new technologies are developed and

more is learned about NF1. It is important to talk with your doctor about appropriate screening tests. In some areas, comprehensive NF1 clinics may be available to help with coordination of medical care.

Learn more about [what to expect when having common tests, procedures, and scans](#) [8].

What are ways of treating tumors related to NF1?

Usually the recommended treatment approach for tumors associated with NF1 is to watch the patient closely for signs of tumor growth or whether he/she is having such symptoms as pain or weakness. This is called watchful waiting, watch and wait, or active surveillance. If symptoms develop over time, then [surgery](#) [9] is done to remove the tumor(s). It is usually possible to remove a tumor growing on or from nerves, and to preserve the nerve involved, unless the tumor is plexiform. Such plexiform tumors are more spread out and often get into the nerve, which makes it harder to remove the tumor without hurting the nerve. A cancerous tumor may be treated with [chemotherapy](#) [10], [radiation therapy](#) [11], or a combination of both. [Clinical trials](#) [12], meaning research studies, for neurofibromatosis are ongoing and currently focus on drugs that affect the *ras* signaling pathway inside the tumor cell. *Ras* communicates signals from outside the cell to the nucleus of the cell. Problems in the *ras* pathway are common in tumors in NF1.

Questions to ask the doctor

If you are concerned about your risk of cancerous or benign tumors, talk with your doctor. Consider asking the following questions of your doctor:

- What is my risk of developing these types of tumors?
- What can I do to reduce my risk of this?
- What are my options for screening?

If you are concerned about your family history and think you or other family members may have NF1, consider asking the following questions:

- Does my family history increase my risk of developing cancer?
- Should I meet with a genetic counselor?

- Should I consider [genetic testing](#) [13]?

More Information

[The Genetics of Cancer](#) [14]

[Genetic Testing](#) [13]

[What to Expect When You Meet With a Genetic Counselor](#) [15]

[Collecting Your Family Cancer History](#) [16]

[Sharing Genetic Test Results with Your Family](#) [17]

Additional Resources

Neurofibromatosis, Inc
www.nfinc.org [18]

Children's Tumor Foundation
www.ctf.org [19]

National Institute of Neurological Disorders and Stroke
<http://www.ninds.nih.gov/disorders/neurofibromatosis/neurofibromatosis.htm> [20]

National Cancer Institute
www.cancer.gov [21]

To find a genetic counselor in your area, ask your doctor or visit these websites:

National Society of Genetic Counselors
www.nsgc.org [22]

National Cancer Institute: Cancer Genetics Services Directory
www.cancer.gov/cancertopics/genetics/directory [23]

Links

[1] <http://www.cancer.net/cancer-types/neurofibromatosis-type-1>

[2] <http://www.cancer.net/node/31308>

[3] <http://www.cancer.net/node/31327>

[4] <http://www.cancer.net/node/31341>

[5] <http://www.cancer.net/node/31379>

[6] <http://www.cancer.net/node/24486>

- [7] <http://www.cancer.net/node/24578>
- [8] <http://www.cancer.net/node/24959>
- [9] <http://www.cancer.net/node/24462>
- [10] <http://www.cancer.net/node/24473>
- [11] <http://www.cancer.net/node/24661>
- [12] <http://www.cancer.net/node/24863>
- [13] <http://www.cancer.net/node/24895>
- [14] <http://www.cancer.net/node/24897>
- [15] <http://www.cancer.net/node/24907>
- [16] <http://www.cancer.net/node/30761>
- [17] <http://www.cancer.net/node/24906>
- [18] <http://www.nfinc.org/>
- [19] <http://www.ctf.org/>
- [20] <http://www.ninds.nih.gov/disorders/neurofibromatosis/neurofibromatosis.htm>
- [21] <http://www.cancer.gov/>
- [22] <http://www.nsgc.org/>
- [23] <http://www.cancer.gov/cancertopics/genetics/directory>