## Q Can neurofibromatosis develop into cancer?

**A** Most tumors, even large tumors, caused by NF, are benign and remain benign. However, in some cases, particularly in NF1, a tumor may become malignant. For this reason, patients with NF should have periodic check-ups by an experienced physician and need to watch for any tumor growth, sudden pain, numbness, tingling or change in function.

## Q Does NF remain stable over a lifetime or does it get worse with age?

**A** NF1, NF2 and Schwannomatosis are progressive disorders, but the rate of progression and specific complications are unpredictable. Individuals with NF1 usually will experience a gradual increase in their number of neurofibromas. Children with NF1 are also prone to the development of brain tumors that may affect vision or growth.

Individuals with NF2 will eventually develop vestibular schwannomas, and may develop other tumors, including meningiomas and other schwannomas.

Schwannomatosis is a highly variable disorder, but many people with the condition will develop painful schwannomas over the years.

## Q During childhood, are the numbers of skin neurofibromas a good way to predict how many skin neurofibromas one will have as an adult?

**A** No, the number of skin neurofibromas that appear in childhood do not predict how many tumors will develop later in life. However, skin neurofibromas are most likely to begin developing around the time of puberty.

## Q Are the number of cafe-au-lait spots an indication of how mild or severe one's experience with NF will be?

**A** No, there is no known correlation between the number of cafe-au-laits spots and severity of NF1.

## Q In women, can birth control pills worsen tumor growth? How about pregnancy? How about estrogen replacement therapy?

**A** Neurofibromas often first appear or grow during puberty and some women report that they notice an increase in tumor activity during pregnancy. However, there has been little research done to establish any link between hormones and neurofibroma growth. Since we do not know whether oral contraceptives will increase NF symptoms, it may be advisable to consider using other forms of contraception, unless the medical reasons for using the pill outweigh the theoretical reasons for not using it. Women who wish to use estrogen replacement therapy need to weigh the benefits of symptomatic relief with these theoretical risks.

## Q Can NF be cured?

**A** At the present time there is no known cure for NF. However, researchers worldwide are working towards the development of more effective treatments.

## Q What research is currently being done on NF?

**A** All three forms of neurofibromatosis are the subjects of intense research. Major advances have been made in studying the genetic basis for NF1 and NF2, resulting in the identification of both genes. These discoveries have led to ideas about therapies that will be tested over the next few years. The hope is that ultimately we will understand the normal role of the neurofibromatoses genes, determine what happens when the NF genes mutate, and how to control the abnormal functioning of the two genes.

## Q What is the Children's Tumor Foundation?

**A** The Children's Tumor Foundation, a non-profit organization, is the leading resource for information on NF. The Foundation's mission is to:

- Encourage and support research and the development of treatments and cures for neurofibromatosis types 1 and 2, Schwannomatosis and related disorders (hereafter collectively referred to as "NF");
- Support persons with NF, their families and caregivers by providing thorough, accurate, current and readily accessible information;
- Assist in the development of clinical centers, best practices and other patient support mechanisms (but not including direct medical care) to create better access to quality healthcare for affected individuals; and,
- Expand public awareness of NF to promote earlier and accurate diagnoses by the medical community, increase the non-affected population's understanding of the challenges facing persons with NF, and encourage financial and other forms of support from public and private sources.

## Q What help can the Children's Tumor Foundation provide?

**A** The Foundation can provide free information about NF; referral to NF Clinics; and referral to Chapters/Affiliates, where local patient support groups, public education programs and special events take place.

## Q How can I reach the Foundation?

**A** Call the Children's Tumor Foundation at 800-323-7938, visit its World Wide Web Site at www.ctf.org, or email to info@ctf.org.

Children's Tumor Foundation™
*Ending Neurofibromatosis Through Research*

**Children's Tumor Foundation, Inc.**
**95 Pine Street, 16th Floor**
**New York, NY 10005**
**Tel: (212) 344-6633**
**Toll Free: (800) 323-7938**
**Fax: (212) 747-0004**
**Email: info@ctf.org**
**Web Site: http://www.ctf.org**



Neurofibromatosis

# Questions & Answers

Children's Tumor Foundation™
*Ending Neurofibromatosis Through Research*

## Q What is neurofibromatosis (NF)?

**A** The neurofibromatoses (NF1, NF2 and Schwannomatosis) are a set of distinct genetic disorders. Individuals with NF1 are prone to the development of benign tumors that grow on the nerves anywhere on or in the body. NF1 can also affect other tissues, including the bones, skin, blood vessels, and eyes. In addition, nearly 50% of all people with NF1 have learning disabilities.

Individuals with NF2 are prone to the development of different benign tumors that grow on nerves anywhere on or in the body (schwannomas) as well as other nervous system tumors. In addition, NF2 commonly affects hearing, vision and balance.

Individuals with Schwannomatosis are prone to the development of benign tumors that grow on nerves anywhere on or in the body (schwannomas), but do not have the other characteristic features of NF2.

## Q Isn't neurofibromatosis the same as Elephant Man's disease?

**A** Neurofibromatosis is not Elephant Man's disease. For many years scientists believed that Joseph Merrick, the so-called "Elephant Man", had NF1, but in 1986 it was established that he had Proteus Syndrome, an extremely rare condition.

## Q Is there more than one kind of NF?

**A** There are three distinct types of NF: NF1, NF2 and Schwannomatosis. Each of these forms of NF can be readily diagnosed by an experienced clinician.

## Q What is the difference between NF1 and NF2?

**A** NF1 (also sometimes called Peripheral or von Recklinghausen NF) is characterized by 6 or more light brown ("cafe-au-lait") spots on the the skin; pea-sized bumps (neurofibromas) on or under the skin; larger areas on the skin that look "enlarged" (plexiform neurofibromas); freckling under the arms or in the groin area; spots on the iris (Lisch nodules); brain tumors (optic nerve glioma); and skeletal abnormalities such as bowing of the legs, curvature of the spine (scoliosis), or thinning of the shin bone. In addition, about 50% of people with NF1 have learning disabilities.

NF2 (sometimes called Central or Bilateral Acoustic NF) is a rarer disorder characterized by multiple tumors. The hallmark of NF2 is tumors on both of the nerves responsible for balance (vestibular nerves). Hearing loss beginning in the teens or early twenties is generally the first symptom. People may also have other tumors involving the brain and spinal cord as well as abnormalities of the lens of the eye.

## Q How many people have neurofibmatosis?

**A** Approximately 100,000 Americans have NF. NF1 affects approximately one out of every 4,000 births.

NF2 is less common, affecting approximately one out of every 40,000 births.

Schwannomatosis is probably even rarer than NF2, although prevalence of that form of NF needs further study.

## Q Does NF occur more often in men or women? Or among certain groups?

**A** NF equally affects men and women of all races and ethnic groups.

## Q Is neurofibromatosis an inherited disorder?

**A** Yes. In 50% of the cases, NF is inherited from a parent. The remaining 50% of NF cases occur as the result of a new (or spontaneous) mutation (change) in the sperm or egg cell. Moreover, a person affected by NF1 or NF2 has a 50:50 chance of passing on the disorder with every pregnancy.

Most cases of Schwannomatosis occur sporadically, but Schwannomatosis can be inherited in some families.

## Q What are the odds someone will get NF?

**A** When a parent has NF, there is a 50% chance of passing on NF with each pregnancy. When there is no family history of NF, the odds are 1 in 4,000 births for NF1 and 1 in 40,000 for NF2.

For Schwannomatosis, the risk of an affected person passing on the trait can be as high as 50%, but it is not clear that all cases are genetically transmitted and some individuals who inherit the gene for Schwannomatosis do not seem to display the trait. More research needs to be done on the genetics of Schwannomatosis.

## Q Will a parent's case of NF predict what his or her child with NF will experience?

**A** The type of NF (i.e. NF1, NF2, or Schwannomatosis) inherited by the child is always the same as that of the affected parent. However, the number and severity of the manifestations of NF differs from person to person, even within the same family. For example, a parent with NF1 who has cafe-au-lait spots, an optic glioma, and neurofibromas on the arms and face may have a daughter who only develops cafe-au-lait spots and scoliosis. The reverse case is possible as well, with a mildly affected parent who has a child with more severe symptoms.

## Q How does someone know if they have NF?

**A** The signs of NF1 usually appear in childhood or adolescence. However, it is possible for someone to show no signs until later in life. In NF2, signs generally appear in the 20s, but may also occur earlier or later in life. The following are signs for NF1 and NF2:

### NF1

If two or more of the following are present, a diagnosis of NF1 is confirmed:

- Family history of NF1
- 6 or more light brown ("cafe-au-lait") spots on the skin
- Freckling under the arms or in the groin area
- Small pigmented bumps on the eye's iris (Lisch nodules)
- Presence of pea-sized bumps (neurofibromas) on the skin
- Larger areas on the skin that look enlarged (plexiform neurofibromas)
- Optic Glioma
- Skeletal abnormalities such as bowing of the legs, curvature of the spine (scoliosis), or thinning of the shin bone

### NF2

If one or more of the following are present, a diagnosis of NF2 is likely:

- Family history of NF2
- Tumors on both auditory nerves which may cause deafness, ringing in the ears, or balance problems
- Tumors of the brain, spinal cord or meninges
- Pre-senile cataracts

### Schwannomatosis

- Presence of multiple schwannomas without other signs of NF2, especially vestibular schwannoma.

## Q What should someone do if they think they have NF?

**A** People who suspect they have NF should see a doctor knowledgeable about NF and discuss their symptoms and concerns. They can obtain more information about NF including NF clinic referrals from the Children's Tumor Foundation at 800-323-7938 or via the Foundation's Web Site at www.ctf.org.

## Q Is there a test for NF?

**A** The diagnosis of NF (NF1, NF2, Schwannomatosis) is made clinically, but in some instances of NF1 and NF2 it is now possible for genetic testing to be done. Genetic testing may be used to confirm a suspected diagnosis or to help with genetic counseling. In NF1, genetic testing is available, and is successful in >95% of cases.

In NF2, pre-symptomatic and prenatal testing is available for the offspring of a person known to have NF2 in about 66% of families.

The Schwannomatosis gene has not been identified, so genetic testing is not yet possible for this condition.

At the present time, genetic testing does not provide information about the severity of NF, and cannot tell us what the likelihood is of specific problems arising in any individual.

## Q Where can someone go for testing? What does it cost?

**A** Many people undergo genetic counseling and testing through a general genetics clinic. Others find it valuable to visit a dedicated NF clinic. Those interested in genetic testing should contact their physician. Information is also available from the Children's Tumor Foundation at 800-323-7938 or www.ctf.org

In NF1, the test costs about $1,200 at present.

In NF2, the test itself costs about $2,000. Additional costs will be involved with counseling and patient care before and after testing.

For both NF1 and NF2, genetic testing should always be performed in conjunction with genetic counseling.