Understanding Neurofibromatosis

What Is NF?

Neurofibromatosis is a genetic disorder that causes growths of tumors to form on nerves. These may occur anywhere in the body. NF is one of the most common genetic disorders. It occurs in every racial and ethnic group and affects both sexes equally. There are at least 100,000 people in the United States with NF. One in every 3,000 to 4,000 babies born has NF. In a city the size of Chicago, as many as 2,300 people have NF. But even though NF is relatively common, not many people have heard about it.

What Causes NF?

Neurofibromatosis is actually the name for a least two separate disorders, which can affect the nervous system as well as a number of other parts of the body. A genetic disorder is caused by an abnormal gene in your body. NF is not contagious. That means you can't catch it from other people. However, parents may pass NF on to their children. It is important to understand the two ways children are born with NF:

- You can inherit an abnormal gene that causes NF from one of your parents OR
- Something can go wrong in one of your genes before birth. This second way of getting NF is called spontaneous gene mutation. If you have a spontaneous gene mutation, you did not inherit NF, but you can pass it on to your children.

We call NF two disorders because two different genes are affected. NF-1 is caused by a change in a gene on chromosome 17. NF-2 is caused by a change in a gene on chromosome 22. Because NF-1 and NF-2 are due to changes in different genes, NF-1 and NF-2 do not occur in the same family.

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- Neurofibromatosis, Inc.
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- The Children's Tumor Foundation: Ending Neurofibromatosis Through Research

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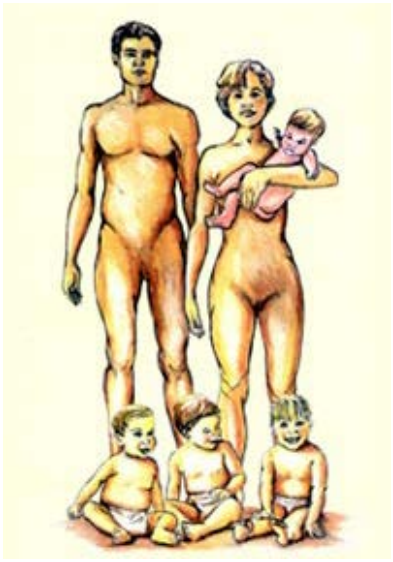
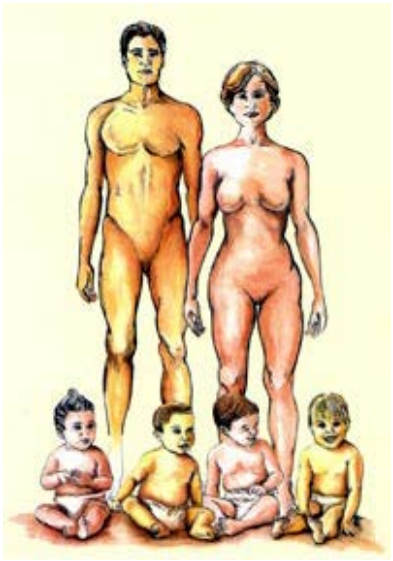


NF Can Be Caused by Spontaneous Gene Mutation

One member of this family--the baby--has NF. Since neither parent has the disease, the baby has NF due to spontaneous change in one of his or her genes. Because this is a spontaneous gene mutation, none of the baby's brothers or sisters is likely to have NF.



NF Is Dominantly Inherited

In this family, the mother has an abnormal gene that has caused NF, and two of her children have inherited this abnormal gene. Everyone has two sets of genetic information. NF is caused by an abnormal gene that dominates the normal gene. As a result, each child in this family has a 50 percent chance of inheriting their mother's abnormal gene. This is called dominant inheritance.

How Do I Know if I, or My Child, Has Neurofibromatosis?

When someone is told that they have neurofibromatosis (NF), one of the first questions is: How can you be sure? How do I know I have NF?

Right now, there is no simple blood test for NF. However, because the genes causing NF-1 and NF-2 have been identified, a simple blood test for NF should be available in the future. Until that time, a doctor who is familiar with NF should make the diagnosis of NF. Knowledge of NF is the best tool you and your doctors have in treating the disorder. A good first step is making sure you understand how a diagnosis of NF-1 or NF-2 is made. To help decide whether someone has NF-1 or NF-2, the National Institute of Health offers the following guidelines:

How Do I Know if I, or My Child, Has NF-1?

Children may have only a few signs of NF-1 and develop other problems when they are older. A person with NF-1 should have at least two of the following features:

- Six or more brown oval or circular spots on the skin, called café-au-lait spots
- Two or more benign skin tumors, called neurofibromas, or one diffuse tumor of the soft tissue or nerves, called plexiform neurofibroma
- Freckles under the arm or in the groin region
- A tumor of the nerve to the eye--called an optic glioma
- Two or more spots on the iris--called Lisch nodules
- A problem of one of the bones, such as bowing of a leg with or without a fracture
- A parent, brother, sister, or child with NF-1

How Do I Know if I, or My Child, Has NF-2?

Signs of NF-2 are usually not present until people are teenagers or older. A person with NF-2 should have either:

Tumors on both sides of the head of the nerves for hearing and balance, called vestibular schwannomas

OR

A mother, father, brother, or sister with NF-2 AND one of the following:

- A vestibular schwannoma
- Benign tumors in the brain or along the spinal cord
- A cataract at a young age

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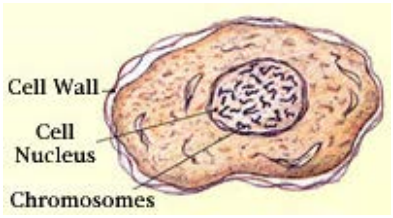
About Other Forms of NF

There are people who do not fit easily into NF-1 or NF-2, or people in whom signs of NF are located on only one side of the body. These are very uncommon forms of NF and we know less about them.

What Is a Gene?

A gene is a tiny portion of DNA that determines personal characteristics, such as eye color. Each chromosome has thousands of genes linked together like beads on a string.

What Are Chromosomes?



Chromosomes are made of chains of genes that contain all of the basic information for a person.

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NF-1

About NF-1

The most common form of neurofibromatosis (NF) is NF-1. It is sometimes referred to as peripheral neurofibromatosis or von Recklinghausen's disease. People with NF-1 generally have brown oval or circular spots on the skin called café-au-lait spots and freckles under the arm or in the groin area. Benign soft tumors or lumps in or under the skin called neurofibroma and brownish red spots in the iris--the colored part of the eye--called Lisch nodules, are present in most people.

NF-1 frequently causes learning difficulties in children. It may affect physical growth. In addition, tumors may form along nerves anywhere in the body.


Cari and her father have NF. An eighth-grader, Cari hasn't had any major problems with her NF.

Some signs of NF-1 are usually visible within the first year of life. Other signs of NF-1 may develop, as people get older. For example, Lisch nodules of the iris are unusual in young children but commonly develop in teenagers and adults. Neurofibromas frequently appear or grow during the hormonal changes that occur in teenage years and during pregnancy. It is important to remember that while the problems caused by NF-1 can be serious, NF-1 usually does not keep people who have it from living a normal and productive life.

Some people with NF-1 will only have café-au-lait spots and neurofibroma, but others may have more difficult problems. At the present time, it is impossible to predict what kinds of problems an individual will have. No two people will be affected in exactly the same way, even within the same family.

Specific Effects of NF-1

While NF-1 can affect almost any organ in the body, many people have only a few difficulties.

People with NF-1 usually have normal intelligence, but as many as 40 to 60 percent of children have short attention span, hyperactivity, or some difficulty learning in school. Problems with visual perception are common and may make spelling and arithmetic more difficult. Children with these problems can be helped to be successful in school. Headaches and difficulties with hearing are also common in NF-1 and may affect schoolwork.

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NF-1 frequently affects growth. People with NF-1 may be shorter or have slightly larger heads than the average person. NF-1 may also affect the growth of bones. A few people will have actual shrinkage of a bone--called atrophy--bending, or fracture of a long bone that won't heal called pseudarthrosis or curvature of the spine called scoliosis.

Jenna has NF-1 and had an optic glioma, a tumor that threatened her vision. Comer Children's Hospital specialists in neurology and oncology worked with Jenna's community physicians to create a comprehensive treatment plan to save her sight. »Read Jenna's story

NF-1 also causes tumors to form in different parts of the body. Small tumors on or below the surface of the skin called neurofibroma present in most people. Some people with NF-1 have tumors or growths that involve the skin and deeper tissues including the nerves called plexiform neurofibroma. These growths can sometimes be large and change the normal shape of a part of the body or can affect internal organs. Occasionally, a cancer can form in a plexiform neurofibroma.

NF-1 may also affect the brain. Bright spots are frequently seen on brain scans. The nerves to the eye may be abnormally large, called optic gloom. In a small number of people, NF-1 causes brain tumors.

Having NF-1 means that there will be changes in your body which nobody can predict. Because doctors cannot tell in advance what will happen to each person and because many of the problems are rare, it is important for you to learn all that you can about NF.

What to Do If You, or Your Child, Has NF-1

People with NF-1 will need regular physical exams to check their vision, blood pressure, and spine to evaluate any new problems or changes. Sudden changes in the size of a neurofibroma, pain, or weakness should be brought to your doctor's attention immediately.

In young children, special attention should be given to growth, hearing, vision, development, long bones, and spine. In addition, parents should ask teachers whether their child is having any trouble with schoolwork and whether a special learning evaluation is needed.

In older children and young adults, plexiform neurofibromas need to be checked carefully. Time needs to be set aside to talk about self-esteem and relationships with friends. Young adults, as well as teenagers, should understand that they could pass NF on to their children.

Special scans of the brain, eyes, and spine are often helpful in NF-1.

Because NF-1 causes spots, as well as small tumors, on the skin that people can see, people with NF-1 may have to be braver than other people. Children with NF are sometimes isolated, rejected, or teased. If this occurs, the best thing to do is to talk

about NF with your family or the child's friends and teachers.

If you are a parent of a child with NF, you should realize that your child might be frightened or angry about changes in his or her body. It is not helpful for you to pretend that there is no problem. Simple, truthful and positive explanations are reassuring to children such as, "You have a problem that causes spots and some bumps on your skin. You need to see a doctor regularly to make sure everything is OK." You should try to talk more in detail with teenagers and make sure they are dealing emotionally with their disorder.

Guidelines for Children and Adults with NF-1

Ages 1 to 5 years

- Check long bones for bowing or fracture.
- Watch developmental milestones and performance in preschool.
- Have an eye exam and hearing test.
- Check blood pressure.
- Assess other family members for evidence of NF.

Ages 6 to 14 years

- Watch school performance.
- Consider tests for learning disabilities or hyperactivity.
- Check back for curvature.
- Have an eye exam and hearing test.
- Monitor height, weight, and sexual development.
- Discuss self-esteem and relationships with friends and classmates.
- Set aside time to talk about NF and how to respond to questions by friends.
- Discuss inheritance of NF and likelihood of having a child with NF.

Ages 15 to 20 years

- Monitor changes in appearance of neurofibroma.
- Continue to talk about NF, relationships with friends and self-esteem.
- Discuss inheritance of NF and likelihood of having a child with NF.

Adults

- Watch for changes in tumors, as these could be a sign of cancer.
- Monitor appearance of headaches, weakness, pain, or changes in sensation.
- Check blood pressure.

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NF-2

About NF-2

NF-2 is sometimes referred to as central neurofibromatosis or bilateral acoustic neuroma disease. NF-2 is different for each person, but because it affects nerves next to the brain or spinal cord, the problems of NF-2 can cause serious disabilities. Even so, many people with NF-2 can lead relatively normal, rewarding lives and learn to compensate for deficits they may have.

In some people signs of the disorder are detectable in childhood, but for the majority of people, NF-2 is not apparent until late teenage years or later. People with NF-2 may only have a few café-au-lait spots--brown oval or circular spots on the skin. Skin tumors are few in number and easily overlooked.

However, everyone with NF-2 has tumors affecting hearing and balance (vestibular schwannoma or acoustic neuroma). Cataracts in the lens or changes in the retina of the eye are present in most people. Tumors, which push on the brain or spinal cord (meningiomas and schwannomas), as well as tumors along the peripheral nerves (schwannomas), or tumors within the spinal cord occur in many people. These problems are serious because they can cause weakness or seizures, but the tumors are benign.

People with NF-2 should realize that while the problems they face can be frightening, there have been important advances in both treating and detection of NF-2, which offer considerable hope.

What to Do If You, or Your Child, Has NF-2

People with NF-2 need regular physical examinations to check their hearing and neurologic function. Hearing tests of pure tones, auditory evoked responses, balance tests, scans of the brain and spinal cord, and eye exams will probably be needed annually. Because NF-2 will ultimately affect your hearing in both ears, you will need to be open to several methods of communication. Sign language opens many opportunities outside of the home for the hearing impaired. It is most useful when the entire family learns to sign. Unfortunately, lip reading is often impractical for people with NF-2.

People with NF-2 may need complicated surgery on more than one occasion. Because some surgery can lead to permanent disabilities, such as loss of hearing, the timing of surgery needs to be carefully considered. Surgery should be performed only by a neurosurgical or

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Once the NF diagnosis has been made, it's important to begin a careful, organized program to spot any change in your body that could be caused by NF. Make regular visits to your doctor part of your routine.

ear, nose, and throat team with special expertise in NF-2 because the surgery can lead to complications in less experienced hands. Specially-trained therapists, audiologists, ophthalmologists, and counselors are also essential in the rehabilitation process. In some instances, surgery should be delayed because some tumors may show very little growth over many years and can be watched carefully. But it is clear that the best results are obtained when problems are identified and treated early.

People with NF-2 need to be cautious of non-standard treatments. Treatment of NF-2 should always be discussed with physicians who treat many patients with NF-2. While the problems in NF-2 can be very frightening, there is evidence of significant improvements in treatment methods, which are very encouraging and indicate real hope for patients to lead extremely functional lives. There is also a community of people who can provide advice and support.

Guidelines for People with NF-2

- Evaluate other family members for vestibular tumors.
- Discuss inheritance of NF-2.
- Learn signing or alternative forms of communication.
- Have eye exam for cataracts and retinal abnormalities.
- Have MRI of brain and entire spinal cord annually.
- Have annual hearing tests.
- Have careful neurologic exam annually.
- Discuss potential impact of NF-2 on career and family.
- Assess resources and support needed for daily living.

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After Neurofibromatosis Diagnosis: What Do You Need to Do?

Identification of the genes for neurofibromatosis (NF)--NF-1 or NF-2--offers tremendous hope for the future. For instance, identification of the NF-1 gene has helped us understand how tumors in NF-1 form. As we understand more about the gene, we hope to be able to predict and control the problems caused by the abnormal gene.

While there is no cure for NF at the present time, there are many things that you can do that will make a significant difference in your life or the lives of you children.

Remember, if your child has NF, you are your child's protector. If you, yourself, are affected, you must be your own advocate as well. This means that you must be sure that you talk to your doctor about any new problems or concerns.

There are also many people besides your doctor that you can rely on for help with NF. They include members of your local NF support group, genetic counselors, teachers, and psychologists.

Learn About NF

Because many of the problems in NF are rare, even among people with NF, most doctors will not have had enough experience with NF to detect or successfully treat all of them. Successful treatment often depends on having a cooperative relationship between your local physician and other physicians in NF programs at major medical centers.

The best treatment also depends on your learning about NF so that you can understand what problems you may expect and bring them to the attention of your doctor. If you notice changes, call the doctor and ask whether the problem could be due to NF. It is often much easier to treat the problems that occur in NF if they are detected early.

Work with Your Doctors

It is important to get the advice of a doctor who treats many patients with NF. Don't hesitate to ask your doctor about his or her experience with NF. Do not be afraid to ask for a second opinion. Your family doctor should be willing to call an NF clinic to ask for advice. You may also want to consult an NF clinic in a medical center on your own for advice. Doctors at an NF clinic can then work with your family doctor to provide you with the best care.

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Jonathon and Kevin are identical twins--both have NF. They love baseball and being in the Boy Scouts. The boys say NF hasn't stopped them from doing anything.

Once a diagnosis of NF has been made, it is important to begin a careful, organized program to spot any changes, which may be due to NF and to keep good records about these problems. Ask doctors for a copy of the examination report to keep for your own records. You may also want to keep copies of your X-rays or scans so that you can discuss them with different doctors. Make regular visits to the doctor as part of your routine.

Evaluate Your Family

It is important to know whether other people in your immediate family have NF. Identifying who in a family is affected may be a sensitive and difficult issue because some people have feelings of guilt or shame associated with having a genetic disorder. However, there are good reasons for knowing who has NF. If, for example, you have NF, then any of your children can have NF. On the other hand, if your child had NF and you are sure that neither of the parents is affected, then the chance of having another child with NF is very small.

The best way of determining if someone has NF is by careful physical examination, although blood tests may eventually be available. You can use this information in planning a family. You may want to discuss this with your doctor or a genetic counselor. Genetic counselors are trained to provide people with information about genetic disorders, family planning, and the risk of passing on a disorder.

Talk About Your Concerns

Don’t be afraid to talk about NF in your family. You may also want to talk to your friends about your concerns. Be prepared for the possibility that your friends or family may not be as understanding or as supportive as you might hope. They may not be ready to talk or they may not want to believe there is a problem or may not know what to say. Some family members may feel guilty or angry about having the disorder.

Unfortunately, there are some situations in which you will need to be cautious about discussing NF. Employers may discriminate against people with NF, even against parents of children with NF, because of potentially higher health insurance costs. Schools may be unsure how to treat a child with NF unless they are given specific directions. Offer information about NF to other people, as it seems necessary. You may also need to be careful not to overemphasize NF so that affected family members feel different from the rest of the family. Remember, that the majority of people with NF can lead normal, functional lives.

Accepting that you or your child has NF may take time and it is important to understand that. Many other families have gone through similar problems. Local NF groups can be source of tremendous support and advice. There are NF support groups in many states. These organizations were formed by parents and patients to provide support, advice, and information on dealing

with many aspects of NF. They are important advocates for patients.

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