



## NEWLY DIAGNOSED WITH NF1: A GUIDE TO THE BASICS

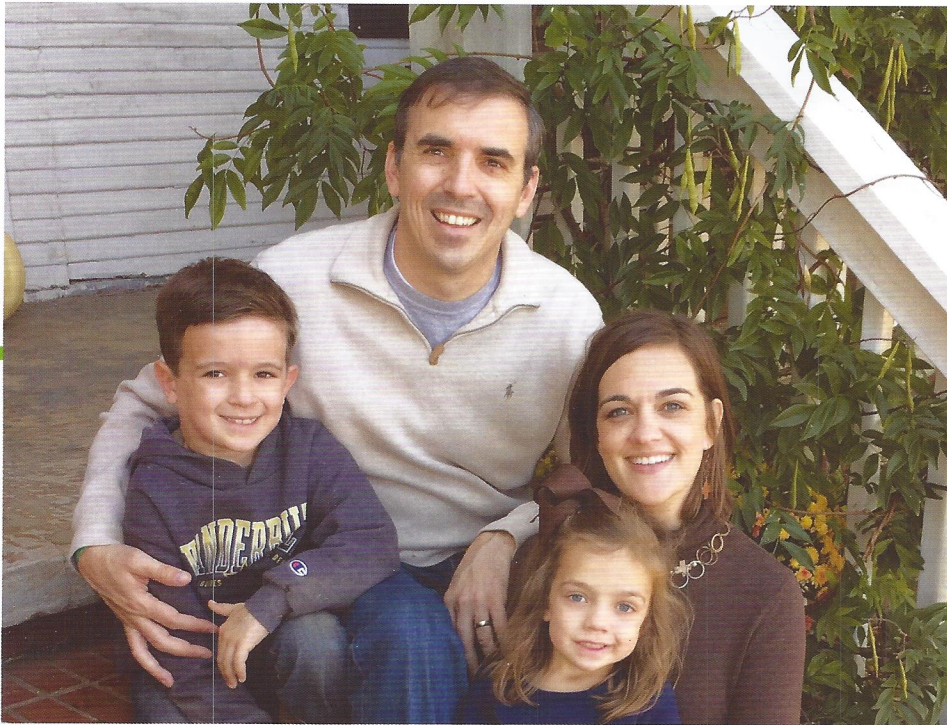
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CHILDREN'S  
TUMOR  
FOUNDATION  
ENDING NF  
THROUGH RESEARCH

# CONTENTS



Newly Diagnosed? You Are Not Alone	2
The Children's Tumor Foundation	4
NF Basics	5
How Does Someone Get NF1?	7
How Is the Diagnosis Made?	8
What Are the Possible Symptoms of NF1?	10
Medical Management of NF1	11
How to Share the News	13
Sample Message to Friends and Family	14
Potential Learning Challenges Associated with NF1	15
Insurance Coverage for Neurofibromatosis	17
Resources	18



## NEWLY DIAGNOSED? You Are Not Alone

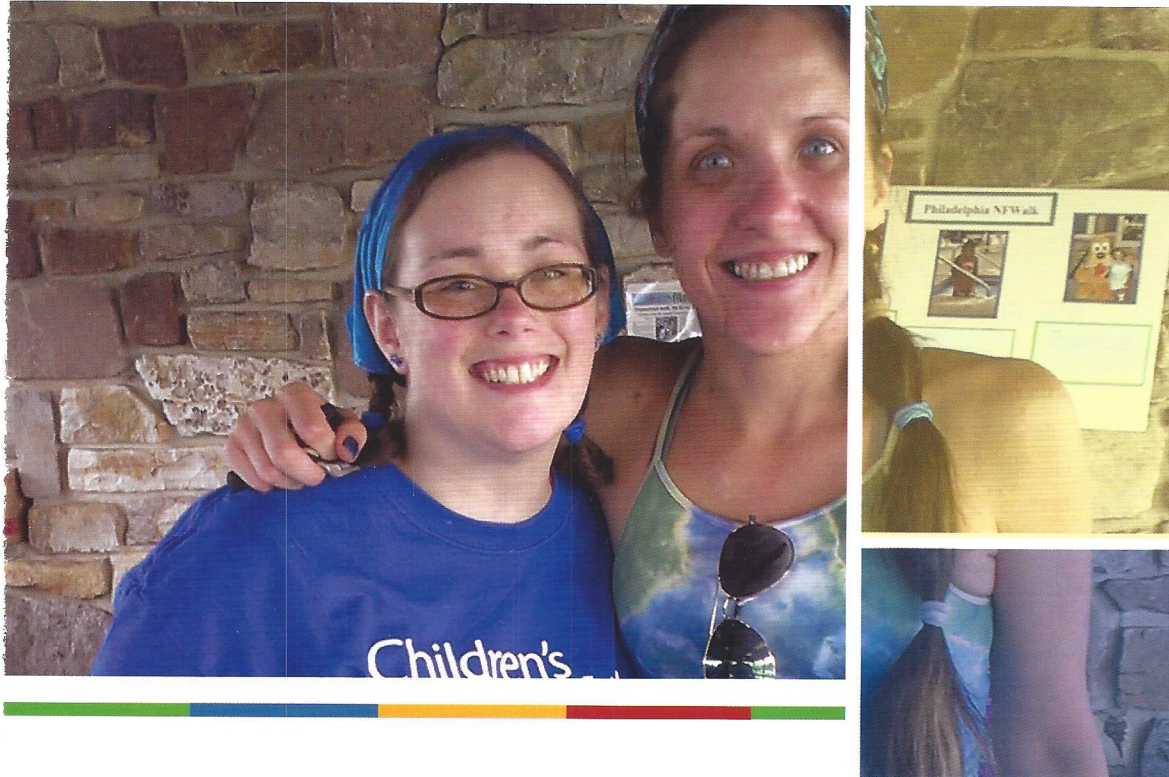
At the Children's Tumor Foundation (CTF), we know that receiving a diagnosis of neurofibromatosis (NF) can be overwhelming and a lot to digest all at once. Everyone deals with difficult news in different ways. Some like to take the information piece by piece so as not to be overwhelmed and so that they can have time to properly absorb everything. Others prefer to delve in immediately and get as much information as they can. Both are perfectly normal approaches.

People also have different emotional reactions to being diagnosed with neurofibromatosis. Some may become anxious, overwhelmed, depressed, or feel a sense of loss or shock from the unexpected news. Instead of fighting and trying to take control of the unpredictable, one of the most helpful mental strategies can be to simply try to accept the diagnosis. Once acceptance begins, it becomes much easier for the negative feelings to lessen and to cope effectively.

What is most important is that you understand that you are not alone. It is estimated that more than 100,000 Americans have NF, making it more common than cystic fibrosis, Duchenne muscular dystrophy, and Huntington's disease combined. The Children's Tumor Foundation has many resources for you all over the United States and would like you to think of us as your safe haven. These resources include clinics throughout the country, local chapters for nearly every state, and various events that you can attend to help you meet more members of the NF community around you.

Dealing with a diagnosis of a genetic disorder such as neurofibromatosis can be hard. We know that the current lack of a cure and the unpredictability of the disorder do not make things any easier. However, there are things that you can do to help make this feel a little more manageable.

- **Get the facts** - Read the "NF Basics" page in this package, or visit the Foundation's website at [www.ctf.org](http://www.ctf.org) to learn more about the three forms of NF.
- **Find a doctor that knows about NF** - Visit the "Find a Doctor" section of [www.ctf.org](http://www.ctf.org).
- **Get support** - You can find social support by getting in touch with a healthcare provider, or visiting the [ctf.org](http://ctf.org) "Living with NF" page, which can direct you to discussion boards, support groups, chat rooms, the NF Forum and NF Camp.
- **Get involved** - National programs like the NF Walk, NF Endurance, Racing4Research, and Cupid's Undie Run are great ways to empower yourself and your community in the fight for treatments and a cure for NF. It is also a way to begin meeting others in your local NF community.
- **Participate** - Join the NF Registry at [www.nfregistry.org](http://www.nfregistry.org)
- **Stay in touch** - Register for the CTF website at [www.ctf.org](http://www.ctf.org), "Like" our Facebook page and follow us on Twitter and Instagram for all the latest news and info.
- **Join** - Find your local CTF chapter and your regional Community Relations Coordinator on CTF's staff by visiting [www.ctf.org/communityrelations](http://www.ctf.org/communityrelations).



## The Children's Tumor Foundation

CTF is a non-profit 501(c)(3) medical foundation, dedicated to improving the health and well-being of individuals and families affected by neurofibromatosis.

The mission of The Children's Tumor Foundation is to:

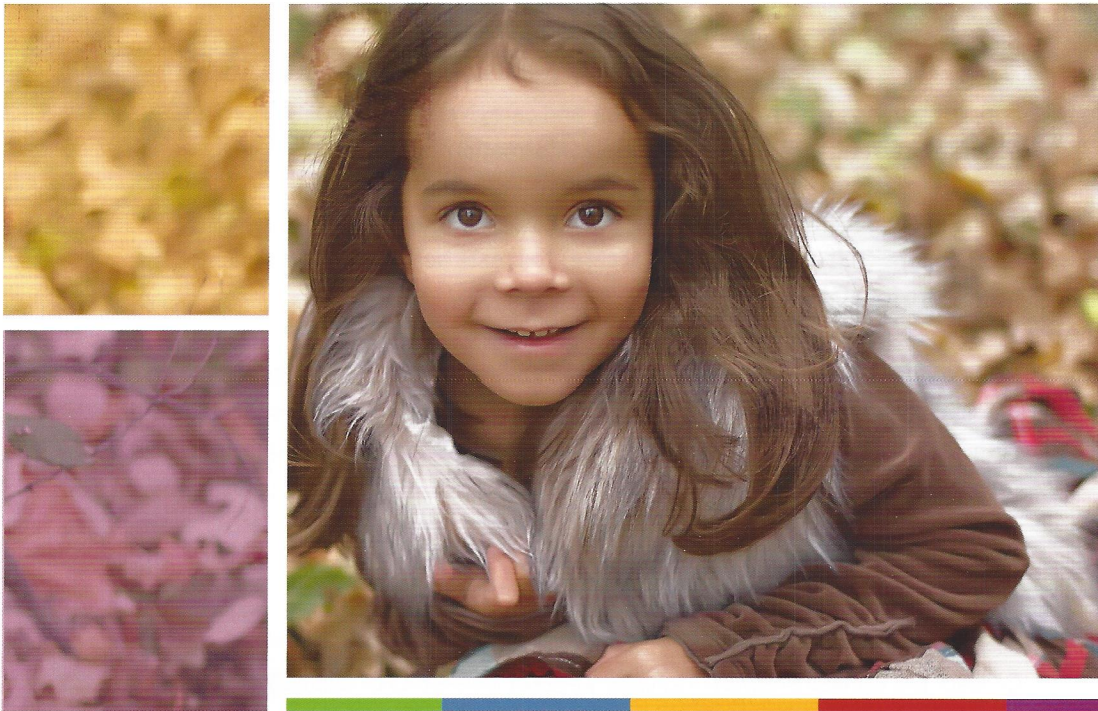
- Encourage and support research and the development of treatments and cures for neurofibromatosis types 1 and 2, and schwannomatosis;
- Support persons with NF, their families, and caregivers by providing thorough, accurate, current, and readily accessible information;
- Assist in the development of clinics, practices, and other patient support mechanisms (but not direct medical care) to create better access to quality healthcare for affected individuals;
- Expand public awareness of NF to promote earlier and accurate diagnoses by the medical community, increase public understanding of the challenges facing people with NF, and encourage financial and other forms of support from public and private sources.

Please feel free to call the Children's Tumor Foundation for information.  
Contact the Clinical Program Manager at (212) 344-6633 or [clinics@ctf.org](mailto:clinics@ctf.org).

## NF Basics

**Neurofibromatosis (NF)** is caused by a genetic change that makes people more likely to develop benign (non-cancerous) tumors around nerves and on the skin. NF can also affect bones, vision, and other body systems. NF1 is a lifelong condition often diagnosed in childhood. People with NF can lead full lives, but require a specialist's care. Neurofibromatosis has been classified into three distinct types: NF1, NF2, and schwannomatosis. One type cannot turn into another type.

**Neurofibromatosis 1 (NF1):** also known as von Recklinghausen NF or Peripheral NF, is the most common form of NF. Occurring in one out of every 3,000 births, it is characterized by multiple cafe-au-lait (light brown) spots and neurofibromas (small benign growths) on or under the skin. About 50% of people with NF also have learning challenges. Softening and curving of bones and curvature of the spine (scoliosis) occur in some patients with NF1. Occasionally, tumors may develop in the brain, on cranial nerves, or on the spinal cord. While NF tumors are not cancerous, they may cause health problems by pressing on nearby body tissues. In a minority (10%) of cases, a benign NF1 tumor may become malignant (cancerous). But 90% of people with NF1 will never develop a malignant tumor from NF1.

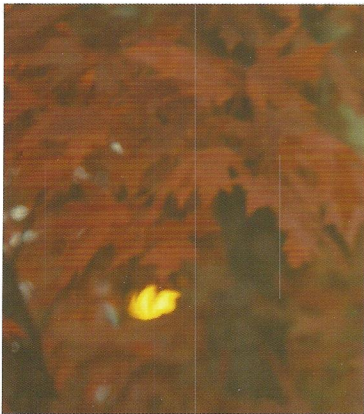


One of the first and most pressing concerns a person might have after a diagnosis of NF1 is how it affects life expectancy. You might see that some research shows that affected people have a shorter-than-average life expectancy. Although two-thirds of people with NF will never develop any major medical symptoms, some people with NF1 may have shortened life expectancies because of uncommon but serious NF1 related complications, mainly malignancies (cancers) and problems with blood vessels. These situations may be rare, but they are concerning and screening for them is an important part of NF1 management.

NF1 symptoms are different for each individual. It is impossible to predict how mildly or severely affected someone will be, or what medical issues they will develop. This can be frustrating for patients and their families. That is why it is very important for patients with NF1 to be treated by a group of healthcare providers knowledgeable about the disorder. You can find a list of NF specialist clinics in the United States at [www.ctf.org](http://www.ctf.org) under "Find a Doctor."

"Around two-thirds of kids with NF will never develop any major medical symptoms."

- Nicole Ullrich, MD, PhD, Boston Children's Hospital



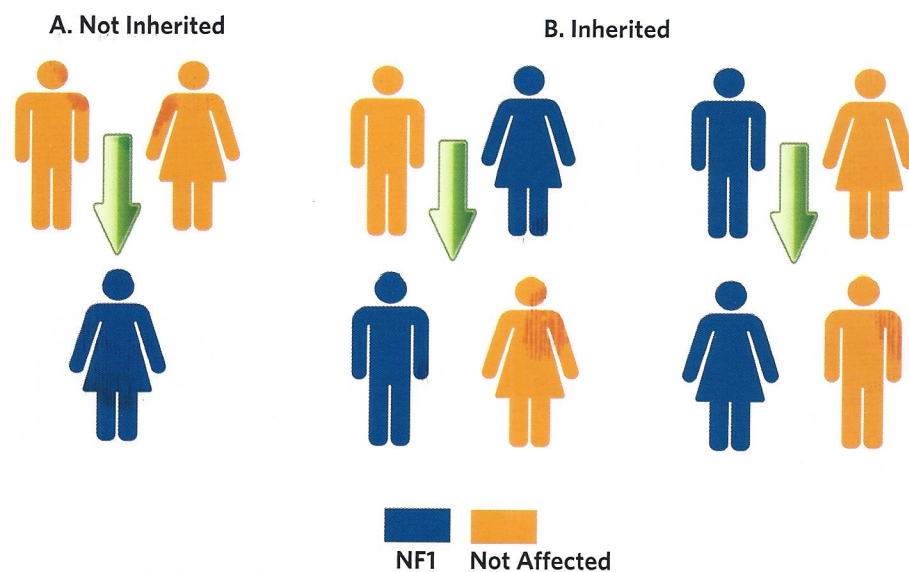
## How Does Someone Get NF1?

When someone has NF, they were born with it. Some people inherit NF1 from a parent, while others are the first case in their family (a spontaneous case). It does not result from anything that occurred during pregnancy. One out of 3,000 people are born with NF1. The cause of NF1 is a change in a gene. A gene is a sequence of DNA. The NF1 gene has a very long sequence, and even a small change can shut down its normal activity. The NF1 gene is responsible for the production of neurofibromin, a protein that keeps cells from growing too quickly. A person with NF1 has a genetic change in the NF1 gene that interferes with the normal production of neurofibromin.

In 50% of cases, patients inherit a changed NF1 gene from a parent with NF1 (see B, below). In the other half of cases, patients are born with a change in the NF1 gene even though neither one of their parents has NF1 (see A, below).

If the change is inherited from a parent, each time that parent has a child, the chance of that child having NF1 is 50/50. Whether or not any particular child develops the disorder is irrelevant to whether their other children develop it. For each child, the probability is still 50%.

If the change is a spontaneous case, the parent couple is very unlikely to have other children with NF1.







## How Is the Diagnosis Made?

The diagnosis of NF1 is most often based on a medical examination of a person's body. This type of diagnosis is called a clinical diagnosis. For example, light brown "café au lait" pigmented areas on the skin are often visible in the newborn period and parents may be told that their newborn may have NF1. Other times it is not until additional signs emerge that a diagnosis is suspected or confirmed.

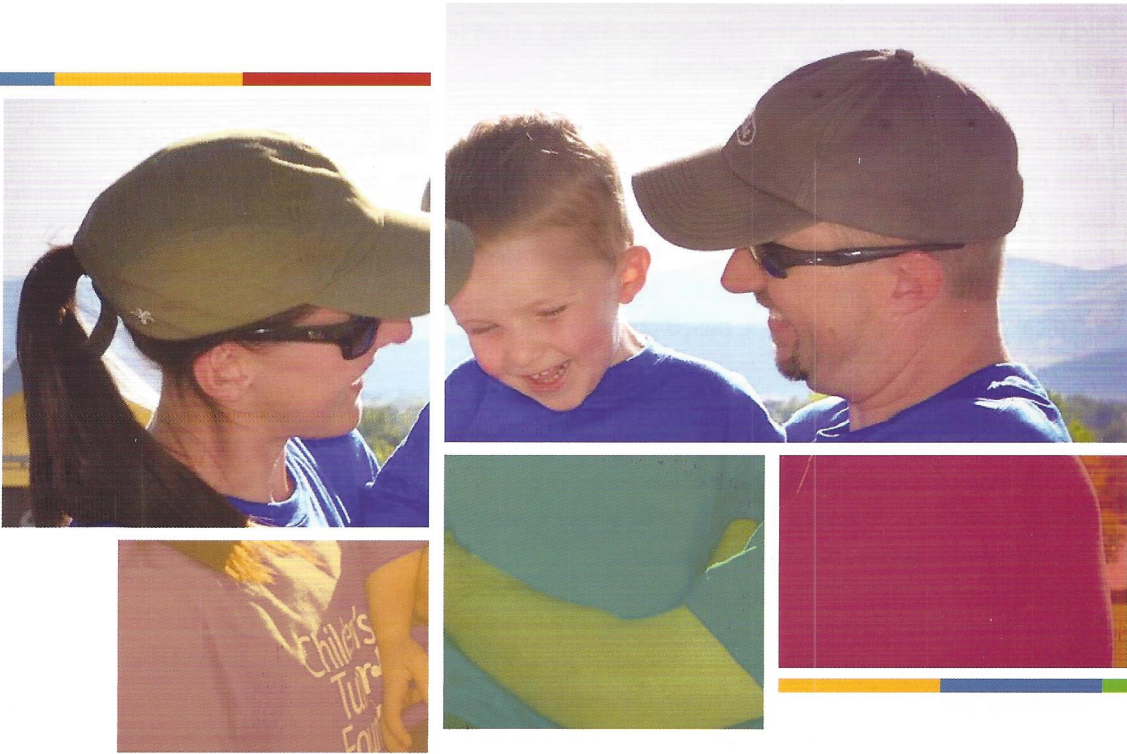
Additional signs that may emerge in childhood can include Lisch nodules (harmless "dots" in the colored part of the eye), freckling (especially under the arms or in the groin region), learning difficulties, or bowing of the long bones in the leg or arm. Sometimes NF1 is not diagnosed until puberty, when typically more neurofibromas appear on the skin.

A clinical diagnosis of NF1 is made based on a medical evaluation finding any TWO of the following:

- Six or more café-au-lait spots that are 5 mm or larger in pre-pubertal individuals or 15 mm or larger in post-pubertal individuals
- Two or more neurofibromas of any type or one or more plexiform neurofibromas
- Freckling in the underarm or groin areas
- Optic glioma (tumor of the optic pathway)
- Two or more Lisch nodules (benign colored spots in the eye)
- A distinctive bony lesion: dysplasia (abnormal growth) of the sphenoid bone behind the eye, or dysplasia of long bones, often in the lower leg
- Having a close relative (parent, sibling, or child) with NF1

People who have a suspected but not confirmed diagnosis of NF1 are usually offered regular medical follow-ups as if they have a diagnosis.

**Genetic testing:** NF1 is caused by a genetic change. This change can be detected by a specialized blood test. Genetic testing (“DNA testing”) for NF1 may be done in certain circumstances, but this test is not done routinely because a clinical diagnosis (based on observable signs of NF1) is considered reliable in most cases. In some instances, genetic testing can help to confirm the diagnosis if it is uncertain, and can be useful to establish a diagnosis in a young child who has not yet developed enough features to make a definite clinical diagnosis. For the most part, genetic testing does not predict the severity or specific complications of NF1. Genetic testing may also be informative for other family members or for deciding about reproductive options. The decision to have genetic testing is a personal one. A genetics healthcare provider or a genetic counselor can help you with this decision.



## What Are the Possible Symptoms of NF1?

No one person will have all the possible symptoms of NF1. Many people are quite mildly affected. There is a range of severities, and we currently have no way to predict which symptoms will appear in an individual. People with NF1 may experience the following:

**Café Au Lait spots:** These flat, light brown spots on the skin are not harmful.

**Neurofibromas:** These are the lumps or bumps on and under the skin that are typical of NF1. They can be on the skin surface (cutaneous neurofibroma) or under the skin (subcutaneous neurofibroma). Most are not a medical issue, but can cause pain and itching. The number of neurofibromas can be different between each person. They can be mild and only a few in number or more severe and be in the thousands. *Please be aware that photos on the Internet of neurofibromas tend to show severe cases. These images are not an accurate picture of what most patients with NF1 look like.*

A less common type of neurofibroma is called a plexiform neurofibroma. They occur in 25% of people with NF1. They can grow to be large and can cause a number of problems such as pressure on nerves and organs. These types of neurofibromas can become malignant in a minority of cases (about 10%).

**Cognitive Differences:** Intelligence in NF1 is usually in the normal range, but children may need extra help in school. Learning challenges occur in about 50% of children with NF1.

**Optic Pathway Tumors:** An optic glioma is a tumor that develops in the cells surrounding the optic nerve. About 15% of children with NF1 will develop an optic glioma. Children are most at risk for optic gliomas when they are under the age of six years. Most of the time, optic gliomas do not cause any symptoms and do not require treatment but sometimes can affect vision and may require chemotherapy.

**Delayed or Early Puberty:** Most people with NF1 will start puberty at the expected age range, but some may have precocious (early) or delayed puberty.

**Small Stature:** Affects about 30% of people with NF1. May be treated with growth hormone.

**High Blood Pressure:** For a small percentage of people, hypertension is one of the most serious complications of NF1. People with NF1 should have their blood pressure checked at least annually.

**Bone Issues:** Tibial bowing (curved lower leg bone) is seen in about 5% of patients. Scoliosis (curvature of the spine) occurs in 12-20% of patients.

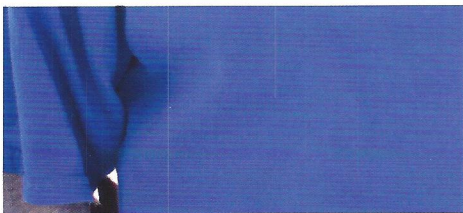
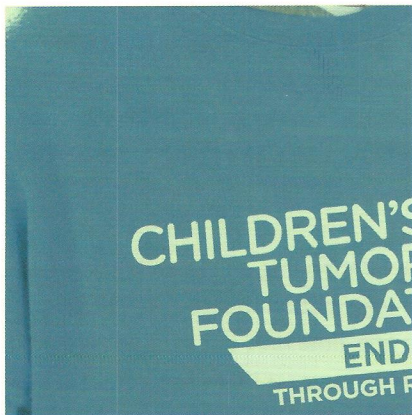
## Medical Management of NF1

Because NF1 involves many different systems of the body, doctors and staff from many different specialties may be involved in your care.

This is most conveniently done at a specialized NF clinic. You can find a list of NF specialty clinics on the CTF website ([www.ctf.org](http://www.ctf.org), "Find a Doctor"). Many of these NF clinics offer coordinated care so that specialists are in communication with one another and may be able to see you on the same day. If you do not have access to a specialty NF clinic, you may need to consult these specialists one at a time. It is important to see specialists who know about NF. For example, seek out an ophthalmologist (eye doctor) who knows what changes to look for in NF1 patients and the right methods to check for them. Your doctor may refer you to healthcare professionals who are experts in different NF1-related issues. You may be referred to the following:

- **Genetics:** Medical Geneticists are physicians with expertise in the diagnosis and management of genetic disorders including NF. Genetic counselors are healthcare professionals with training in medical genetics and personal counseling. Their job is to explain the genetic contribution to a medical condition and the various choices that can be made to deal with that condition.
- **Dermatology:** Dermatologists are trained to evaluate and treat conditions of the skin, hair, and nails.
- **Neurology:** Neurological specialists are trained in the treatment and diagnosis of conditions affecting the brain and nerves.
- **Ophthalmology:** These specialists are trained to diagnose, monitor, and treat (through medicine or surgery) conditions affecting the eyes and vision-related structures in the body.
- **Orthopedics:** This branch of medicine deals with conditions involving the muscles and bones.
- **Oncology:** Oncologists specialize in the diagnosis and treatment of different types of benign and malignant tumors.

- **Psychology:** Psychologists diagnose and treat emotional and behavioral issues.
- **Cardiology:** Cardiologists specialize in the heart and major blood vessels. They manage cardiac conditions such as heart abnormalities.
- **Pediatrics:** Pediatricians are experts in the medical care of children and adolescents.



## How to Share the News

One of the more difficult parts of dealing with a new diagnosis is telling close friends and family, or even the child who has the condition. Although everyone has their own strategies and personal preferences for what and how to tell others, the following suggestions may help make the process a little bit easier.

### WHO TO TELL

It is not uncommon for this question to come up when talking with a family member, a close friend, or maybe even a teacher—especially if the child has visible signs on the body, or NF-related learning difficulties that affect their school performance. Many people reveal their diagnosis to others only if it seems important to their relationship (for example, to a close friend, or a teacher). Letting the child also have a say in who to tell can be helpful as well.


### WHAT TO TELL

When you decide to share your or your child's diagnosis with others, you must also decide how much information to share. You might share in a limited way, such as discussing the learning difficulty aspects of NF1 with your child's teacher. Other times, it is helpful to have someone with whom you can share more details, including all of the emotional ups and downs that go along with the diagnosis.

### SOME TIPS FOR TELLING CHILDREN

Children prefer routine and certainty. Although neurofibromatosis is filled with uncertainty, the more you can establish certain routines and normalcy for the child, the more comfortable the child will be. It is also important that you tell the truth. Creating a trusting and honest relationship is very important. Keeping a child in the dark can sometimes create more anxiety.

How much you choose to tell your child will vary depending on the child's age and maturity level. Many parents allow the level of information that they tell their child to grow with them as they get older. This is not a one-time talk, but instead an ongoing conversation that evolves over time. Some studies suggest that until a child reaches age eight, they only need basic information such as the name of the disorder, the parts of the body that are affected, how it will be treated (if at all), and how their lives will be affected in the short-term.



Another tip to keep in mind is the way in which you are reacting to the diagnosis in front of your child. Before you begin the conversation with your child, it may be helpful to make sure that both parents, or caregivers, are on the same page and have come to terms with their own feelings. Children often depend on a parent or adult's reaction to decide how they themselves should react. Although neurofibromatosis is a serious matter, the calmer you remain, the less anxious the child will be.

### **SAMPLE MESSAGE TO FRIENDS AND FAMILY**

Dear \_\_\_\_\_,

I want to share with you that \_\_\_\_\_ has been diagnosed with neurofibromatosis type 1.

Neurofibromatosis type 1, or NF1, is a disorder that can cause benign (non-cancerous) growths called neurofibromas to grow on and under the skin. The severity and symptoms vary greatly from person to person. People with NF1 require special medical care, but usually enjoy full, active lives.

The most obvious signs of NF1 are neurofibromas and light brown patches on the skin. NF1 is something that people are born with, although it may not be diagnosed until later in life. It is caused by a genetic change and occurs in one in 3,000 births. Scientists and doctors are working towards understanding and treating NF1.

If you would like to learn more, please visit [www.ctf.org](http://www.ctf.org).  
We appreciate all of your love and support.

Best,

## Potential Learning Challenges Associated with NF1

The most common problems in children with NF1 are learning challenges and related problems. These affect about 50% of children with NF1. Most children with NF1 have normal intelligence; however, they may have issues in the processing of information. Problems with working memory, attention, visual-motor function, and spatial orientation are common issues. Keep in mind that these types of issues in NF1 are NOT universal.

Early intervention to address any learning issues can make a big difference. Working with your child's school and teachers to address these issues is vital.

What is known about cognitive and developmental issues in NF1:

1. For babies, delayed crawling, sitting, walking, and talking may be observed.
2. For school-age children, difficulties in handwriting, focus and attention, and verbal memory may occur.
3. At all ages, patients with NF1 often have difficulties in organization and time management.
4. Learning challenges in NF1 do not get worse over time, but it may appear that way as school tasks become more complex at each grade level.

### **Some general recommendations for identification and intervention of these problems:**

1. Talk to your doctor about your concerns. There are many things that you can do to help your child with daily school activities.
2. Talk to the school. Educate them about NF1 and learn about what they do for children with learning issues.
3. Learn about your educational rights. Get in touch with parent support groups. They can provide additional resources. Learn about school support, special education laws, and additional resources in the school system.



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- An Individualized Education Program (IEP) for your child may include extra time, shorter tests, or being assigned a desk near the front of the classroom.
  - Keep an eye on your child's self-esteem. The better they feel about themselves, the more willing they will be to put in the hard work needed to overcome any learning difficulties.
  - Check in periodically with CTF or other NF-related organizations. As we learn more, new interventions and treatments may become available.

*CTF thanks Maria T. Acosta, MD, Clinical Director, The Gilbert Family NF Institute, Children's National Medical Center, Washington DC, for contributing information about learning and NF1.*



## Insurance Coverage for Neurofibromatosis

Billing and payment for healthcare services in the United States are based on procedure and diagnosis codes. These codes are used to determine medical necessity for health services, but the billing and coding processes are quite complex. It is not uncommon for healthcare providers to make unintentional errors that can result in services not being covered.

It is important to know the correct diagnosis and procedure codes for NF. Being proactive and working closely with providers and insurance companies can eliminate most of the hassle and frustration in this process.

As of May 2014, the diagnosis codes in the United States are the version known as ICD-9. They are in the process of being updated to version ICD-10. The changeover is currently set to occur in October 2015.

Current ICD-9 codes for neurofibromatosis in use as of May 2014 are as follows:

DIAGNOSIS	DIAGNOSIS CODE
<i>NF, Type 1</i>	237.71
<i>NF, Type 2</i>	237.72
<i>Schwannomatosis</i>	237.73
<i>NF, unspecified</i>	237.70

ICD-10 codes, when they become active, will be as follows:

<i>NF, Type 1</i>	Q85.01
<i>NF, Type 2</i>	Q85.02
<i>Schwannomatosis</i>	Q85.03
<i>NF, unspecified</i>	Q85.00

A different set of codes covers medical procedures. These are CPT codes. One of the most common insurance issues for NF1 patients is covering the procedure of the removal of neurofibromas. Generally, removal of a skin growth for cosmetic reasons is not covered by health insurance. However, the situation is different for the removal of neurofibromas for patients diagnosed with NF. Medicare and Medicaid will cover this procedure because it is medically justified. Private insurance plans may reimburse at various levels, but the claim should be considered medically valid.

The CPT codes for removal of cutaneous (skin) neurofibromas in people with NF1 are as follows:

DIAGNOSIS	PROCEDURE CODE
<i>Excision of neurofibroma</i>	64788
<i>Excision of neurofibroma, extensive</i>	64792

If an insurance company does deny a claim, check with your doctor's office to make sure the claim was fully documented. A lack of information may be the reason why the claim was denied. Make sure that the claim filed includes an NF diagnosis code, the reason for the procedure (e.g., pain, loss of function), and the correct procedural code.

If you do not have health insurance, contact your state's Medical Assistance Administration for additional information.

## RESOURCES

The key thing to keep in mind while searching for information about NF1 is that your doctor is your best resource! If you come across information that you find confusing or strange, or if you just have more questions, you should ask your doctor.

### Books

**NEUROFIBROMATOSIS: A HANDBOOK FOR PATIENTS, FAMILIES AND HEALTH CARE PROFESSIONALS**

By Dr. Bruce Korf and Dr. Allen Rubenstein

*Information about the natural history and genetics of NF, as well as management and research.*

**THRIVING WITH NEUROFIBROMATOSIS**

By Kristi Hopkins

*This book is written by Kristi Hopkins, who has NF1 and three children with NF1. Kristi wrote this book about her personal experience with NF1. She also writes a blog about NF1, and is active in the NF1 community.*

### Websites

**Children's Tumor Foundation's website ([www.ctf.org](http://www.ctf.org))** The website of the oldest and largest national foundation for NF in the United States. Here you can find information on NF, support groups, NF specialists, research news, educational events, family events, and many other ways to support the NF community.

**NF Registry ([www.nfregistry.org](http://www.nfregistry.org))**. A patient-centered resource for sharing your experiences to help guide NF research. You can find clinical trials that you may want to consider participating in, see how your NF experiences compare to others, and be a part of an international NF awareness effort. There is strength in numbers, and the NF Registry needs your participation to make a difference.

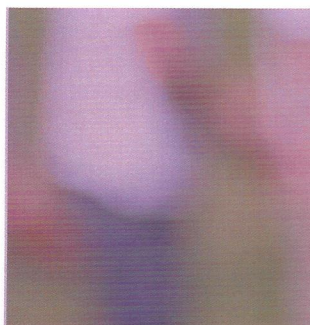
**Understanding NF1 ([www.UnderstandingNF1.org](http://www.UnderstandingNF1.org))** was created by Dr. Bruce Korf, Medical Director of the Children's Tumor Foundation. It provides information about NF1 in patient-friendly language. There are also videos and pages with perspectives from people with NF1 and their families. You will find patient care logs you can print out and use for your own record-keeping.

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