



**TALKING ABOUT
NEUROFIBROMATOSIS:
A GUIDE FOR TEENS**

children's

HOSPITAL • ST. LOUIS

 HealthCare

WHAT IS NEUROFIBROMATOSIS?

Whether you were just diagnosed with neurofibromatosis or you have been seeing an NF doctor for years, you probably have some questions about what neurofibromatosis really is. Chances are that none of your friends has ever heard of it. In fact, most people have never heard of neurofibromatosis, but believe it or not, it is not a rare disorder. One in every 3,000 people has NF, which makes it really pretty common.

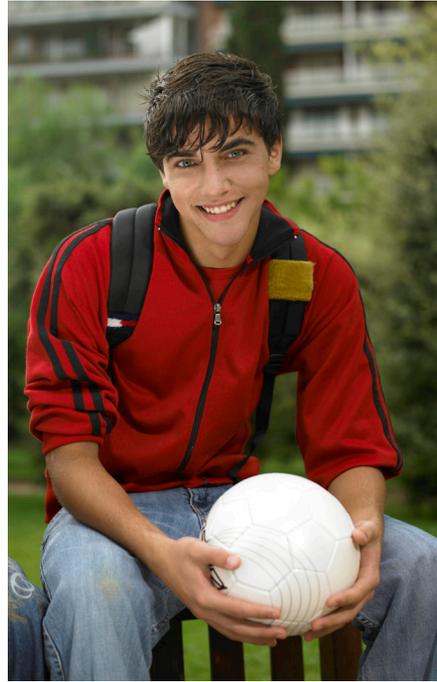
Neurofibromatosis is called "NF" for short. It also is sometimes called von Recklinghausen disease, after a German doctor named Frederick von Recklinghausen. But this is really just a very long name for the same thing.



The Two Types of NF

There are two conditions with “NF” in their name, which can sometimes make things really confusing, because they are totally different conditions. There is **NF Type 1** (called NF1) and **NF Type 2** (called NF2). NF1 and NF2 have completely different features and medical concerns. All that they have in common is the name “NF.” NF1 can never become NF2 or the other way around.

This booklet is written only for people with NF1. Some of the stuff you might read about NF2 might be scary if you don’t realize that you have NF1, not NF2. If your doctor says you have NF1, you can ignore everything you read about NF2. You won’t develop the problems that people with NF2 can have.



The Features of NF1

To be given the diagnosis of NF1, someone must have two features from the list below:

- Six or more café-au-lait spots
- Freckles in the armpit or groin
- Lisch nodules (freckles on the colored part of the eye)
- Two or more neurofibromas or a plexiform neurofibroma
- An optic glioma
- A specific bone problem — either sphenoid dysplasia or tibial dysplasia
- A parent, brother, sister or child with NF1

Most people with NF1 don’t develop all of these features. The good news is that most of the features are cosmetic, don’t cause any medical problems, and certainly aren’t life-threatening. Some of them, however, do need medical attention. Each one is discussed here so you’ll better understand what they are.

Features of NF

The most common feature seen in NF1 is the flat, tan birthmarks that you have on your skin. They are called **café-au-lait spots**, which is just a fancy French term referring to the light coffee color of the spots. They don't cause any medical problems. Some people may have six spots and others may have 20; but having more spots doesn't mean that you have a "worse" case of NF1. You won't keep getting more spots throughout your life, either. Most people have all the spots they will ever have by the time they are five years old.

Most people with NF1 develop **freckles** in their armpits or groin (the crease where your leg meets your body). These freckles are helpful in making the diagnosis of NF1 but don't cause any medical problems. They usually develop during elementary school.

Lisch nodules are freckles on your iris (the colored part of your eye). They don't affect your eyesight in any way but they are important for diagnosing NF1. Nearly all people with NF1 will develop Lisch nodules by the time they are adults.

The "neuro" in neurofibromatosis refers to the body's nerves. People with NF1 can develop bumps on the nerves called **neurofibromas**. Neurofibromas look like lumps on or under the skin. They can grow any place where there are nerves — on the face, scalp, chest, as well as inside the body.

Neurofibromas often start to show up during puberty. They may keep getting bigger for a while but will eventually stop growing. Typically, people gradually develop new ones as they get older. There is no way to predict how many neurofibromas someone will develop. Neurofibromas usually don't hurt but are sometimes tender if they are irritated by something rubbing on them, like a shoe or waistband. Doctors may sometimes call them a tumor, but they aren't a cancer and won't turn into a cancer.

Plexiform neurofibromas are also benign (meaning non-cancerous) tumors but they are a little different than the kind of neurofibroma we just talked about. Plexiforms, as your doctor may call them, are usually bigger than a regular neurofibroma and may feel like a lot of small lumps bunched



together. A plexiform neurofibroma is often present at birth and often does much of its growing during childhood. Most people with NF1 will develop regular neurofibromas, but only a third of all people with NF1 will have a plexiform neurofibroma. You may want to ask your doctor if any of your tumors are a plexiform, so you know the difference.

Although they are not a cancer, a plexiform neurofibroma can sometimes turn into a cancer. This doesn't happen very often, though. If you have a plexiform neurofibroma, you need to tell your doctor right away if it hurts constantly, if it starts growing quickly, if it changes in the way it looks or feels, or if you have a change in your usual activity (like weakness or persistent numbness of your arm or leg). Even if a plexiform neurofibroma hasn't become cancerous, it may hurt if it is hit or bumped.

An **optic glioma** is a tumor that can develop on the optic nerve, which is the nerve that connects your eye to your brain. Without proper management and treatment, an optic glioma can sometimes go on to cause problems with someone's eyesight. When you were younger, you may have had an MRI, a special test that takes pictures of the brain, to look for an optic glioma. An optic glioma, however, is something doctors usually worry about in little kids with NF1. It would be really unusual for a teenager to suddenly develop an optic glioma that would cause any problems.

The **bone problems** seen in NF1 are present at or just after birth, and aren't very common. So if your doctor hasn't told you that you have either sphenoid dysplasia (a problem in the way some of the bones in the eye socket developed) or tibial dysplasia (a problem with the way the lower leg bones developed), then it isn't something you need to worry about.

The last thing from the list of features that we started with is having a **family history of NF1**. Not everyone with NF1 has someone else in the family with NF1. We'll explain more about this later.

What can happen as a teen with NF?

Now that you're a teen with NF1, you already have freckles and café-au-lait spots.

Typically, people with NF1 start to see neurofibromas appear during their teens. The neurofibromas grow slowly and may look like a pimple at first. You won't wake up one morning, or next year, and be covered with neurofibromas. They develop gradually over a period of many years.

If a neurofibroma is painful or you just don't like the way one looks, it can be removed by a doctor who has experience with NF1 and neurofibromas; but having a neurofibroma removed doesn't guarantee that it won't grow back or that you won't grow a new one in the same area.

As a teenager, your height may be more important to you than it was before. Most people without NF1 are around the height of their parents. People with NF1, though, are sometimes a little shorter than their classmates.

Some people with NF1 have learning concerns. It doesn't mean that they are dumb or can't learn; it just means that they may need some additional help in certain subjects in school. Lots of people with NF1 go on to college even if they had learning difficulties as a teenager. Whether you choose to go to college or get a job right out of high school, you can enjoy a career in a variety of interesting and successful fields.

How does someone get NF1?

NF1 is a genetic condition that happens when a certain gene changes. It is something you were born with, not something you can catch from another person like you can catch a cold.

Genes provide the instructions that tell our bodies how to grow and develop. We all have genes that influence how our body grows — our eye color, height and heart development, just to name a few. There is also an *NF1* gene. Everyone has two copies of the gene, even people who don't have NF1. In people with NF1, one copy of the *NF1* gene is changed. When one copy of the *NF1* gene is changed, it can't work the way it is supposed to anymore, and that person has NF1.



Half of all people with NF1 inherit the changed *NF1* gene from a parent with NF1. When a parent has NF1, they must either pass on the *NF1* gene that is changed or the *NF1* gene that is not changed. There is nothing a parent can do to control whether their child inherits the changed gene, so there is a 50-50 chance that the child will inherit NF1.



The other half of all people with NF1 are the first in their family to have NF1. This happens when the gene change occurs as a brand new change in that person. It has nothing to do with anything mom or dad may have done before or during pregnancy.

If you have NF1, you will have a 50-50 chance of passing it on to each of your kids one day. NF1 is not something that skips generations, and it affects boys and girls equally. If you have NF1, it won't turn into Type 2 in your kids. However, a child can be less severely affected or more severely affected than his or her parent. There is no way to predict what problems a child with NF1 will have based on the problems their parent with NF1 had.

Hopefully since we're talking about having kids, you realize that people with NF1 do get married. A lot of people with NF1 worry about developing neurofibromas and what they will look like. But the bottom line is that people with NF1 are still attractive to the opposite sex, get married, and have kids just like people without NF1.

Other Random Things

OK — So here are some other common questions or things you might be wondering but were afraid to ask:

Who is the Elephant Man?

The Elephant Man is a guy who had thick lumpy skin that resembled that of an elephant — but he didn't actually have NF1. A doctor once said he had NF1, and unfortunately, the name association stuck.



Does having NF1 make me “different”?

No! Most likely a lot of your friends at school don't even know you have NF1, because you really don't look any different from them. There is no reason to think that you are any different from your friends. Being a teenager can be a lot of fun — so enjoy all the other things that your friends do!

Will I be covered with neurofibromas?

There is no way to predict how many neurofibromas you might develop in your lifetime. Like we discussed before, neurofibromas can be safely removed by a doctor who has experience removing them.

Is there anything I can do to stop the neurofibromas from growing?

There is no medication that can prevent neurofibromas from growing. And, there is nothing you can do that would make more neurofibromas develop. Neurofibromas often appear or grow in size during times of hormone changes such as puberty (which you can't avoid) and pregnancy. Common things like caffeine and exercise do not cause an increase or decrease in the number or size of neurofibromas.

Are there things I shouldn't do — like play certain sports — because I have NF1?

Generally, there is nothing you can't do just because you have NF1. Most kids can play sports, learn to drive, sing in the choir, go away to camp, hang out with friends, go to college, get married and have children. If you have any concerns about certain restrictions you may have, you should always ask your doctor first.

Will I develop cancer or die from NF1?

This is very unlikely. Most people with NF1 live normal, full lives. There are some rare complications, including cancer, that people with NF1 can develop. These problems happen a little more often in people with NF1 than they do in people without NF1, but they are still pretty rare.

Neither my mom nor my dad has NF1. Does this mean I'm adopted?

No. Only half of all people with NF1 have a parent who has NF1. The other half of the time, NF1 is caused by a new change in the person's *NF1* gene. In other words, you could be the first person in your family to have it because of a random change that occurred in your *NF1* gene.

Should I tell my friends I have NF1?

Some people tell their friends they have NF1 and others don't. It's a personal decision and one you should probably talk about with your parents. Having NF1 is nothing to be ashamed of, but not everyone will necessarily understand what having it means. You should think about who your good friends are, and who will understand and be supportive if you tell them. There is nothing wrong with just telling people that you have a bunch of birthmarks. The fact is that most people won't notice or even think to ask.

What does the future hold as far as treatment?

There is a lot of research going on with NF1 and possible treatments. It is our hope that better treatments will be available in the near future for people with NF1.

What if I have more questions?

Feel free to ask your parents and doctor any other questions you may have. Don't be afraid. They want to help!

In our own words



"People don't even know I have NF. I still do what I want, and I have fun no matter what. I won't let NF take control of my life."

— Sean, 13

"Being 16, I can't help wanting to look perfect. But even though I do look a little different, I still have confidence in myself. I just live a normal life."

— Alex, 16



"NF really doesn't affect my life."

— Hart, 12

"Everyone has challenges in life. It is what you do in spite of them that makes you who you are as a person."

— Brittani, 16



"I'm active in swimming, piano, gymnastics, school and choir. These are all the things NF doesn't stop me from doing!"

— Lindsay, 12



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