YOUR CHILD'S DIAGNOSIS OF NEUROFIBROMATOSIS

Children's HOSPITAL • ST. LOUIS
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A Guide for Parents

As a parent, you want what is best for your child. Finding out that your child has a condition you have never heard of may be frightening, particularly as you wonder what this may mean for your child's future.

This booklet is designed to help answer your questions about Neurofibromatosis type 1. It is our hope that information about your son or daughter's diagnosis will relieve some of your concerns, help you to better understand your child's specific health care needs, and empower you as a parent to become an active part of your child's medical team.

The Two Types of NF

Neurofibromatosis is called "NF" for short. And while you have probably never heard of it before, it is a fairly common genetic condition, affecting one in every 3,000 individuals. It also is sometimes called von Recklinghausen disease, after a German doctor named Frederick von Recklinghausen.

Two different conditions use the name "NF". This can make things confusing as families search the Internet for information. There is NF type 1 (called NF1) and NF type 2 (called NF2). NF1 and NF2 are two different medical conditions. They have completely different features and associated medical concerns. All that they have in common is the name "NF." NF1 can never become NF2 or the other way around. And while TV and popular magazines may sometimes call it the "Elephant Man's disease," NF is a different condition altogether.

This booklet is written only about NF1. Some of the information you may read about NF2 might be particularly frightening if you don't realize that your child has NF1, not NF2. Your child will not develop the problems associated with NF2.

Most people with NF1 do very well. The most common features of NF1 are birthmarks and freckles on the skin. These do not cause any medical problems. But some of the other features that are seen less frequently may require treatment by an NF doctor. Since no one can predict which child will develop what features of NF1, all children are followed to screen for anything that may need medical attention.

How is the diagnosis of NF1 made and what are the features of NF1?

To be given the diagnosis of NF1, someone must have two features from the list below. It is uncommon that an individual has all these features.

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

Each one of these features is discussed below for you to better understand what the terms mean.

The most common feature seen in NF1 is flat, tan birthmarks on the skin that are typically found at birth or during the first few months of life. They are called *café-au-lait spots,* which is the French medical term referring to the light coffee color of the spots. These spots do not cause any medical

concerns. Some children will have six spots while others may have 20. The number of spots does not indicate how "severe" your child's diagnosis is and does not correlate with other medical problems.

Most people with NF1 also develop freckles in their armpits *(axillary freckles)* or freckles in their groin *(inguinal freckles)*. These freckles are helpful in making the diagnosis of NF1, since they develop in areas where freckles do not usually appear in people without NF1. The freckles do not cause any medical problems.

Lisch nodules are bumps on the iris (the colored part of the eye). Lisch nodules are best detected by an eye doctor and are not the flecks of color that you may see in your own eyes when you look in the mirror. Lisch nodules do not affect eyesight in any way, and people with NF1 can't feel the bumps beneath their eyelids. 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 growths 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 appear during puberty and may grow in size and number over time. 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.

A *plexiform neurofibroma* is also a benign (meaning non-cancerous) tumor, but they are a little different than the kind of neurofibroma described in the previous paragraph. Plexiforms (as doctors may call them) are usually bigger than a regular neurofibroma and may feel like a lot of small lumps bunched together. Most people with NF1 will develop 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 child's tumors are plexiform neurofibromas. People who have plexiform neurofibromas may say they are painful if hit or bumped. Although they are not a cancer, a plexiform neurofibroma can sometimes turn into a cancer. Warning signs include significant pain, weakness in a leg or arm, or rapid growth.

An *optic glioma* is a tumor that can develop on the optic nerve, which is the nerve that connects the eye to the brain. Without proper diagnosis and treatment, an optic glioma can sometimes go on to cause problems with a child's eyesight. Eye exams once a year are the best way to help detect an optic glioma. Only 15 to 20 percent of children with NF1 will develop an optic glioma, and half of these children will never have any problems with their vision.

The bone problems seen in NF1 are present at or just after birth: *sphenoid dysplasia* (a problem in the way some of the bones in the eye socket develop) and *tibial dysplasia* (a problem with the way the lower leg bones develop). A tibial dysplasia can lead to a break in the bone (fracture).

The last thing from the list of features is a *family history* of NF1. About half of all children with NF1 will also have a parent who has NF1. Sometimes, a parent may learn that he or she has NF1 at the same time a child is diagnosed. If you are concerned that you or your spouse may have features of NF1, please raise the issue with your child's NF1 doctor.

The features of NF1 develop over time. Frequently, infants and young children with NF1 have several café-au-lait spots but will not yet have two features of NF1. In such instances, the diagnosis of NF1 cannot be made until the child gets older and develops a second feature of NF1. Most people with NF1 will develop at least two of the features of NF1 by late childhood or early adolescence. The majority of people with NF1 will, over time, develop café-au-lait spots, freckling, Lisch nodules and neurofibromas.

Your doctor may discuss genetic testing with you. Genetic testing is not necessary to confirm your child's diagnosis if your child has two of the features from the earlier list. If your child does not yet have two features of NF1, a change in the *NF1* gene will confirm the diagnosis of NF1. However, if a change in the *NF1* gene is not found, it cannot rule out the diagnosis of NF1. Genetic testing does not help your child's doctors predict which features of NF1 your child may or may not develop.

Are there other medical concerns associated with NF1?

About half of children with NF1 have a learning disability. It is uncommon, however, for a child with NF1 to be mentally retarded. The specific learning problems in children with NF1 are similar to and managed in the same way as learning concerns seen in persons who don't have NF1. In addition to specific learning disabilities, children with NF1 may have low muscle tone, coordination difficulties, and behaviors associated with attention-deficit hyperactivity disorder.

Children and adults with NF1 are often slightly shorter than their classmates and other family members. Some children may develop scoliosis (a curvature of the spine). Less frequently, children can have seizures, high blood pressure, or a heart defect at birth. Some specific types of cancer are slightly more common than in persons without NF1.

What should I expect as my child gets older?

Since various features of NF1 may appear as your child gets older, it is important to know what to expect during the phases of your child's growth and development.

In *infancy,* you may notice that your child has several café-au-lait spots. Your child's doctor will count the number of café-au-lait spots and also look for any concerns with how the bones developed. Your doctor should also take a family history to see if there are other family members who may have NF1.

In *early childhood,* your child may develop more café-au-lait spots. During this time, most children with NF1 will also develop axillary and inguinal freckling. Your child's doctor will look for any new café-au-lait spots and

for any freckling. Eye exams once a year by a pediatric ophthalmologist are very important during early childhood, since this is the most common age at which optic gliomas present. Also, half of children will have Lisch nodules by the time they are 5 years old, and almost all people with NF1 will have them by the time they reach adulthood. During this time, you and your child's doctor will follow your child's development to identify any possible learning concerns. Plexiform neurofibromas are typically diagnosed in early childhood but may go undetected if they are not causing problems and are not visible. They often do much of their growing during childhood, but can also enlarge in adolescents or adults

During *late childhood* and through *adolescence,* neurofibromas start to appear. A neurofibroma may look like a pimple at first and may grow gradually in size. A person with NF1 may continue to gradually develop new neurofibromas over the years throughout adulthood.

What special medical care does my child need and what should I pay attention to at home?

Your child should be seen once a year by a *doctor who specializes in NF.* At this appointment, your child's doctor will see if he or she has developed any new features of NF1. The doctor will also do a physical exam to make sure there are no medical concerns related to NF1.

Your child should have an eye exam once a year by an *ophthalmologist* familiar with NF1. This exam will look for Lisch nodules as well as for any changes in your child's vision that may be associated with optic glioma. You should call your child's doctor if you notice any changes in vision or signs of early puberty such as body odor, breast development, axillary or pubic hair, or enlargement of the male genitals. These could be signs of an optic glioma. Optic gliomas that are affecting eyesight may lead to blindness if undiagnosed and untreated. Eye exams once a year by an experienced pediatric ophthalmologist are the best way to find a change in your child's vision.

If there is any concern regarding your child's vision, your doctor will order an *MRI* (Magnetic Resonance Imaging). An MRI is a special test that takes pictures of the brain and is used to diagnose an optic glioma. If your child has an optic glioma that is affecting vision, your doctor may recommend chemotherapy to treat the tumor.

There is nothing to prevent or slow the growth of neurofibromas. They can be safely removed but this should be done by an experienced physician. Ask your NF doctor for a referral. Having the neurofibromas removed does not guarantee that they won't grow back or that new ones won't develop.

Unlike neurofibromas, plexiform neurofibromas are very difficult to remove, and surgery is generally not recommended. Most plexiform neurofibromas do not become cancers, but sometimes changes in the tumor are an indicator that it has become a cancer. If your child has a plexiform neurofibroma, you should call the doctor if your child complains that it hurts constantly, if it starts growing quickly, if it changes in the way it looks or feels, or if your child has a change in usual activity (like weakness or persistent numbness of an arm or leg).

What causes NF1?

NF1 is a genetic condition caused by a change in a single gene. It is something your child was born with.

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, to name a few. There is also an *NF1* gene. Everyone has two copies of this gene. 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 and that person has NF1.

Half of all people with NF1 inherit the changed *NF1* gene from a parent. When a parent has NF1, the parent 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 a child inherits the changed gene, so there is a 50-50 chance that the child will inherit NF1. Sometimes, a parent does not even realize he or she has NF1 and may be diagnosed at the same time as the child.

The other half of all people with NF1 are the first person in his or her family to have NF1. The gene change occurs for the first time in that child.

When your child grows up, he or she will have a 50-50 chance of passing NF1 on to each of his or her children.

NF1 is not something that skips generations, and it affects boys and girls equally. A child can be less severely affected or more severely affected than the parent. There is no way to predict what problems a child with NF1 will have based on the problems a parent with NF1 had. If your child has NF1, it won't turn into NF2 in his or her children.

Regardless of whether your child inherited the *NF1* gene change from you or is the first person in the family to have NF1, the fact that your child has NF1 is not your fault. There is nothing you did before, during, or after your pregnancy that caused your child to have NF1.

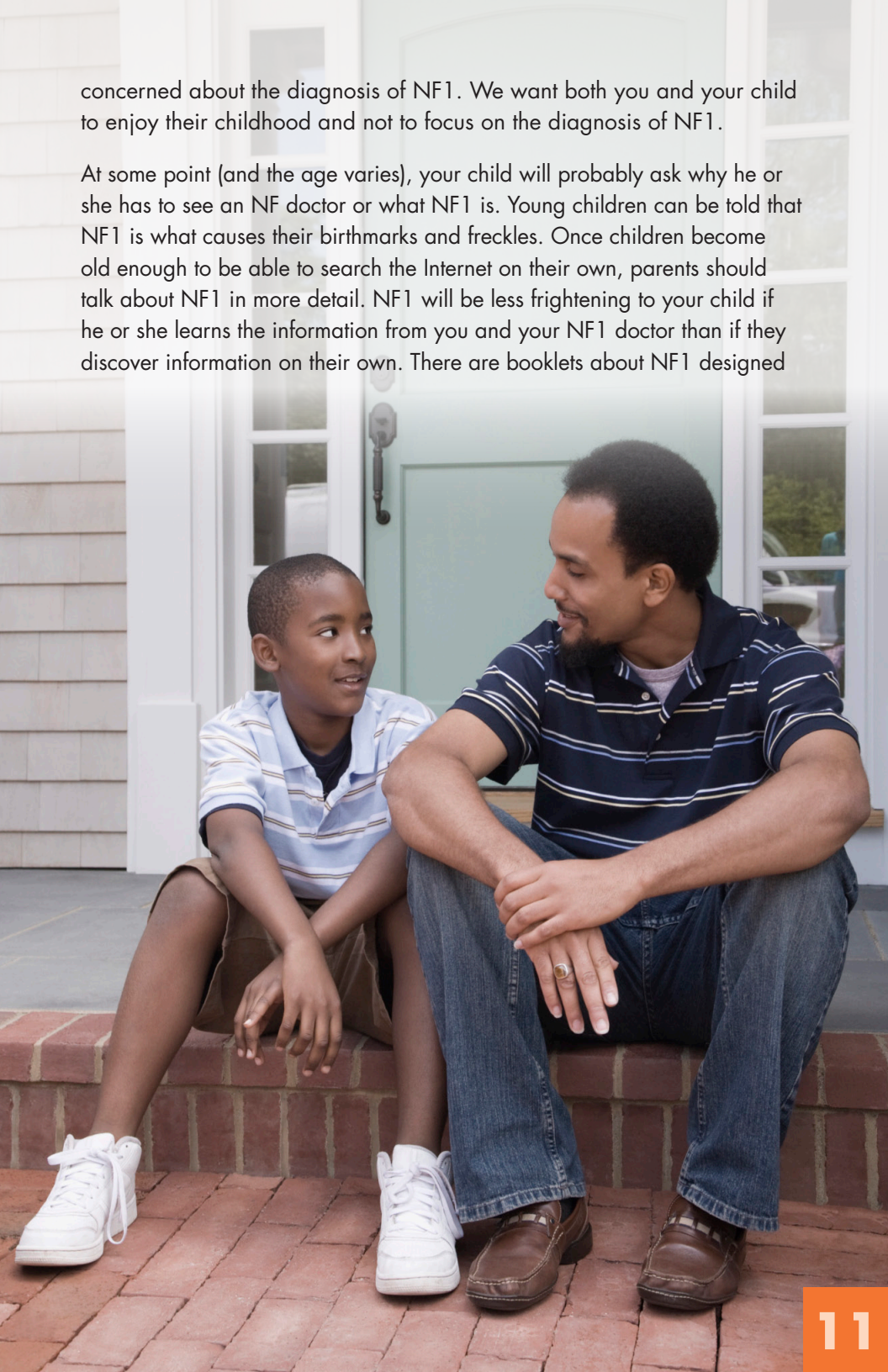
What should I tell my child about NF1 and when?

Presenting information to your child in a straightforward way will go a long way in developing open and honest communication between you and your son or daughter. It allows your child to know that you aren't hiding anything so that they will feel comfortable to come to you later with questions and concerns.

Your son or daughter will also take cues from you. If you are honest while maintaining a positive attitude, it will help your child avoid being overly

concerned about the diagnosis of NF1. We want both you and your child to enjoy their childhood and not to focus on the diagnosis of NF1.

At some point (and the age varies), your child will probably ask why he or she has to see an NF doctor or what NF1 is. Young children can be told that NF1 is what causes their birthmarks and freckles. Once children become old enough to be able to search the Internet on their own, parents should talk about NF1 in more detail. NF1 will be less frightening to your child if he or she learns the information from you and your NF1 doctor than if they discover information on their own. There are booklets about NF1 designed



for children and teens that may help your conversation. These are available on the Washington University NF Center website and from the NF Clinical Program. You should also emphasize to your child that the NF1 will never turn into NF2. It is also important to remember that the quality of information available on the Internet varies. Some of the information may be alarming and not accurate.

As children enter their teens, they may become self-conscious about their appearance. You can encourage your child by telling him or her that the café-au-lait spots are simply birthmarks. Children also may notice that they are a little shorter than many of their classmates. A lot of people with NF1 worry about developing neurofibromas and what they will look like. It will be important to emphasize that neurofibromas grow and develop slowly over years, not within a period of a few days or even weeks. The bottom line is that people with NF1 are still attractive to the opposite sex, get married, and can have children just like people who don't have NF1.

Whenever your child asks a question that you can't answer, say that you don't know but that you will find out. They will appreciate your honesty. Then call your child's NF1 doctor's office so that both you and your child can learn more.

Below are several questions that your child or teenager may ask you.

Will I be covered with neurofibromas?

There is no way to predict how many neurofibromas you might develop in your lifetime. 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? Or does anything make more grow?

There is no medication currently available that can prevent the neurofibromas from growing. And there is nothing you do that will make you develop more neurofibromas. 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, exercise and smoking do not cause an increase or decrease in the number or size of neurofibromas.

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 some cancers 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. 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.

Common questions parents ask:

What is the chance that we will have another child with NF1?

Your child's doctor should take a detailed family history to determine if either parent may have NF1. When a parent has NF1, there is a 50-50 chance with each pregnancy that the child will inherit NF1. Using new technology, NF1 can now be diagnosed before or during pregnancy when one member of a couple has NF1. Ask your doctor for more information about pre-implantation genetic diagnosis and prenatal genetic diagnosis. When neither parent has NF1, the chance that another child will have NF1 is less than 1 percent.

Is there a way to fix the gene?

Even though we can identify the exact change in the gene, there are millions of cells throughout the human body, and we do not know how to correct the change in the gene inside every cell. Research is currently focused on finding ways to reverse the effects of having an *NF1* gene that doesn't work the way it should.

Why are MRIs not done on every child to look for optic pathway gliomas?

Studies have shown that there is no difference in outcome between those children whose optic glioma was diagnosed on a screening MRI and those children who are diagnosed following an abnormal eye exam.

Should I tell my child's teacher?

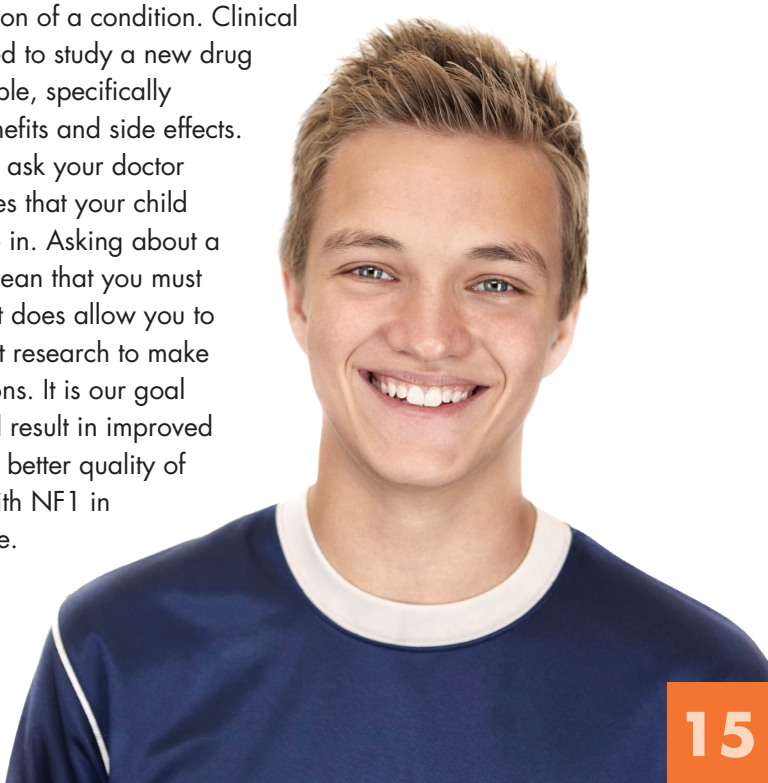
Approximately half of all children with NF1 have a learning disability. If you and your child's teacher feel that your child has a learning disability, formal testing should be done. This may include IQ testing and other tests to identify strengths and weaknesses. If a learning concern is found, the teachers and school staff will meet with you to develop an Individualized Education Plan (IEP) for your son or daughter. Early identification of learning concerns will help your child to maximize long-term success in school.

Should I limit my child's activities because they have NF1?

In most instances, having a diagnosis of NF1 does not limit someone's activities and hobbies. Encourage your child to participate in those activities in which he or she displays an interest and a talent. These things will show your child how they are more like their friends than they are different.

What is being done to learn more about NF1 and to find better treatments?

There is active research focusing on NF1 at many universities and hospitals around the country. Some of this research does not involve human subjects but is done in the lab to learn more about the basic biology of NF1. Other research involves patient participation through registries and clinical trials. Registries, or natural history studies, track the normal progression of a condition. Clinical trials may be used to study a new drug or device in people, specifically looking at its benefits and side effects. You may want to ask your doctor if there are studies that your child could participate in. Asking about a study does not mean that you must participate, but it does allow you to learn more about research to make educated decisions. It is our goal that research will result in improved treatments and a better quality of life for people with NF1 in the years to come.



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