YOUR CHILD’S DIAGNOSIS OF NEUROFIBROMATOSIS TYPE 1
A GUIDE FOR PARENTS
As a parent, you want what is best for your child. Finding out that your child has a condition that 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 answer your questions about Neurofibromatosis type 1 (NF1). It is our hope that information about your child’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.
Neurofibromatosis, or NF, actually refers to several medical disorders, including Neurofibromatosis type 1 (NF1), Neurofibromatosis type 2 (NF2), and Schwannomatosis. These are different medical conditions with distinct features and associated medical concerns. Of the three conditions, the most common is NF1. It is important to understand that NF1 does not develop into NF2. In addition, although NF1 has been referred as the “Elephant Man’s disease” in the popular press, this is incorrect.

This booklet specifically addresses your child’s diagnosis of NF1. While you have probably never heard of NF1, it is a fairly common medical condition, affecting one in every 3,000 individuals worldwide. You might also see it referred to as von Recklinghausen disease, named after Dr. Frederick von Recklinghausen, who first described this disorder in the 1880s.

The most common features of NF1 are birthmarks and freckles on the skin. While these birthmarks and freckles do not cause any medical problems, some of the other features of NF1 may require medical attention and treatment. Since no one can predict which features a child with NF1 will develop, all children should be closely monitored to allow for the early detection and management of potentially serious medical problems.
How is the diagnosis of NF1 made, and what are the features of NF1?

To receive a diagnosis of NF1, a person must have either a positive genetic test, or two or more of the following features:

- Six or more café-au-lait spots
- Freckles in the armpit or groin
- Lisch nodules
- 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

The most common feature of NF1 is café-au-lait spots. These are flat birthmarks that are slightly darker than the surrounding skin, and are typically found at birth or during the first few months of a child’s life. The number of spots does not indicate how “severe” your child’s diagnosis is, and does not correlate with other medical problems. Most
people are not bothered by their birthmarks, and notice that they usually lighten by adulthood.

Most people with NF1 also develop freckles in their armpits (axillary freckles) or groin area (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. Similar to cafe-au-lait spots, these freckles do not cause any medical problems.

Lisch nodules are small bumps that appear on the colored part of the eye (iris). They are best detected by an eye doctor (ophthalmologist). Lisch nodules do not affect vision, 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.

People with NF1 can also develop neurofibromas, which are benign tumors that grow near nerves in the body. Neurofibromas look like skin-colored lumps or purple-colored areas on the skin, or may feel like small bumps under the skin. They can grow anywhere on or inside the body. Neurofibromas usually start to appear just before puberty, and typically grow in size and number over time. There is no way to predict how many neurofibromas someone will develop over the course of their life. Neurofibromas usually don’t hurt, but are sometimes tender if they are irritated by something rubbing on them, like a shoe or waistband, and may be itchy when first appearing. Although doctors call them tumors, they are not cancerous and will not turn into a cancer.

Most people with NF1 will develop neurofibromas, but only a third of all people with NF1 will have a plexiform neurofibroma. A plexiform neurofibroma is a benign (non-cancerous) tumor that involves a network of nerves in the body, and is usually bigger than a dermal (skin) neurofibroma. It may feel like a cluster of small lumps bunched together, or may feel soft and “pillowy”, like there are feathers under the skin. People who have plexiform neurofibromas may say they are painful or uncomfortable when hit or bumped. Plexiform neurofibromas are different than dermal neurofibromas, because they can sometimes turn into cancer. For this reason, you should ask your child’s doctor if your child has a plexiform neurofibroma, and notify them immediately if there is significant pain, weakness in an arm or leg, rapid growth or hardening of the tumor.
An optic glioma is a tumor that can develop on the nerve that connects the eye to the brain (optic nerve). Without proper diagnosis and treatment, an optic glioma can cause problems with vision, even blindness. Yearly eye exams are the best way to monitor vision in children with NF1. 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 abnormalities seen in people with NF1 are usually present at, or just after, birth. These include a defect involving the bones of the eye socket (sphenoid dysplasia) and a problem with the way the lower leg bones develop (tibial dysplasia). Tibial dysplasia may appear as bowing of the lower leg, and can lead to a break in the bone (fracture), which can be difficult to treat.

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 their child is diagnosed. If you are concerned that you or your spouse may have features of NF1, please let your child’s NF doctor know.

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, or by genetic testing. 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 or more features of NF1. If your child does not yet have two features of NF1, NF1 genetic testing will confirm the diagnosis of NF1. However, if an abnormality (mutation) in the NF1 gene is not found, it does not exclude the diagnosis of NF1 in your child. Importantly, genetic testing does not necessarily help your child’s doctors predict which features of NF1 your child will develop.
Are there other medical concerns associated with NF1?

About half of all children with NF1 have a specific learning disability. In contrast, it is uncommon for a child with NF1 to have an intellectual disability (formerly referred to as “mental retardation”). The specific learning problems in children with NF1 are similar to the learning problems seen in individuals who don’t have NF1, and are managed in the same way (e.g., therapy services, individualized education plans, resource classes, and academic accommodations). In addition to specific learning disabilities, children with NF1 are at increased risk for Attention-Deficit/Hyperactivity Disorder (ADHD), Autism Spectrum Disorder (ASD), low muscle tone, and coordination difficulties.

In addition, children and adults with NF1 are often shorter than their classmates and other family members, and some children may develop a curvature of the spine (scoliosis). Less frequently, children with NF1 may experience seizures, high blood pressure, or a heart defect at birth. Some specific types of cancer are also slightly more common in children with 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 each phase of your child’s growth and development.

At birth, children with NF1 typically have a few café-au-lait spots, and problems with bone development, including sphenoid and tibial dysplasias, are identified. During infancy (0-2 years), these birthmarks typically increase in number and size. Delayed onset of major
developmental milestones (e.g., walking, first words) is also common during the first few years of life.

Children with NF1 may continue to develop more café-au-lait spots throughout early childhood (3-12 years of age), and most develop axillary or inguinal freckling during this time. Vision problems related to an optic glioma are commonly identified in early childhood, and approximately half of all children with NF1 will have Lisch nodules at 5 years. Plexiform neurofibromas are typically diagnosed in early childhood, but may go undetected if they are not visible or not causing any problems. Plexiform neurofibromas tend to grow most rapidly throughout childhood. Specific learning disabilities and behavioral problems including Attention-Deficit/Hyperactivity Disorder (ADD/ADHD) and Autism Spectrum Disorder (ASD) are also commonly identified at this age.

During late childhood and through adolescence (13-18 years), neurofibromas typically start to appear, and may gradually increase in number and size over time. Scoliosis may worsen during this time, and academic and behavioral difficulties may continue to persist.

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 whether he or she has developed any new features of NF1, and will do a physical exam to make sure there are no new medical concerns related to NF1.

Your child should have an annual eye exam performed 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 an optic glioma. Optic gliomas that affect eyesight may lead to blindness if they are undiagnosed and untreated. 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, as these could be signs of an optic glioma. While early onset of puberty may seem like an unusual indicator of optic glioma, it can sometimes be associated with the growth of an optic glioma. If there is any concern about your child’s vision or
early onset of puberty, your doctor will order a brain 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.

Most people with NF1 will develop neurofibromas at some point in their life, and there is currently no available treatment that prevents or slows the growth of neurofibromas. If your child develops pain or discomfort associated with a neurofibroma, it can be safely removed by asking your NF specialist for a referral to a surgeon experienced in the removal of neurofibromas.

If your child has a plexiform neurofibroma, you should call the doctor immediately if the plexiform neurofibroma becomes painful, starts growing quickly, changes the way it looks or feels, or if your child has a change in usual activity (such as weakness or persistent numbness of an arm or leg), as these can be signs that the plexiform neurofibroma is changing into a cancer. Unlike neurofibromas, plexiform neurofibromas are very difficult to remove, and surgery is generally not recommended.
What causes NF1?

NF1 is a genetic condition caused by a change (mutation) in the *NF1* gene. People are born with NF1.

Genes provide the instructions for how each one of us is made. We all have thousands of genes that influence how our body grows – from the color of our eyes to how our heart is shaped. Within these thousands of genes, there is the *NF1* gene. Everyone has two copies of this gene in each cell of their body. However, in people with NF1, one copy of the *NF1* gene is changed (mutated). When one copy of the *NF1* gene is changed, this causes NF1.

Half of all people with NF1 inherit a mutated *NF1* gene from an affected parent, while the other half of individuals are the first person in their family to have NF1. When a parent has NF1, there is a chance that they will pass on their mutated copy of the *NF1* gene. There is nothing a parent can do to control whether a child inherits the changed gene, and there is a 50% chance that the child will inherit NF1 from that parent.

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

NF1 arises in boys and girls equally, and does not skip generations. A child can be more or less severely affected than their parent. There is currently no way to predict what complications a child with NF1 will have based on their parent’s medical history.

What should I tell my child about NF1 and when?

It is important to develop open and honest communication with your child, and present information to them in a straightforward way. This will allow your child to feel comfortable coming to you with questions or concerns, and will promote trust between you and your child.

Your child will also take cues from you. If you are honest, while maintaining a positive attitude, it may ease any concerns your child may have about the diagnosis of NF1.
At some point, your child will probably want to know more about NF1, and may ask you to tell him or her about it. Young children can be told that NF1 is what causes their birthmarks (spots) and freckles. However, 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 specialist, rather than learning about it from online sources. It is also important to remember that the quality of information available on the internet varies. Some of the information may be scary, and is often inaccurate. Additional booklets, available on the Washington University NF Center website (nfcenter.wustl.edu) and from your care team at the NF Center Clinical Program at St. Louis Children’s Hospital, are available to aid you in this conversation.
As children enter their teens, they may become self-conscious about their appearance. Your child may notice that they are a little shorter than many of their classmates, and may worry about developing neurofibromas and how these will affect his or her appearance. It is important to emphasize that neurofibromas grow and develop slowly over years, not within a period of a few days or even weeks, and that people with NF1 are still attractive, get married, and have children, just like people who don’t have NF1.

If your child asks a question that you can’t answer, please do not hesitate to contact your NF care team. They will answer any questions that you or your child may have.

Below are some questions that your child or teenager may ask about their diagnosis of NF1.

Q: Will I be covered with neurofibromas?

A: There is no way to predict how many neurofibromas you might develop in your lifetime. Any neurofibroma that is bothersome can be safely removed by an experienced surgeon.

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

A: There is nothing that you do that makes your neurofibromas grow. Neurofibromas often appear or grow in size during times of hormone changes, such as puberty (which you can’t avoid) and pregnancy. There is no medication currently available that can prevent the neurofibromas from growing, and common things, like caffeine, exercise and smoking, do not cause an increase or decrease in the number or size of neurofibromas.

Q: Will I develop cancer or die from NF1?

A: There are some complications associated with NF1, including certain cancers that are slightly more likely to develop, but these are still pretty rare.
Q: Neither of my parents has NF1. Does this mean I’m adopted?

A: No. Only half of all people with NF1 have a parent who has NF1. The other half are the only people in their family to have NF1. Since neither of your parents have NF1, your NF1 was caused by a new change that happened in your NF1 gene before you were born.

Q: Should I tell my friends I have NF1?

A: 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, who will understand, and who will be supportive if you tell them. It is perfectly acceptable to tell 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:

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

A: Your child’s doctor should take a detailed family history to determine if either parent has NF1. When a parent has NF1, there is a 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%.

Q: Is there a way to fix the gene?

A: 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 this 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.

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

A: Studies have shown that there is no difference in outcome between those children whose optic glioma was diagnosed on a screening MRI compared to those children who were diagnosed following an abnormal eye exam. Performing brain MRIs in young children (under 7 years old) is also not without some risk, since many children will require sedation during the procedure.

Q: Should I tell my child’s teacher that my child has NF1?

A: Approximately half of all children with NF1 have a learning disability, and many have attention deficits. If you and your child’s teacher feel
that your child has a learning disability, formal testing should be done. This may include tests to identify academic strengths and weaknesses. If a learning problem 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 to maximize your child’s long-term success in school.

**Q: Should I limit my child’s activities because they have NF1?**

**A:** 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 activities will show your child how they are more like their friends than they are different from them.

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

**A:** There is active research focusing on NF1 at many universities and hospitals around the country, including the Neurofibromatosis Center at Washington University. Some of this research is performed in research laboratories to learn more about the basic biology of NF1, while other research involves patient participation through clinical trials. Clinical studies, including DNA and tissue repositories, are designed to learn more about how NF1 affects children and adults, while clinical trials are 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.

nfcenter.wustl.edu

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