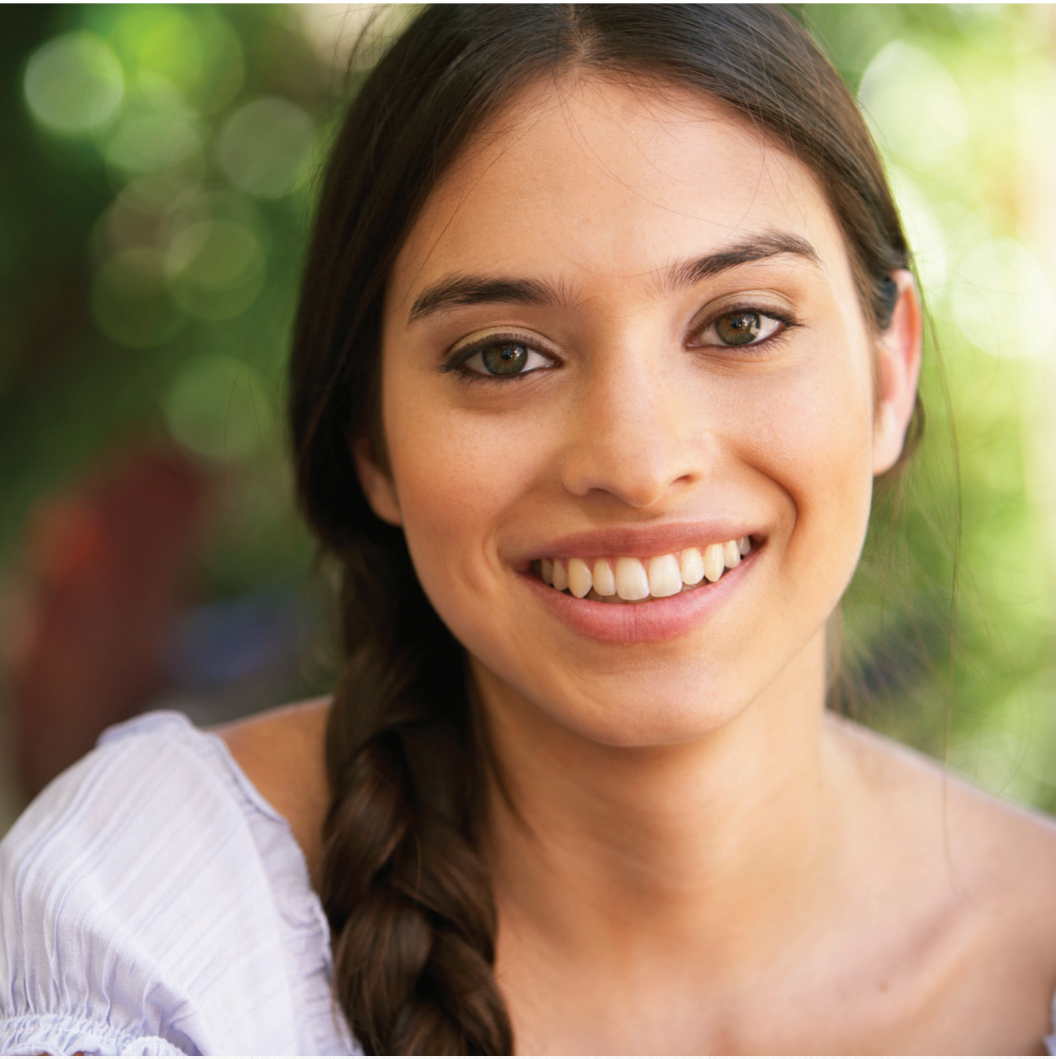
LIVING WITH NEUROFIBROMATOSIS TYPE 1:

A GUIDE FOR ADULTS

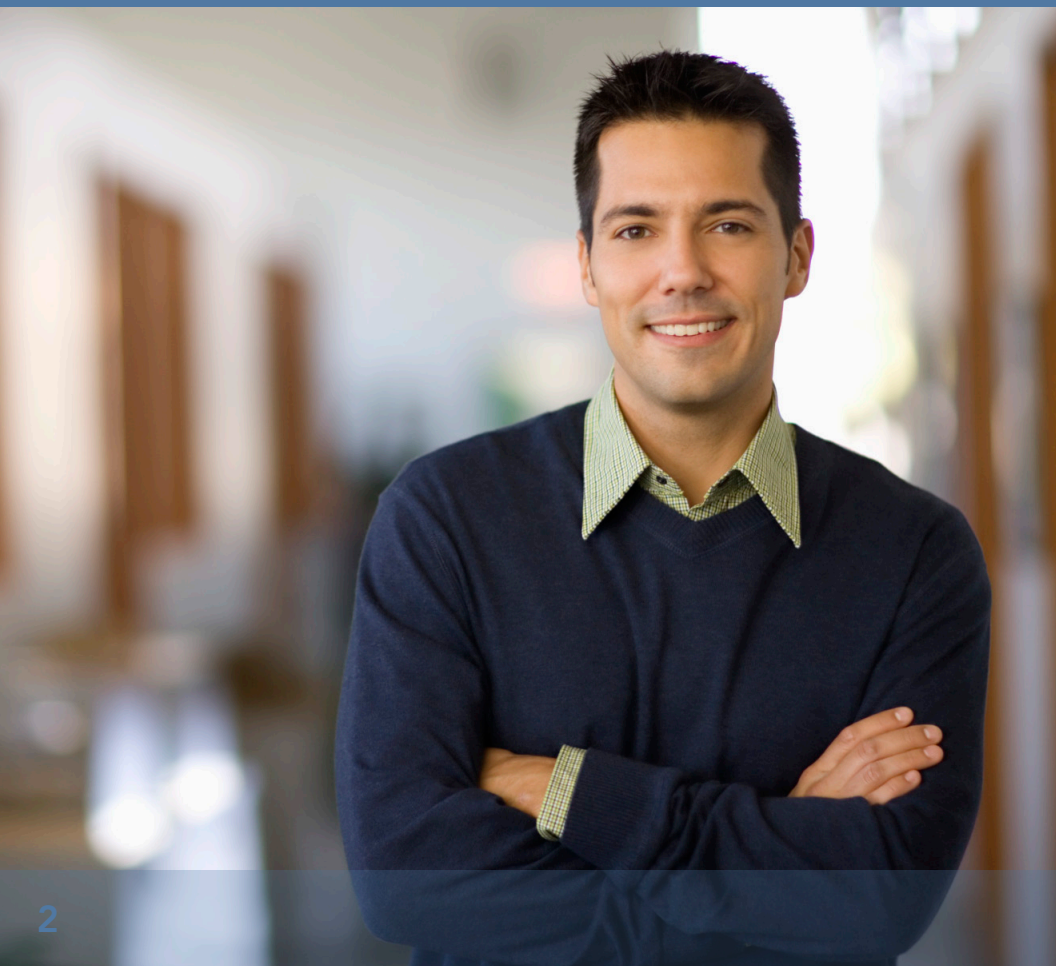


Children's HOSPITAL • ST. LOUIS
BJC HealthCare

Washington University in St. Louis
Physicians

Neurofibromatosis (NF) is a set of genetic conditions that affect nearly every organ system in the body, including the nervous system (brain, spinal cord, and nerves). There are three distinct types of NF, referred to as NF1, NF2, and Schwannomatosis. Each of these types represents a completely different condition, and people with NF1 do not develop NF2.

NF1 is the most common of the three conditions and affects approximately 1 in 3,000 individuals worldwide. Individuals with NF1 can develop a wide variety of clinical problems, ranging from bone deformities, raised blood pressure, and attention deficit to nerve tumors (neurofibromas), rare cancers and brain tumors (gliomas). The specific medical concerns that affect children with NF1 are frequently different from those experienced by adults. This booklet is designed to address questions and concerns unique to adults with NF1.



TUMORS AND CANCER

Tumors are common in adults with NF1. The majority of these tumors are benign growths, and are not cancers (malignant tumors). The types of benign tumors seen in people with NF1 include neurofibromas and plexiform neurofibromas. Malignant peripheral nerve sheath tumors (MPNSTs) and brain tumors (malignant gliomas) are less frequent.

Neurofibromas are small benign tumors associated with nerves. They can arise on the skin, underneath the skin, or deep within the body. The tumors that form on the skin are benign and do not become cancers. Neurofibromas may be first noticed during childhood, but most commonly appear during teenage years. Their appearance and growth often coincides with periods of hormonal change, such as puberty and pregnancy. Many women will report that the number and size of their neurofibromas increases during pregnancy. Typically, adults with NF1 will develop more neurofibromas over time. They may grow for a period of time and then stop growing. In addition, they may change in shape or color as they grow. Sometimes, they can also be associated with itching or slight discomfort when bumped. These are not signs of cancer development.

Unfortunately, there is no way to determine how many neurofibromas a person with NF1 may develop or where on the body they will appear. They can appear anywhere there are nerves – on the face, the scalp, the chest, as well as inside the body. There is nothing a person can do to prevent the formation or growth of neurofibromas. Certain foods, smoking, caffeine, and heat do not stimulate their growth.

The large number of neurofibromas that can develop in people with NF1 may have a significant psychological impact on that individual's self-esteem, relationships, and social interactions. Neurofibromas that grow along the belt or bra line, on the hands and forearms, or on the ankles or feet may be painful, bleed, or become infected.

In situations when select neurofibromas cause problems, some individuals choose to have them removed either by conventional surgery or using a procedure called electrodessication. With electrodessication, an electrical current is applied to the neurofibromas through a needle point tip. This technique is less invasive than traditional surgical removal methods, and allows your doctor to treat large numbers of neurofibromas at a single time.

In all cases, neurofibromas should be removed by a doctor who has experience with NF1. It is important to discuss your expectations prior to surgery, as some neurofibromas may grow back after surgery.

Plexiform neurofibromas are another type of neurofibroma. However, these tumors are frequently larger and involve multiple nerves. Plexiform neurofibromas can arise just underneath the skin (superficial), deeper within the body including the eye socket (orbit), neck, and pelvis (hip area) or along the spine. These tumors typically develop in childhood and can be painful if they are hit or bumped. Depending

on their location, plexiform neurofibromas can also lead to abnormal bone growth or pressure on nerves, blood vessels, and other important structures in the body. For example, plexiform neurofibromas along the spine can press on the spinal cord or nerves and lead to weakness or pain, or cause curvature (scoliosis) requiring surgery. Plexiform neurofibromas found in the neck or pelvis can cause swallowing or breathing problems or difficulties with urination, respectively.

Current treatments for plexiform neurofibromas include surgery and chemotherapy. Radiation therapy is not recommended for these tumors, as it can lead to cancer development. Surgery can reduce the overall size of the tumor, but will not stop the tumor from growing. Over the past several years, new cancer drugs have been used to slow the growth of plexiform neurofibromas. You should discuss the treatment options with your NF1 specialist.

Unlike the neurofibromas, some plexiform neurofibromas can turn into cancers, called *peripheral nerve sheath tumors* (MPNSTs). Some MPNSTs are highly aggressive cancers, which can grow rapidly and spread to other regions of the body (bone and lung). These aggressive cancers are frequently difficult to treat with conventional therapies. **Individuals with NF1 who have a plexiform neurofibroma should immediately let their doctors know if they develop a new persistent pain or a change in their usual physical abilities and activity (weakness, numbness, and problems with bladder or bowel function).** You should also call your NF1 specialist if your plexiform neurofibroma grows rapidly, changes in shape or color, or feels harder. These are often signs that the plexiform neurofibroma is turning into a cancer.

Brain tumors in adults with NF1 are much less common than those arising in children with NF1. Whereas 15-20% of children with NF1 will develop an optic glioma (brain tumor along the nerve that carries vision from the eye to the brain), adults with NF1 are at a higher risk of developing malignant brain tumors (gliomas). While gliomas are more common in individuals with NF1 than in people without NF1, they are still rare tumors, seen in fewer than one in one hundred adults with NF1. Signs of these types of brain tumors in adults include new seizures, persistent headache, weakness, numbness, or a change in thinking. If you notice these problems, you should contact your NF1 specialist.

LEARNING AND SPEECH PROBLEMS

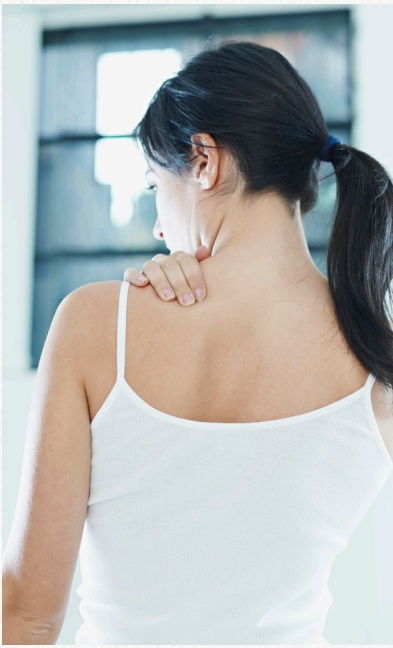
Some people with NF1 may have been diagnosed with a learning disorder as a child. These learning disabilities are not progressive and will not worsen over time during adulthood. Some adults have difficulties with reading, writing, concentrating, coordination, and finding their way around. They may experience problems in filling out application forms for jobs or for social services. Individuals should consult their doctors for adult literacy programs and support networks available to help with these difficulties.

Some adults with NF1 may have had speech problems in childhood, which may continue into adulthood. There is no evidence that speech problems get worse in adulthood, and they are usually mild. Individuals may have difficulty in adjusting the rate, volume, and tone of their speech, and sometimes their speech has a nasal quality. Speech therapy can improve these problems.

OTHER ASSOCIATED MEDICAL CONDITIONS

High blood pressure, also called hypertension, can be seen in people with NF1. Similar to adults without NF1, it is important to have your blood pressure checked at least once a year by your doctor. Sometimes, high blood pressure is not specifically related to NF1. However, in rare situations, individuals with NF1 can have hypertension caused by a tumor that secretes blood pressure-raising hormones, called a pheochromocytoma. In some adults with NF1, high blood pressure can result from problems with the large blood vessel from the heart (aorta) or with the blood vessels to the kidney (renal arteries). Regardless of the cause, high blood pressure is treatable.

Strokes are known to occur at an increased frequency in people with NF1. Blood vessels in the brain can be damaged, particularly in people who were treated with radiation therapy for a brain tumor. If you experience sudden weakness, numbness of your limbs or face, change in your speech, balance or vision, you should call 911 and go immediately to the emergency room.



Some adults complain of *chronic pain.* Back pain is sometimes related to neurofibromas along the spine. However, in most situations, surgery to remove these spinal neurofibromas will not alleviate the pain. For adults with severe and chronic pain that has been fully investigated, a referral to a doctor who specializes in pain management can sometimes be helpful to reduce the degree of pain you are experiencing. Physical therapy has also proven beneficial in some individuals with NF1 and back pain.

Bone problems can also be seen in adults with NF1. In this regard, adults with NF1 are prone to reduced Vitamin D levels, which can lead to low bone mineral densities, osteoporosis and abnormal bone fractures. Supplementation with Vitamin D may improve general bone health in some adults with NF1. In contrast, curvature of the spine (scoliosis) sometimes seen in growing children, is not a feature that starts in adulthood. Scoliosis diagnosed in childhood will persist in adulthood and can occasionally cause problems.

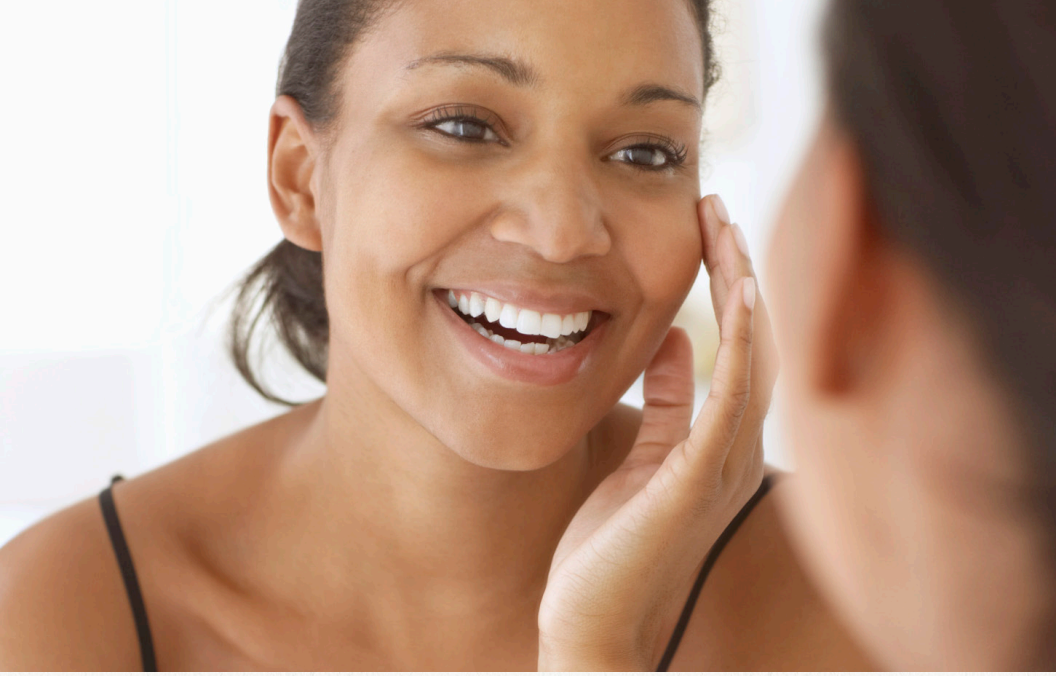
STARTING A FAMILY

Marriage and starting a family are important issues to consider for adults with NF1. Ultimately you and your partner will decide what is best for you and your family. It is therefore recommended that you know as much as you can about NF1. You may wish to have your fiancée or spouse come with you during your visit to the NF specialist to learn more and to get their own questions about NF1 answered.

There are several ways in which NF1 can affect a pregnancy. First, women with NF1 who become pregnant may experience an increase in the development and growth of their neurofibromas. In addition, if you have a plexiform neurofibroma which is growing rapidly, it will be difficult to obtain radionuclide imaging studies (PET scans) during pregnancy. Second, some women with NF1 may have additional problems with their pregnancy and delivery than women without NF1. If there are internal neurofibromas, rarely these can interfere with the growth of the baby in the uterus (womb) or cause problems during delivery (if close to the uterus or pelvic floor where the vagina and uterus come together). In these situations, you and your obstetrician may decide to deliver the baby by Caesarian section (C-section). You should discuss these potential issues and options with your NF1 specialist and obstetrician.

Third, there is a 50/50 chance with each pregnancy that you and your partner will have a child with NF1. You may wish to speak with a genetic counselor before deciding to have children to help you better understand the chances of having a child born with NF1. Genetic counselors help by providing additional information, but will not tell you and your family what you should do. There are many options now for families who want to make decisions about having a baby with NF1. Genetic testing can now be performed prior to implantation (before the fertilized egg attaches to the uterus and starts growing) or during early pregnancy. These genetic tests require that the mother or father with NF1 have their DNA tested for a change in the *NF1* gene. This testing may or may not be covered by your insurance. You should ask your NF specialist about NF1 genetic testing.

While these genetic tests are accurate, they cannot tell you how severely affected a child with NF1 may be. Just because the child may be born with NF1, it is not currently possible to know whether they will have more or fewer medical problems than their affected parent has. If you decide to have children, it is very important that your NF1 specialist examines the newborn child during the first 3 to 6 months of life to check for café-au-lait macules or other features of NF1.



PHYSICAL APPEARANCE AND SELF-ESTEEM

During the transition to adulthood, many individuals may become more self-conscious about their appearance. Since neurofibromas tend to appear more frequently during adulthood, some people may be embarrassed by their physical appearance. Although most adults will develop neurofibromas on their skin, severe disfigurement is not common. Plexiform neurofibromas can sometimes be disfiguring depending on their size and location on the body.

It is always important to remember that NF1 is not your identity, and will never be. You define who you are and who you will become. The interactions with those who care about you will help you with these issues. If you feel that there are aspects of NF1 that are taking more of a toll on your life than you can handle, be sure to discuss these concerns with your NF1 specialist. You may find it difficult not knowing how your NF1 will progress as you get older. Though there is no cure for NF1, there are new treatments and procedures that might help minimize some of the features of NF1. It is important to remember that you are so much more than NF1, and that you are not alone. Talk to your NF specialist for resources in your area.

SCREENING RECOMMENDATIONS

It is recommended that adults with NF1 have evaluations at least every one to two years by a specialist expert in NF1. You should also see your regular doctor (internist and gynecologist) for routine health care, including yearly blood pressure assessments. Your NF1 specialist will also check your blood pressure, but will additionally perform a detailed physical and neurologic examination. MRI exams are not part of the usual recommendations unless there is a specific concern or problem.

ORGAN AND BLOOD DONATION

People with NF1 can donate their blood and organs to other people who are in need of blood or organ transplants. The person who receives their blood and/or organs will not develop NF1 as a result of the donation.

INSURANCE

Occasionally people will have difficulty with insurance coverage. Individuals with NF1 should see their NF1 specialist on an ongoing basis. Insurance can become an issue because NF1 is a chronic condition and insurance carriers tend to limit coverage for chronic conditions. If you are planning on changing your insurance, you should check with your insurance carrier or future employer about your coverage. Additionally, there is a federal program entitled COBRA (the Consolidated Omnibus Budget Reconciliation Act) that gives workers and their families who lose their health benefits the right to choose to continue group health benefits for limited periods of time under certain circumstances. Other state and federally funded programs exist to provide additional support, but do have some exclusions and income limits.

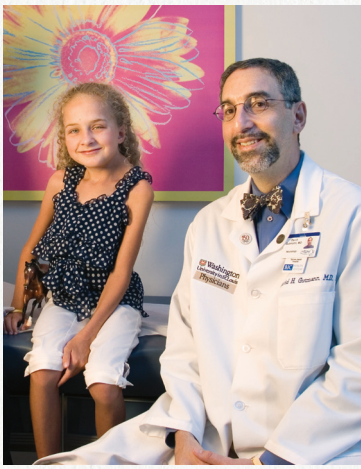
RESEARCH AND CLINICAL TRIALS

A clinical trial is a research study in which a person participates to help doctors learn more about a particular treatment or way of looking at a medical problem. In a clinical trial, the treatment/evaluation has not yet been proven to work, but the trial is an important step in finding out whether it will be beneficial in the treatment of NF1-related medical problems. The doctor directing the clinical trial will review the potential

benefits and risks before a person agrees to participate. Everyone has the right to decline participation.

Clinical trials for NF1 are listed on the National Institutes of Health website (www.clinicaltrials.gov).

FINDING A DOCTOR



It is important that people with NF1 see a doctor who can answer their questions and who knows about NF1. For help finding a physician call 314.454.KIDS or visit www.StLouisChildrens.org. The websites for the Washington University Neurofibromatosis Center (http://nfcenter.wustl.edu), Children's Tumor Foundation (www.ctf.org) and Neurofibromatosis, Inc. (www.nfnetwork.org) have information about nearby doctors, medical centers, and NF resources for advice and treatment of people with NF1.

OTHER RESOURCES

Washington University Neurofibromatosis Center
http://nfcenter.wustl.edu

St. Louis Children's Hospital
StLouisChildrens.org

Children's Tumor Foundation
www.ctf.org

Neurofibromatosis, Inc.
www.nfnetwork.org

The National Council on Independent Living
Provides information and resources on employment, housing, transportation, and civil rights for individuals with disabilities.
www.ncil.org

Research Clinical Trials
www.clinicaltrials.gov

ST. LOUIS CHILDREN'S HOSPITAL

Neurofibromatosis Center
One Children's Place | St. Louis, Missouri 63110

314.454.KIDS (5437) | http://nfcenter.wustl.edu
StLouisChildrens.org

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