



LIVING WITH
NEUROFIBROMATOSIS TYPE 1:

A GUIDE FOR ADULTS

CHILDREN'S
TUMOR
FOUNDATION
ENDING NF
THROUGH RESEARCH

This resource serves as a general guide for adults with neurofibromatosis type 1 (NF1) and is not meant to replace treatment or ongoing care from a doctor or NF specialist.

Neurofibromatosis type 1 is a genetic condition that occurs in 1 in 2,500 individuals, affecting more than 4 million people around the world. Many people living with NF1 are healthy and do not experience serious medical issues. For some people, however, more significant health issues may occur. It is important to know the common manifestations of NF1, as well as the various complications and concerns that adults with NF1 can face.

Welcome Message from the Children's Tumor Foundation

The Children's Tumor Foundation (CTF) is a global non-profit organization dedicated to improving the health and well-being of children and adults affected by all types of NF, which refers to all types of neurofibromatosis and schwannomatosis.

OUR MISSION: Drive research, expand knowledge, and advance care for the NF community.

OUR VISION: End NF.

We are called the Children's Tumor Foundation because NF1 is most often diagnosed in childhood. However, NF1 is a lifelong condition, and CTF is committed to finding treatments and offering support for all who live with NF, young and old, throughout the lifespan.

We know that living with NF is a journey, and we hope to provide guidance and support every step of the way. Neurofibromatosis type 1 affects each individual differently, just like each individual with NF is unique separate from the diagnosis. Often patients with visible signs of NF struggle to be seen as more than just their NF, while patients whose NF is invisible sometimes struggle to make others understand.

We are here to support you throughout this process, and we hope this resource, along with our other resources, will offer insight and guidance. Thank you for letting us join you as you navigate your unique path with NF1.

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Adults with NF1

This resource serves as a supplement to the Children's Tumor Foundation booklet, **Diagnosed with NF1: A Guide for Those Living with NF1**, which is available to read or request at ctf.org/education.

Often NF manifestations, or symptoms, change over time, and there are important considerations specifically for adults living with NF1.

Tumors and Cancer

One of the most common features of NF1 is the presence of tumors (increased division of cells). A majority of NF1 tumors are benign (non-cancerous) rather than malignant. Tumors can be located inside or outside the body, including cutaneous neurofibromas, plexiform neurofibromas, and brain or spinal tumors (gliomas). Additionally, NF1 can be associated with other types of tumors, such as tumors of the nailbeds (glomus tumors, which are involved in temperature control), the neuroendocrine system (pheochromocytoma, which develop in adrenal glands), and the gastrointestinal system (gastrointestinal stromal tumors, or GIST). NF1 is also associated with an increased risk of breast cancer.

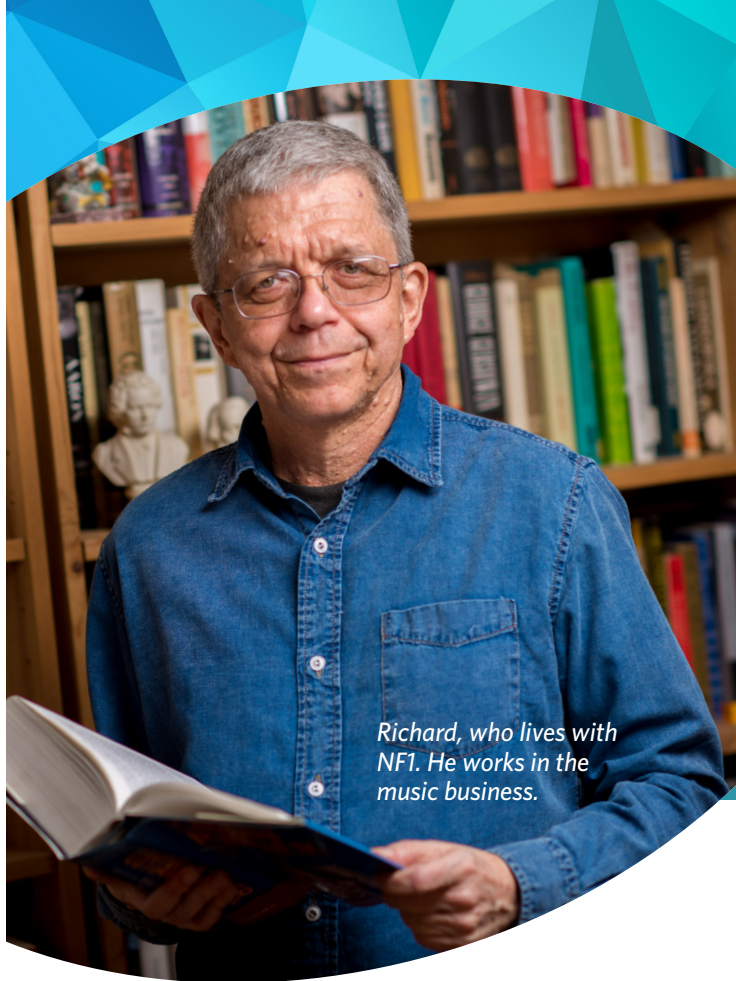
Cutaneous Neurofibromas

Cutaneous neurofibromas are the most common type of neurofibroma. These are benign tumors that develop along a nerve, on or under the skin, and appear as small lumps, bumps, or nodules. Cutaneous neurofibromas usually first appear during puberty. Typically, adults with NF1 will develop more cutaneous neurofibromas as they age, as well as during pregnancy. There is no way to predict when, where, or how many neurofibromas a person will develop over the course of their life. Some people with NF1 may have only a few neurofibromas, while others may develop hundreds.

Cutaneous neurofibromas may be itchy, tender, or affect a person's self-image. They may occasionally bleed. Some people choose to have them removed. Current treatment options include surgery, laser, and electrodesiccation. Treatment should be performed by a provider familiar with NF1. At this point, there are no proven methods to prevent neurofibroma development.

Plexiform Neurofibromas

Plexiform neurofibromas are another type of tumor that may impact people with NF1, visible in 30%, and present on imaging in over 50% of affected individuals. These tumors are different than cutaneous neurofibromas because they can grow to be quite large. They may cause pain, disfigurement, bleeding, and impaired function. Unlike cutaneous neurofibromas, plexiform neurofibromas are thought to be present at birth and grow slowly over time, but more rapidly in childhood than in adulthood. They can be located inside the body or more externally visible on the surface of the skin. They may have a mixed or lumpy texture, sometimes with hair growth over the affected area. Most importantly, plexiform neurofibromas have the potential to become a malignant cancer, and should be monitored closely.



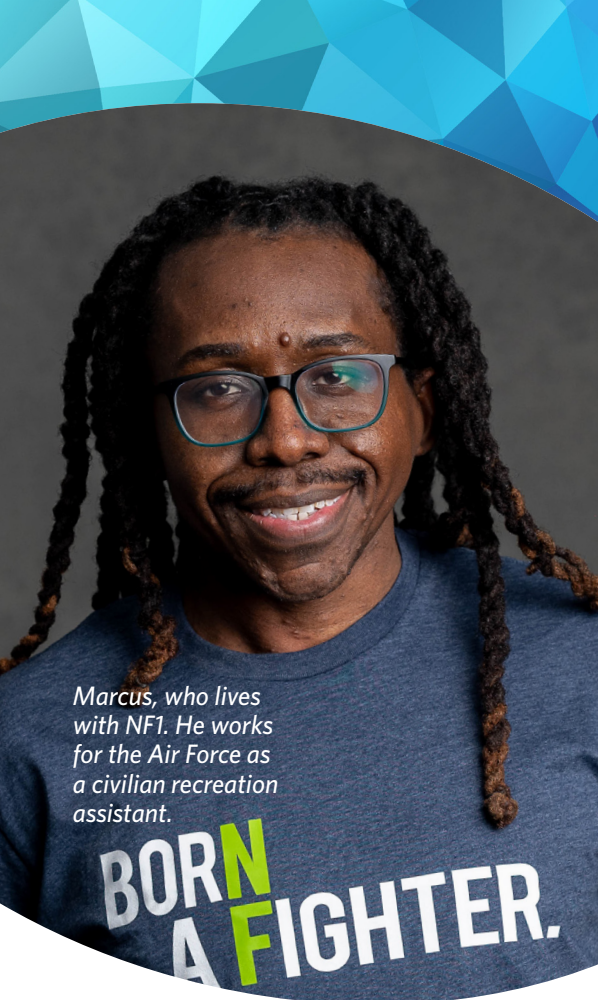
Richard, who lives with NF1. He works in the music business.

Sometimes surgery or other interventions are recommended to manage a plexiform neurofibroma, especially if it is large or painful, or has developed changes consistent with malignancy. Surgical intervention and treatment for plexiform neurofibromas should be provided by doctors familiar with NF1.

Individuals with NF1 are also at an increased risk of developing a malignant peripheral nerve sheath tumor (MPNST), most often occurring within a plexiform neurofibroma. These are very concerning tumors and may be difficult to treat. Therefore, it is important to recognize any new or acute changes in symptoms.

“Red Flag” symptoms to promptly alert your healthcare provider about:

- new persistent or progressive pain
- pain that wakes you from sleep
- change in physical abilities or function, such as weakness, numbness, change in bladder or bowel function, or swallowing and breathing problems
- rapid growth of a portion of a pre-existing plexiform neurofibroma
- change in shape or texture of a pre-existing plexiform neurofibroma (becomes hard)
- associated weight loss, fatigue, and malaise



Marcus, who lives with NF1. He works for the Air Force as a civilian recreation assistant.

Gliomas

Adults with NF1 can develop brain or spinal tumors called gliomas. The most common type of glioma seen in adults with NF1 are low-grade gliomas, which are typically slow growing (low-grade) and benign. They can cause symptoms and may require periodic monitoring with scans. Adults with NF1 can also develop malignant gliomas (including glioblastoma multiforme). Adults with NF1 who experience unexplained headaches, or a change in their usual physical abilities or function (such as weakness, numbness, a change in personality, or new onset seizure) should promptly notify their NF specialist.

Breast Cancer

Women with NF1 have an increased risk of developing breast cancer, which may occur at an earlier age than in the general population. Current guidelines advise yearly screening mammography starting at age 30 with consideration of contrast-enhanced breast MRI between the ages of 30-50. After age 50, the risk of breast cancer in women with NF1 becomes similar to that of the general population. At that time, the screening recommendations are the same as for the general population.

Other NF1-Associated Tumors

Other tumors seen more often in people with NF1, relative to the general population, include:

- **Glomus tumor.** A benign tumor of the nail bed affecting the fingers and toes. These tumors can be multiple and very painful, especially when bumped, and are sensitive to heat and cold. Treatments include pain medication and surgery.
- **Gastrointestinal stromal tumor (GIST).** A tumor of the gastrointestinal tract that can cause bowel disturbances, bleeding, abdominal pain, and appetite or weight changes. Management typically includes surveillance or surgery.
- **Pheochromocytoma.** A tumor of the adrenal gland that can cause high blood pressure, anxiety, headache, sweating, and rapid heartbeat. Treatments include blood pressure management and surgery.

Other NF1 Manifestations

Lisch Nodules

The majority of adults with NF1 have Lisch nodules. These are small raised bumps on the iris (colored part of the eye). Lisch nodules do not affect vision or cause any problems, but are unique to NF1 and may be useful for confirming a diagnosis. These should not be confused with optic pathway gliomas which may affect vision during childhood.

Bones

Adults with NF1 have an increased risk of osteoporosis and fracture due to decreased bone mineral density. Many NF providers recommend that individuals with NF1 take a daily Vitamin D supplement. Occasionally, scoliosis from childhood can progress in adulthood, and people with severe scoliosis may have breathing issues, or develop weakness in their limbs due to pressure on the spinal cord. These problems should be monitored by an NF specialist and an orthopedic surgeon familiar with NF1.

High Blood Pressure and Blood Vessel Abnormalities

Adults with NF1 are at risk for high blood pressure (hypertension) and are advised to monitor their blood pressure regularly, and are therefore at increased risk for cardiovascular disease and stroke. There are many causes of hypertension; however, in individuals with NF1, there may be specific NF1-related causes that require additional evaluation and may need to be treated differently. High blood pressure can be caused by narrowing of the vessels near the kidneys (renovascular hypertension) or by the presence of pheochromocytoma (see "Other NF1-related tumors").

Itching

Itching is a frequent complaint in people with NF1. Although not fully understood, it may be related to the presence of mast cells (inflammatory cells that release histamine). Itching may be localized, such as in the area of a neurofibroma, or more generalized (occurring throughout the body). Individuals with bothersome itching should contact their NF providers.

Seizures

Seizures (epilepsy) are seen slightly more often in people with NF1 than in the general population. Sometimes medication is required to control the seizures, and some individuals require follow up with an epilepsy specialist. Anytime there is a new onset or unexplained seizure in an adult, prompt evaluation is needed.

Pain

Some adults with NF1 experience chronic pain, and the source and intensity of the pain may vary. Back pain is sometimes related to scoliosis, or can result from neurofibromas of the spine. Headaches and migraines are common. Neurofibromas may cause pain in any area of the body.

New or worsening pain or change in headache frequency or intensity can be a warning signal of complications in NF1. For adults with severe and chronic pain that has been fully investigated and has not responded to other treatments, a referral to a doctor who specializes in pain management can be helpful in reducing the degree of pain experienced. It is important to remember that not all pain in individuals with NF1 is related to NF1.

Learning in NF1

NF1 can be associated with learning disabilities and/or attention difficulties. These issues usually are diagnosed in childhood, and can last throughout life. Learning and attention issues can affect concentration, communication skills, organization, time management, and employment opportunities. Supportive therapies may be helpful in adulthood to optimize success. Some individuals benefit from discussing their concerns with a social worker, psychologist, or vocational therapist. Learning and attention issues are managed and treated with the same approaches as individuals who do not have NF1.

Sleep

Some individuals with NF1 report sleep problems and daytime sleepiness. Causes of sleep disturbances may include anxiety, depression, pain, or side effects of medications. Occasionally, neurofibroma tumors in the neck region can disrupt sleep and cause loud snoring.

Depression and Anxiety

Many individuals with NF1 struggle with concerns related to mental and emotional well-being. This may be related to feelings of isolation, having a chronic medical condition, or persistent pain. Symptoms include feeling down, having low energy, not experiencing joy, crying, moodiness, and feeling on edge. Sometimes these feelings interfere with sleep or appetite. A psychologist, psychiatrist, or counselor may assist with coping, and other strategies have been helpful as well. We encourage adults living with NF1 to seek help and support through family and friends, support groups, or professional counseling services.



Salina, who lives with NF1. She works as a document control manager.

Physical Appearance

Some adults worry about changes in their physical appearance as a result of their NF1. Although most adults will develop neurofibromas on their skin, significant disfigurement is not common, and people vary in how they experience physical differences. NF1 is not the "Elephant Man's disease," which is a rare and an entirely different condition. Plexiform neurofibromas can sometimes be visibly noticeable depending upon their size and location. Curvature of the spine, called scoliosis, can be seen in growing children, but it is not a feature that usually starts in adulthood. Some people experience distress from cutaneous neurofibromas that are visible on the face or other body areas. Interventions are available to treat cutaneous neurofibromas (see "Cutaneous Neurofibromas"). Discomfort with one's appearance can have a profound impact on individuals and their relationships, so it is important to seek help to work through these challenges if they occur.

Pregnancy and Family Options

A diagnosis of NF1 does not affect the ability to conceive a child. Although many women with NF1 do not have medical issues during pregnancy, some report an increase in the number of neurofibromas and/or enlargement of already existing neurofibromas. Some women experience elevated blood pressure or other complications, and women with NF1 have a higher cesarean section rate than women without NF1. Women should let their obstetricians know that they have NF1 and also inform their NF1 doctors when they become pregnant.

When a couple decides to start a family, if one of the parents has NF1, each child will have a 50% chance of also having NF1. It is important to remember that, because NF1 is so variable, a child may or may not have the same medical problems as their parent. Talking with a genetic counselor or an NF specialist before conception is recommended to help individuals with NF1 better understand the chances of having a child born with NF1 and the spectrum of medical concerns associated with NF1.

Some individuals with NF1 may want to review all family planning options, which might include genetic testing. Many couples have successful pregnancies resulting in healthy children. Any family planning decision is a personal one, and entirely up to the couple; however genetic counselors can assist in the decision-making process. More information about the genetics of NF1 and reproductive options can be found at ctf.org/education.

Children of individuals with NF1 who are at risk for NF1 should be evaluated for NF1 features by a doctor specializing in NF1 in the first few months after birth, and by an ophthalmologist beginning around one year of age.

Samantha, who lives with NF1. She is a business development manager and mom.



NF1 Screening Recommendations

In 2018, a resource was published to provide physicians with guidance on caring for individuals with NF1. It may be useful for individuals with NF1 to share this document with their healthcare providers. The publication can be accessed free of charge at ctf.org/acmg.

It is usually recommended that adults with NF1 have medical evaluations once a year or more by a doctor who knows about NF1. Annual physical evaluations should include a detailed physical exam including spine evaluation, blood pressure test, and a neurologic assessment. Vitamin D levels may be checked and additional supplementation prescribed. In addition, any concerning or changing symptoms should be reported, and psychologic well-being should be assessed. Depending on age and life stage, family planning and reproductive options should be discussed for personalized care.

Scans and Imaging in NF1

While there is not a universal recommendation, many major medical institutions recommend baseline scanning in adults with NF1 to evaluate for the presence of NF1 tumors; other providers recommend scanning only when there is a specific concern.

Because NF1 is associated with tumor formation and increased risk for certain types of cancers, radiation exposure should be limited as much as possible. Magnetic resonance imaging (MRI), which does not expose a person to radiation, is typically preferred over computed tomography (CT) or X-rays. Other types of NF1-related imaging may include positron emission tomography (PET scan) to evaluate for cancer; dual-energy X-ray absorptiometry (DEXA scan) to evaluate bone health; and mammography (see “Breast Cancer”).

Other topics related to NF1

Organ, Blood, and Body Donations

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

Insurance

Occasionally people have had difficulty managing their insurance and healthcare costs. Some individuals qualify for disability benefits. Individuals should contact their NF1 specialists or a social worker who can assist with available programs.

NF1 and the Military

Unfortunately, because NF1 is considered a chronic neurologic condition, it is considered a disqualification for enlistment into all branches of the military.

NF1 Treatments

Currently, there is no cure for NF1, but there are treatments for the various manifestations seen in NF1. Interventions for cutaneous neurofibromas include surgery, electrodesiccation, and/or laser treatments. One should be aware that there is a chance of regrowth, the possible need for multiple treatments, and that these procedures may not be covered by insurance. A group of medications called MEK inhibitors (such as selumetinib or mirdametinib) are used to reduce the size and effects of plexiform neurofibromas, and other NF1-related tumors. See MEK inhibitor information for patients at ctf.org/mek. These medications require close monitoring for side effects. Individuals should consult with their NF care provider for personalized medical care.

NF Research, Clinical Trials, and the NF Registry

Researchers from around the world seek to learn more about NF. Many areas of research focus on the tumors in NF and understanding the underlying mechanism of tumor growth, as well as exploring different medications to treat NF tumors. Other areas of NF research include studies looking at how individuals with NF learn, develop, and cope with their condition.

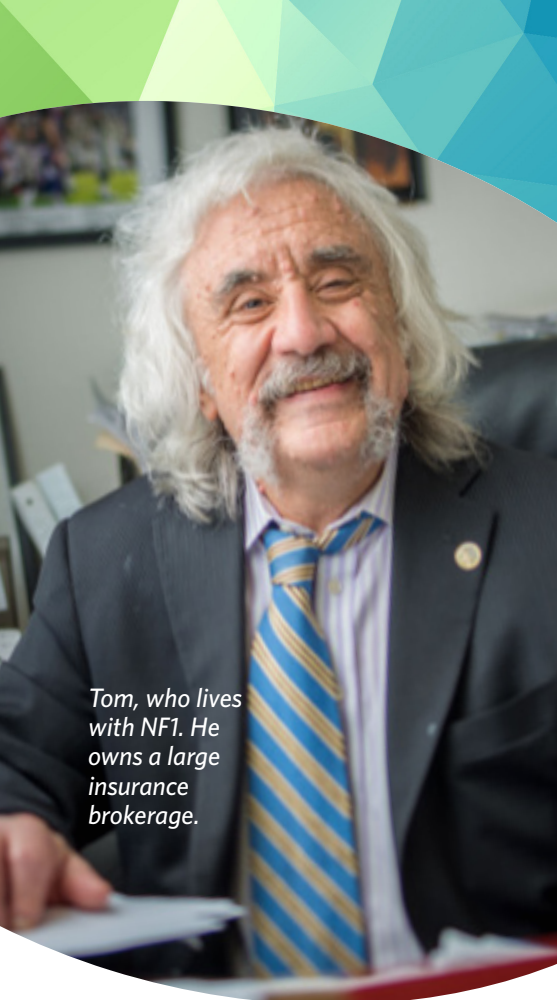
A clinical trial is a scientific study in which a person participates to help researchers learn more about a particular treatment or way of looking at a medical issue. In a clinical trial, the treatment/evaluation has not yet been proven to be effective, but the trial is an important step in finding out whether it will be beneficial in the treatment of NF1-related medical problems. The researcher directing the clinical trial will review the potential benefits and risks before a person agrees to participate.

The CTF NF Registry is the largest worldwide database of individuals with NF, designed to speed the development of promising treatments and expand our knowledge about NF. Individuals register online and provide information about their diagnosis, symptoms, and treatment. Participants can decide whether or not they wish to be notified about clinical trials or other studies. Individuals should discuss the possibility of participating in a study with their NF provider. Current studies and clinical trials are listed at **clinicaltrials.gov**.

For more information about the NF registry, go to **nfregistry.org**.



Ken, who lives with NF1. He is a lawyer in New York City.



Tom, who lives with NF1. He owns a large insurance brokerage.

Find a Doctor







It is important that people with NF1 see a doctor or healthcare provider who is familiar with NF1 and can answer their questions. To locate an NF clinic or a specialist familiar with NF, please go to the Children's Tumor Foundation Find a Doctor page at ctf.org/doctor.

Children's Tumor Foundation Resources and Support

It is estimated that more than four million people worldwide are living with NF, and the Children's Tumor Foundation is committed to providing informational resources and support that will help guide you on your journey with NF.

Below are some of the many ways that CTF supports adults living with NF:

- Organizes a social gathering for adults with NF at the NF Summit
- Hosts an "Adults with NF" Facebook group, moderated by a trained CTF volunteer
- Highlights stories of adults with NF in our "This is NF" and "Make NF Visible" photo series (many of which are being used in this brochure) and video series
- Answers questions from adults in our "Ask Kate!" video series
- Holds a camp for young adults
- Distributes and updates CTF publications, such as this one, in English and various languages
- Provides a searchable feature on our NF Clinic listing to easily find existing healthcare providers that serve adults with NF1
- Creates events and programs for parents of children with NF
- Offers courses in becoming a Patient Advocate
- Promotes awareness of the NF1 Adult Care guidelines
- Supports health care professionals in staying current with the latest in clinical research and patient care at our yearly NF Conference
- Funds adult-focused NF research

Through social media, CTF posts the latest news and information, and allows individuals to ask questions, send comments, and connect with other members of the NF community. CTF is active across all social media channels at [@childrenstumor](https://www.childrenstumor.org).      

CTF offers regional and national events, such as Shine A Light NF Walks, NF Endurance events, NF Camp, clinic-hosted medical conferences, and social events in which individuals and families can find support and education, and connect with others living with NF1. These events are listed at ctf.org/calendar.

Visit ctf.org to download informational brochures in various languages, learn about CTF programs, or register for a virtual or local event.

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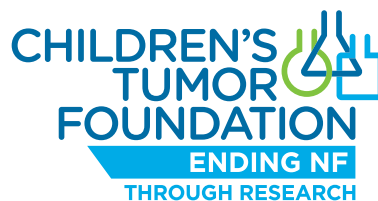
*Olivia, who lives with NF1.
She is a librarian and
practicing martial artist.*

Children's Tumor Foundation

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