

# DIAGNOSED WITH NF2-RELATED SCHWANNOMATOSIS

PREVIOUSLY REFERRED TO AS  
NEUROFIBROMATOSIS TYPE 2



## A MESSAGE FROM THE Children's Tumor Foundation

This booklet is designed to help you on your journey living with *NF2*-related schwannomatosis, also known as *NF2*-SWN (and previously referred to as neurofibromatosis type 2, or *NF2*). Whether this is a recent diagnosis or an established diagnosis, you will find information and support here to help you and your loved ones.

The Children's Tumor Foundation (CTF) created this guide to help answer some of the most common questions that people living with *NF2*-SWN have about coping with a diagnosis, understanding how this genetic condition occurs, recognizing common and less common symptoms, and managing care while living a full life.

We are also here to help you connect with other people diagnosed with *NF2*-SWN and their families and share opportunities for you to be part of the active and vibrant CTF community around the world.

On the Cover:  
*Sequoyah, who lives with NF2-SWN, is a photographer and photo editor from Brooklyn, NY. Pictured on the cover with her father Darien, who also lived with NF2-SWN.*



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*McKinnon, who lives with  
NF2-SWN*

# A DIAGNOSIS OF *NF2*-RELATED SCHWANNOMATOSIS

## Newly diagnosed with *NF2*-related schwannomatosis

At the Children's Tumor Foundation (CTF), we want you to know that you are not alone on your journey with *NF2*-related schwannomatosis, or *NF2*-SWN. For most people, many questions and concerns arise after receiving a diagnosis. You may need support as you learn about this condition, how to navigate it, and how to live your best possible life. This brochure is designed to give you essential information and resources.

There is a lot of information to absorb at one time, and you probably want to know how your diagnosis will impact your life. It can be helpful to remember that everyone deals with health-related news in different ways. While some prefer to receive small pieces of information at a time, others like to get as much information as possible right away. Either of these approaches is perfectly normal.

The Children's Tumor Foundation has many resources available for individuals diagnosed with any type of schwannomatosis. These resources include information on specialized clinics around the world, local events to help you meet others affected by *NF2*-SWN, and online opportunities.

## Previously diagnosed with *NF2*-related schwannomatosis

Coping with a diagnosis of a genetic condition can be difficult at any stage of life. Individuals and families must continually learn new things and adjust their coping strategies as new challenges come up. Considerations at the initial diagnosis may be very different from those that are applicable many years later. Considerations and challenges can change over time based on new symptoms and how you are feeling about living with *NF2*-SWN.

Because of new discoveries, in 2022 a group of experts published updated guidelines for the diagnosis of all types of schwannomatosis, including *NF2*-SWN. These changes may be hard to accept for some patients and may cause confusion or uncertainty. The Children's Tumor Foundation, along with your healthcare providers, is here to help you navigate your feelings and your understanding of *NF2*-SWN as we continue to learn more about these conditions.



*Laura, who lives  
with *NF2*-SWN*

# NF2-SWN: GET THE FACTS

## What is NF?

The term NF refers to a group of distinct genetic conditions, which include neurofibromatosis type 1, or NF1, and all types of schwannomatosis, including *NF2*-related schwannomatosis (previously known as NF2). They each have different genetic causes, so one type of NF cannot change into another type.

These are lifelong conditions that affect all populations equally, regardless of gender, race, or ethnicity. NF is not caused by anything you or your parent did or did not do, and NF is not contagious. People who have NF can lead productive lives, but they often require specialized medical care.

**did you know?**

One type of NF cannot change into another type.

## Introduction to Schwannomatosis

Schwannomatosis is a type of NF that causes tumors, called schwannomas, to grow on nerves in the central nervous system (brain and spine) and on peripheral nerves (the nerves throughout the rest of the body). Schwannomas are benign, meaning they are not cancer. However, sometimes they press on nerves, blood vessels, or other nearby organs, which can cause pain or other symptoms.

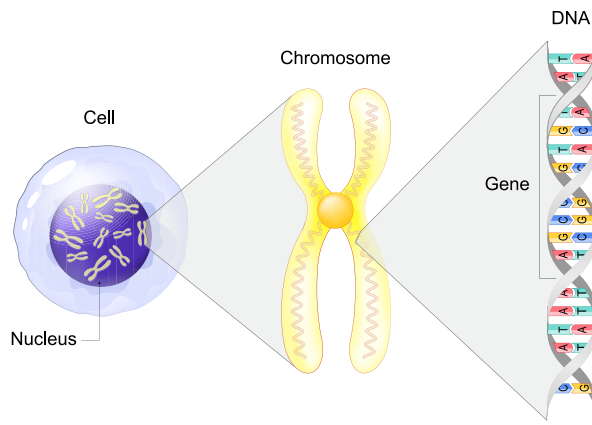
## Types of Schwannomatosis

The term schwannomatosis is an umbrella term for several genetic conditions that lead to a risk for multiple schwannomas to grow on nerves.

## did you know?

Chromosomes are made up of long strands of DNA, and a gene is a small part of DNA.

All of the known types of schwannomatosis are a result of a change in a gene (a sequence of DNA) located on chromosome 22. When a gene change causes the gene to stop working correctly, it is called a pathogenic variant (formerly called a genetic mutation) or disease-causing variant.



Before 2022, schwannomatosis and “NF2” were classified as two distinct conditions. However, researchers and doctors have learned that they are very similar, and it is more accurate to group them together under the umbrella term of schwannomatosis.

Recently, there has been a movement to change the way we name genetic conditions. This includes a combination of the name of the gene that is affected (has a pathogenic variant), and a main symptom or feature of the diagnosis. Therefore, we now refer to the types of schwannomatosis according to the gene affected, if it is known. *Gene names are always in italics.*

### ***GENE*-related schwannomatosis**

For example, *NF2*-related schwannomatosis is caused by a pathogenic variant in the *NF2* gene. Likewise, if the *SMARCB1* gene is involved, the condition is instead called *SMARCB1*-related schwannomatosis.

All of the known genes that cause schwannomatosis cause tumors called schwannomas. However, there are some differences in other health issues with each type of schwannomatosis, and researchers are continuing to learn more about these and how this impacts someone’s health.

The table below includes the names of the different types of schwannomatosis that are currently known. Researchers believe there are more genes to be discovered that cause schwannomatosis. As researchers continue to study these conditions, their findings will inform healthcare professionals more about the types of schwannomatosis and how to better take care of their patients.

Type of Schwannomatosis	Causative Gene	Estimated Incidence
<i>NF2</i> -related schwannomatosis	<i>NF2</i>	1 in 25,000 births
<i>SMARCB1</i> -related schwannomatosis	<i>SMARCB1</i>	1 in 280,000 births
<i>LZTR1</i> -related schwannomatosis	<i>LZTR1</i>	1 in 500,000 births
22q-related schwannomatosis	Unknown – genetic testing of blood/saliva is negative, and tumor testing localizes the disease-causing variant on chromosome 22q	Unknown
Schwannomatosis-NOS (not otherwise specified)	Unknown – genetic testing is not available or not performed	Unknown
Schwannomatosis-NEC (not elsewhere classified)	Unknown – genetic testing of blood/saliva and tumors is performed but does not identify any disease-causing variants	Unknown

**The information that follows in this brochure will refer only to *NF2*-related schwannomatosis.** For information about the other types of schwannomatosis, a separate brochure titled **Diagnosed with Schwannomatosis** is available to download or request at [ctf.org/education](http://ctf.org/education)



## Introduction to *NF2*-Related Schwannomatosis

*NF2*-related schwannomatosis (*NF2*-SWN) is the most common form of schwannomatosis and affects about 1 in 25,000 people. *NF2* carries the instructions for making a protein called merlin (which stands for Moesin-Ezrin-Radixin-Like protein). Merlin helps prevent cells from multiplying too many times. Without merlin, cells continue to multiply, becoming tumors.

The most common tumors in *NF2*-related schwannomatosis are called vestibular schwannomas (formerly called acoustic neuromas) on the nerve that carries sound and balance information from the inner ear to the brain (the eighth cranial nerve). In *NF2*-SWN, these tumors often affect both ears, and may lead to partial or complete hearing loss. *NF2*-SWN can also cause schwannomas to develop on other nerves in the central nervous system (brain and spine) or the peripheral nervous system (nerves in other places in the body, including on the skin).

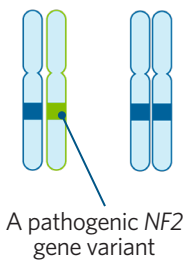
People with *NF2*-SWN may also develop other tumors called meningiomas (tumors of the membranes surrounding the brain and spinal cord) and ependymomas (tumors that develop from cells lining the ventricles of the brain and center of the spinal cord). This condition can also cause different problems with the eyes, some of which can cause vision loss. Most people develop symptoms in their late teen and early adult years, although about 10% of people develop symptoms during childhood. The most common first symptoms of a diagnosis of *NF2*-SWN are ringing in the ears (tinnitus), gradual hearing loss, and/or balance problems.

These symptoms are discussed in more detail on pages 11-13.

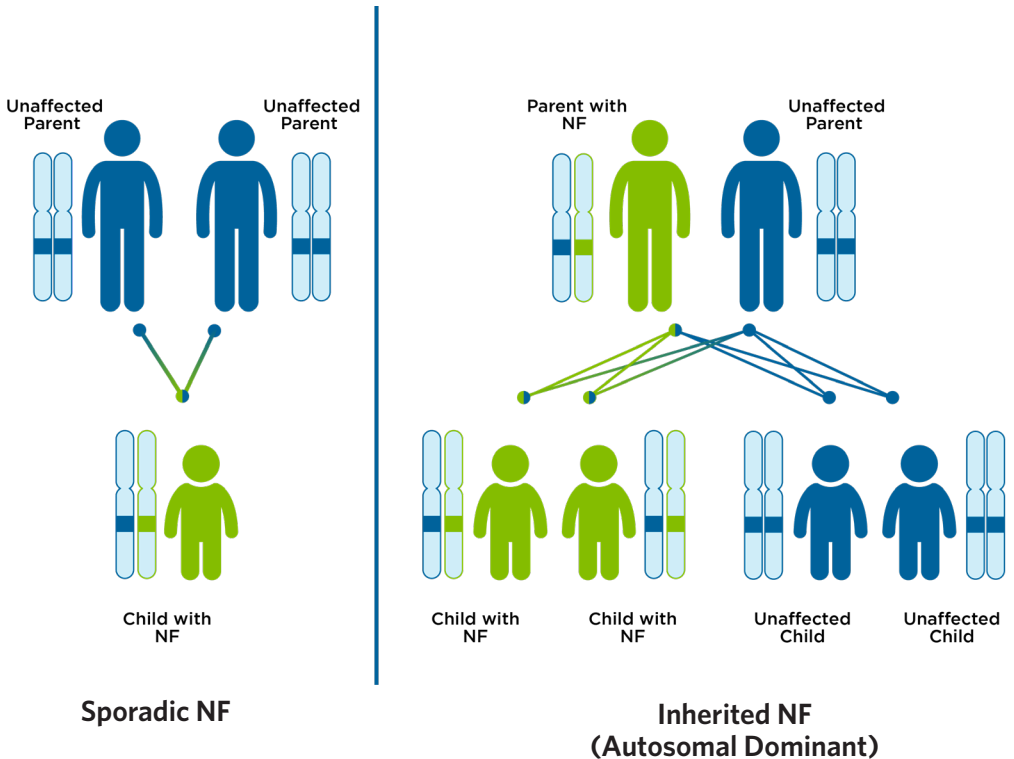
# What Causes *NF2*-Related Schwannomatosis?

## Familial *NF2*-SWN

Some cases of *NF2*-SWN are familial, or inherited, meaning more than one person in a family is affected by the condition. Approximately half of people born with *NF2*-SWN inherit it from an affected parent.



*NF2*-related schwannomatosis follows an autosomal dominant pattern of inheritance. We have two copies of most of the genes in our body, one from each parent, including the *NF2* gene. Autosomal dominant means that it only takes a pathogenic variant in one copy of the gene to cause the condition; the second copy of the gene from the other parent will not have a pathogenic variant and is working correctly. In general, a parent with an autosomal dominant disorder (such as *NF2*-SWN) has a 50% chance of passing along the condition to a child.



## Sporadic (non-familial) *NF2*-SWN

Approximately half of the people with *NF2*-SWN did not inherit the condition from a parent and are the first person in their family to be diagnosed. This results from a new change in the *NF2* gene, causing it to not work properly. We refer to this as sporadic or *de novo*. Because individuals with sporadic *NF2*-related schwannomatosis have a pathogenic variant in the gene, there is a risk of passing it along to their children, just like individuals with familial *NF2*-related schwannomatosis.

## Mosaic *NF2*-SWN

Over half of people with sporadic (but not familial) *NF2*-SWN have the pathogenic *NF2* variant in some parts of the body and not others. This is called genetic mosaicism, a situation in which an individual is composed of two genetically distinct cell types. People with mosaicism may have a milder form of the condition since the disease-causing variant is not present throughout the body; for example, mosaic *NF2*-SWN patients may develop hearing problems only in one ear. In individuals with mosaic *NF2*-SWN, the risk of passing on the condition to their child is usually less than 50%. Sometimes specialized genetic testing is needed in people with mosaic *NF2*-SWN to help better understand the risk to their children.

### did you know?

A parent with *NF2*-SWN usually has a 50% chance of passing on the condition to a child. If a parent has mosaic *NF2*-SWN, the chance can be less than 50%.



## Genetic Testing

Genetic testing for *NF2*-SWN is available but not always necessary to make a diagnosis of *NF2*-related schwannomatosis, as the diagnosis can often be made just on the basis of clinical features. *NF2* testing, along with testing for other genes associated with schwannomatosis, may be helpful when there is some question about the diagnosis. Additionally, *NF2* testing is sometimes helpful in understanding and predicting the severity of *NF2*-SWN and allowing for testing of family members who are at risk. It may also be helpful as part of family planning for future children. Testing may be done in a blood or saliva sample but can also sometimes require tumor tissue, if available.

### The Role of Genetic Counseling

Genetic counselors and medical geneticists have expertise in the study of genes and inherited disorders and how they impact families and their health. They are familiar with the complexity of these conditions. Genetic counselors can:

- Provide information about the inheritance patterns involved in the different types of schwannomatosis
- Explain the testing options available and discuss the benefits, risks, and limitations of genetic testing
- Help you decide the best genetic test options for your situation
- Interpret and explain the results of genetic testing
- Discuss potential uses of genetic testing for family planning (including prenatal testing during pregnancy and embryo genetic testing during *in vitro* fertilization), or testing of other family members
- Help families cope with a genetic diagnosis and its impact on the family
- Find resources that may assist families throughout the journey of living with *NF2*-SWN

# CLINICAL FEATURES

## Symptoms of *NF2*-Related Schwannomatosis (Formerly known as *NF2*)

There are different ways that *NF2*-related schwannomatosis (*NF2*-SWN) can initially be suspected.

First, there may be a concern with hearing or balance. For others, findings on an eye exam (cataracts or epiretinal membrane) can lead to a diagnosis. *NF2*-SWN is sometimes diagnosed before any symptoms develop, either because it was suspected due to family history or if *NF2*-SWN tumors were found incidentally (unexpectedly) on a body scan obtained for another reason (for example, a brain scan is done after a head injury, and reveals a vestibular schwannoma tumor).

*NF2*-SWN is highly variable, and not every individual with this condition will experience all manifestations. Some of the difficulties and symptoms that individuals with *NF2*-SWN might experience are:

- Ringing in the ears (tinnitus)
- Hearing loss
- Problems with balance
- Facial weakness
- Speaking and swallowing difficulties
- Seizures
- Vision loss
- Loss of balance and mobility due to tumors of the spine, foot drop, pain, muscle wasting, or other nerve damage.



## Schwannomas

The most common tumors seen in people with *NF2-SWN* are schwannomas. Schwannomas are noncancerous (benign) tumors made up of abnormal Schwann cells, which are the cells that insulate the nerves. They can develop on any nerve in the body, including the cranial nerves, spinal nerves, and peripheral nerves.

### Vestibular Schwannomas

The most common schwannoma seen in *NF2-SWN* are vestibular schwannomas, sometimes also called acoustic neuromas. These schwannomas often affect the acoustic (cochlear) nerves (which carry information about sound to the brain) and the vestibular nerves (which carry balance and equilibrium information to the brain). Vestibular schwannomas can be unilateral (on one side), or bilateral (on both sides). An MRI scan (picture of the different parts of the brain) is routinely done to monitor the presence or progression of vestibular schwannomas.

In *NF2-SWN*, vestibular schwannomas are usually bilateral and therefore affect hearing on both sides. There are different approaches to the treatment and management of vestibular schwannomas, and individuals might consider multiple opinions reviewing these options.

### Non-vestibular Schwannomas

The most common places for non-vestibular schwannomas to develop in *NF2-SWN* are the spinal nerves (spinal schwannoma), other cranial nerves (nerves of the head and face), and peripheral nerves (peripheral schwannoma) in the rest of the body. These are often monitored by MRI scans and monitoring for symptoms (such as weakness, pain, and numbness). The primary treatment for non-vestibular schwannomas is surgery.

## Meningiomas

These are benign tumors found on the lining of the brain and spinal cord. They occur in about 80% of people with *NF2-SWN* at some point in their lifetime but don't always cause noticeable symptoms. Symptoms of meningiomas can include headaches, seizures, blurred vision, weakness, or numbness. Many meningiomas in people with *NF2-SWN* never require treatment, but they are often monitored by MRI scans. Surgery is the primary treatment for meningiomas.

## Ependymomas

These are benign tumors that typically develop from the cells that line the spaces inside the spinal cord. Ependymomas will occur inside the spine in about 20% of people with *NF2-SWN*. Spinal ependymomas may cause pain, sensory changes, or weakness but are most commonly asymptomatic. If needed, surgery is the primary treatment for ependymoma, although other treatments, such as radiation therapy or chemotherapy, may occasionally be considered.

## Eye/Vision Issues

Visual function is impacted in some way in about one-third of people with *NF2-SWN*. Juvenile cataracts are common in young people diagnosed with *NF2-SWN* and may or may not cause vision problems. In some cases, people with *NF2-SWN* may encounter eye problems due to schwannomas on the cranial nerves. This can impact vision directly, can cause problems with eye movements (sometimes leading to double vision), can affect the opening and closing of the eyelids due to facial weakness, and can cause clouding of the eye due to eyelid closing problems and loss of sensation in the eye. Additionally, people with *NF2-SWN* can develop a membrane on the back of the eye (epiretinal membrane), or have tumors that grow behind the eyes that can result in vision loss.

# MEDICAL MANAGEMENT

## Treatment of *NF2*-Related Schwannomatosis

There is currently no cure for *NF2*-related schwannomatosis (*NF2*-SWN). However, promising research and clinical trials are underway evaluating the biology of *NF2*-SWN tumors and testing drugs. There is hope that these potential treatments may be able to effectively shrink *NF2*-SWN tumors and manage or prevent symptoms.

Because *NF2*-SWN involves many different parts of the body, healthcare providers from different specialties may be involved in caring for people with *NF2*-SWN. An NF clinic is the facility most likely to offer a full range of necessary services. You can find a list of NF specialty clinics on the CTF website at [ctf.org/doctor](https://www.ctf.org/doctor). Look particularly for facilities that specialize in *NF2*-SWN, which means that they are familiar with the complexities of *NF2*-SWN and the treatment and management of individuals living with the condition. Ideally, the various specialists required for *NF2*-SWN care can coordinate and communicate with each other for management and treatment planning (see page 21).

In the course of diagnosis, treatment, and ongoing care, a person with *NF2*-SWN might seek care from a number of specialists:

- Neurology – conditions affecting the brain and spine
- Otolaryngology (ENT) – surgeons for ear, nose, throat, and vestibular (inner-ear balance) systems
- Neurotology – surgery for ear problems related to the nerves
- Audiology – evaluations for hearing, hearing aids, cochlear and auditory brainstem implants
- Neurosurgery – surgery for the brain and spinal cord
- Oncology and neuro-oncology – management and treatment for benign and malignant tumors
- Ophthalmology – monitoring and treatment of eyes and vision
- Orthopedics – surgery to remove schwannoma tumors
- Neuro-ophthalmology – monitoring and treatment of visual problems related to nerves
- Genetics – diagnosis and family risk assessment
- Pediatric and adult primary care – routine medical care
- Physical, occupational, or speech therapy – improve function and treat pain and disability



## Coping with Hearing Loss

Communication is a meaningful exchange of information between two or more individuals. In *NF2-SWN*, when there's hearing loss or speech difficulties, it can profoundly impact your relationships and social interactions.

A number of tools and strategies can be utilized to maximize communication options in the workplace, with your friends and family, and in everyday life. This can include learning visual means of communication that are not dependent on hearing such as American Sign Language (ASL) and Cued Speech. Assistive technologies, such as voice-to-text apps as well as telephone and in-person transcription services, can be very helpful. For some individuals, hearing aids can improve their understanding

of spoken language and other sounds. In others, implanted hearing options such as auditory brain implant (ABI) and cochlear implant (CI) may be available.

Strategies and tools to help you cope with hearing loss can be found at [ctf.org/hearingloss](https://ctf.org/hearingloss)



*I've been there, and I know what it's like to slowly lose your hearing and your face. And then you have to try to learn about all the different things that are available to help regain your hearing. . . . The good news in all of this is that there has never in the history of man been a better time to be deaf. Between the ABI (Auditory Brain Implant) texting and some of the transcription apps - those tools are out there. And they really do help; every little bit helps to normalize your day.*



—Matt, who lives with *NF2*-related schwannomatosis

## Facial Nerve Injury

Individuals living with *NF2*-SWN often develop injury of the facial nerve due to the growth of their vestibular or facial nerve schwannomas or, more commonly, during the treatment of their tumors. Damage to the facial nerve can result in facial palsy (weak forehead, eyelids, or mouth), slurring of speech, change in taste, eating, and incomplete eye closure. Patients undergoing these issues should seek facial physical therapy (a subspecialty of physical therapy) as a first treatment option. Botox injections or surgery might be recommended.



“

*NF shouldn't stop us from pursuing our dreams. Nothing stops me from trying. And others should know that just because we have a disease that is challenging, we can succeed in life.*”

”

—Christine, who lives with  
*NF2*-related schwannomatosis

# HOW YOUR DOCTOR MAKES A DIAGNOSIS OF *NF2*-RELATED SCHWANNOMATOSIS

Based on a detailed medical history and examination, healthcare providers use a list of diagnostic criteria (a checklist of signs and symptoms) to determine whether or not an individual may be given a diagnosis of *NF2*-related schwannomatosis (*NF2*-SWN). Additionally, pathologic examination of a removed tumor and/or genetic testing of blood, saliva, or tumor may contribute to making a diagnosis of *NF2*-SWN.

## 2022 Diagnostic Criteria Update

The diagnostic criteria for schwannomatosis and the condition previously called “neurofibromatosis type 2” were carefully reviewed by a large group of NF experts. They published an update to these guidelines in 2022, in which neurofibromatosis type 2 was renamed *NF2*-related schwannomatosis. The purpose of the revisions is to improve diagnostic accuracy so that clinicians can make the best decisions to help improve the health and well-being of patients.

While the diagnostic criteria below were developed specifically for healthcare providers, it may be helpful for you to be aware of them to enhance your understanding of the diagnostic process of *NF2*-SWN and discuss your own individual diagnosis with your healthcare providers.

### For Healthcare Providers and General Practitioners

Complete information about the diagnostic criteria for all types of NF, including links to the *Genetics in Medicine* publication, summary documents, and an NF Diagnosis mobile app, may be found at [ctf.org/criteria](https://ctf.org/criteria)

# Diagnostic Criteria for NF2-Related Schwannomatosis

Formerly called neurofibromatosis type 2

A diagnosis of NF2-related schwannomatosis can be made when a patient has **one of the following**:

1. **Bilateral vestibular schwannomas** (tumors on the hearing nerves of both ears)
2. **An identical NF2 pathogenic variant\* in at least two separate NF2-related tumors** (including schwannoma, meningioma, and/or ependymoma)
3. **Either two Major OR one Major and two Minor criteria are present as follows:**

Major criteria	Minor criteria
<ul style="list-style-type: none"> <li>▪ Unilateral vestibular schwannoma (tumor on only one side)</li> <li>▪ A parent with NF2-related schwannomatosis</li> <li>▪ Two or more meningioma tumors (Note: a single meningioma qualifies as a minor criteria)</li> <li>▪ NF2 pathogenic variant* is found when blood or saliva is tested</li> </ul>	<p><b>Can count more than one of a type</b> (e.g., two schwannomas = two minor criteria)</p> <ul style="list-style-type: none"> <li>▪ Ependymoma tumor OR schwannoma tumor</li> </ul> <p>(Note: if the major criteria is vestibular schwannoma (on the hearing nerve), at least one schwannoma must be located in the skin)</p>
	<p><b>Can count only once</b></p> <ul style="list-style-type: none"> <li>▪ Juvenile cataracts of the eye (either subcapsular or cortical in type), OR</li> <li>▪ Retinal hamartoma (tumor on the nerve of the retina), OR</li> <li>▪ Epiretinal membrane (a thickening of the tissue in the eye's retina) in a person less than 40 years old, OR</li> <li>▪ One meningioma tumor</li> </ul> <p>(Note: multiple meningiomas qualify as a major criterion; meningioma cannot be used as both a major and a minor criteria)</p>

*\*When the variant is present at reduced level, the diagnosis is mosaic NF2-related schwannomatosis*

## MOSAICISM

Mosaicism is confirmed for NF2-related schwannomatosis by **either** of the following:

- Clearly less than 50% pathogenic variant allele fraction in blood or saliva OR
- Pathogenic variant is not detected in blood or saliva, but a shared pathogenic variant is detected in two or more independent (anatomically unrelated) tumors

**SUMMARY:** Mosaicism is confirmed when blood tests show that only some of the cells being tested show the same gene variant or when blood or saliva shows no pathogenic variant. A variant must be found in two separate tumors.

# DISCUSSING YOUR DIAGNOSIS

## Telling Others

A difficult aspect of dealing with a new diagnosis is deciding how and when to tell family and close friends. Here are some suggestions that may help make the process a little easier.

### Who should I tell?

Who you tell is up to you. You do not have to tell anyone about your diagnosis if you do not want to. However, sharing your diagnosis can be important in some relationships to explain what you have been experiencing. It can also be helpful to receive support from other people.

### What should I tell them?

When you decide to share your or your family member's diagnosis with others, you must also decide how much information to share. You might only feel like sharing in a limited way, such as discussing the fact that hearing loss is usually the main symptom. Other times it is helpful to have someone with whom you can share more details, including all of the potential ups and downs that go along with the diagnosis.

## Sample Message to Family & Friends

Dear \_\_\_\_\_,

I want to share with you that \_\_\_\_\_ has been diagnosed with NF2-related schwannomatosis, also called NF2-SWN.

NF2-SWN is a genetic condition that causes benign (noncancerous) tumors to grow on the nerves of the brain and spine. \_\_\_\_\_ will likely require special medical care and lifelong monitoring.

The most obvious signs of NF2-SWN may be hearing loss, facial weakness, or balance issues. NF2-SWN is something that people are born with, although it may not be diagnosed until later in life. It is caused by a genetic change that occurs in about 1 in 25,000 people. It is not contagious. There is not yet a cure. However, scientists and doctors are working toward understanding and treating NF2-SWN

If you would like to learn more about NF2-SWN, the Children's Tumor Foundation has free online information about the condition at [ctf.org](http://ctf.org).

We appreciate all of your love and support.

Sincerely, Your Name

# ADDITIONAL SUPPORT AND RESOURCES

## Connecting With Other Patients and Families


After your diagnosis, you might experience a range of feelings that may include shock, sadness, anger, and uncertainty. While all of these feelings are completely normal, it can be difficult to deal with these emotions by yourself. In addition to getting love and support from friends and family, it can also be helpful to connect with other patients and families who are facing similar challenges. These individuals might have a special understanding of your thoughts and feelings and offer a uniquely personal perspective about their own experiences and challenges with *NF2-SWN*.

It may be comforting and useful for you to read stories about others living with *NF2-SWN* and their families. The CTF newsfeed at [ctf.org/news](https://ctf.org/news) frequently posts stories of NF. You may also enjoy reading the stories from NF Heroes of all ages at [ctf.org/storiesofnf](https://ctf.org/storiesofnf)

Numerous videos featuring *NF2-SWN* patients of all ages can be viewed on the Children's Tumor Foundation and Make NF Visible YouTube channels.

[YouTube.com/ChildrensTumor](https://www.youtube.com/ChildrensTumor)

[YouTube.com/MakeNFVisible](https://www.youtube.com/MakeNFVisible)



**“** Making NF visible cultivates community, connection, and deep friendships with others just like you. It creates so many opportunities for support, encouragement, guidance, and an improved life. This kind of advocacy helps provide a guide on this hard journey and is a constant reminder that you can do this and you are not alone. By ourselves, we can only go so far, but together we can go even further. **”**

—Nissa, who lives with *NF2*-related schwannomatosis

## Finding Specialized Care

Because *NF2-SWN* is a rare genetic condition, finding a specialist with experience in diagnosing and managing the condition can be challenging. If you have a confirmed or suspected diagnosis of *NF2-SWN* or any type of schwannomatosis, it's important to look for a trusted, experienced clinician.

The Children's Tumor Foundation (CTF) knows how important it is to find healthcare professionals who have experience diagnosing and caring for patients with *NF2-SWN*. CTF has established a nationwide network of NF clinics called the NF Clinic Network (NFCN) that recognizes clinics that provide comprehensive medical care.

Clinics specializing in all types of NF provide a comprehensive, multidisciplinary approach to care. These specialized clinics are led by experts who collaborate in the treatment and management of all forms of schwannomatosis and related complications. Genetic counselors, nurses, and other caring specialists affiliated with these clinics can provide patients and families with assistance and support in managing the lifelong medical, psychological, and social implications of an *NF2-SWN* diagnosis.

To find a clinic that is a NFCN member and other specialists throughout the country who are familiar with NF, go to our Find a Doctor page at [ctf.org/doctor](https://ctf.org/doctor)

### For your Healthcare Providers

We understand that everyone may not have access to an NF specialist, so CTF has developed the NF Diagnosis mobile app for primary care providers, available for iPhone and Android phones. Within the app is up-to-date NF diagnostic information specifically for clinicians. There are also links to important publications about *NF2-SWN* for the general practitioner. Ask your doctor to go to [ctf.org/nfapp](https://ctf.org/nfapp) for more information.

## The NF Registry

The NF Registry is a patient-entered resource for sharing information about your symptoms to help guide schwannomatosis research. This safe and effective tool will empower patients and their caregivers by inviting them to take an active role in advancing NF research.

When you join the NF Registry, you have access to the latest discoveries about the many ways living with NF can affect individuals and families. This information will help you and your family find the best possible care. As an NF Registry participant, you complete a yearly health survey. This data helps researchers study how NF affects everyone differently and how NF changes over time. You can then choose whether to receive personalized emails about any or all of the following topics:

- Clinical trials and research studies relevant to you or your child
- Updates to NF care recommendations
- Research announcements and news
- Surveys designed to get patient input on key NF challenges
- Educational materials specific to you
- Resources to help you on your path with NF

The NF Registry's first principle is that people living with *NF2*-related schwannomatosis (*NF2*-SWN) are always in control of their own information. You only share what you want to share, and you control the permissions on when or if you are to be contacted. All information is carefully protected with the strictest privacy protocols in place. Even if you choose not to be contacted, your participation helps researchers learn from NF patients and families. To learn more or join the NF Registry, go to **[nfregistry.org](https://nfregistry.org)**

### did you know?

Joining the NF Registry will give you access to the latest *NF2*-SWN research, and can alert you to clinical trials and research studies relevant to you.



## Research

The Children's Tumor Foundation is passionately working toward breakthroughs in treating *NF2*-related schwannomatosis.

Our commitment to progress shines through in the *NF2*-SWN research field. We are actively seeking therapies through powerful, ongoing initiatives. Over the years, numerous individual researchers have been supported by CTF funding. Notably, our "Synodos for *NF2*" team science initiative brought together experts from 12 world-class labs over three years. These dedicated teams collaborated, sharing information, datasets, and results in real-time, and have generously made that data freely available.

The Synodos project discovered several promising treatments and played a crucial role in the creation of the INTUITT-*NF2* platform trial. This collaborative effort between academic institutions and pharmaceutical companies aims to expedite the availability of promising treatment options for *NF2*-SWN patients.

CTF remains committed to accelerating the time it takes to bring effective *NF2*-SWN treatments to the clinic and, ultimately, to patients. We achieve this by expanding the clinical drug pipeline for *NF2*-SWN, enhancing drug selection through innovative testing models, and exploring gene therapy options that target the root genetic causes of *NF2*-SWN.

Explore more about our impactful work that continues to make life-saving progress for patients living with *NF2*-SWN at [ctf.org/impact](https://ctf.org/impact)

**Searching for information is a positive step that can empower you as a patient. Keep in mind that your doctor is the best resource for information or to answer questions. If you come across any information that you find confusing or strange, it's important that you talk to your doctor.**

# THE CHILDREN'S TUMOR FOUNDATION

Founded in 1978, the Children's Tumor Foundation (CTF) began as the first grassroots organization solely dedicated to the goal of finding treatments for NF, which includes neurofibromatosis type 1, or NF1, and schwannomatosis, including NF2-related schwannomatosis, or NF2-SWN. Today, CTF is a highly recognized global nonprofit foundation, the leading force in the fight to end NF, and a model for other innovative research endeavors.

## **The Mission of the Children's Tumor Foundation:**

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

**Our Vision:** End NF.

“ *It is important to make NF visible so that all people with the condition, whether it is obvious or not, get a voice.* ”

—Alexandra, who lives with NF2-SWN





## CTF Educational Resources

There is a lot to learn about when you or someone you love receives a diagnosis of *NF2-SWN*. The unpredictability of the condition requires keeping up with new and innovative research and clinical information. This can feel overwhelming at times. However, there are resources to help make your journey feel a little more manageable.

### Get the Facts

Read the information in this booklet, and visit the Foundation's website at [ctf.org](https://ctf.org) to learn more about the different types of schwannomatosis, or attend a local clinic symposium or a national event like the CTF NF Summit, which is held each year.

### Translations

The Children's Tumor Foundation is a global organization and is working to translate our educational resources, including this one, into Spanish and additional languages. To learn more, go to [ctf.org/education](https://ctf.org/education)

### Download the NF Care Patient App

For patients and caregivers, the Children's Tumor Foundation NF Care App contains quick access to the CTF newsfeed, research updates, patient resources, and more. Learn more at [ctf.org/nfapp](https://ctf.org/nfapp)

### Sign Up for Research

Join the NF Registry at [nfregistry.org](https://nfregistry.org) to learn about and participate in advanced scientific research for NF. Read more on page 22 of this brochure.

## Get Involved

Getting involved is a great way to meet others who are also affected by *NF2-SWN*.

### CTF National Programs

The Children's Tumor Foundation's fundraising opportunities include Shine a Light NF Walk, NF Endurance, and Cupid's Undie Run. These are great ways to empower yourself and your community in the fight for treatments and a cure for all types of NF.

### The NF Summit

An annual gathering of patients, families, volunteers, and clinicians, the NF Summit allows those living with NF and their families to connect, support, and learn from one another while attending seminars on relevant topics pertaining to *NF2*-related schwannomatosis. Find out more at [ctf.org/nfsummit](https://ctf.org/nfsummit)

### Spread the Word

Families and organizations from around the world participate in NF Awareness Month each May and join the Children's Tumor Foundation's Make NF Visible and Shine a Light awareness campaigns. You can share our infographics and videos on social media, secure a proclamation in your local town or state, light up a local landmark, and even advocate for NF research funding. Learn more at [ctf.org/nfawareness](https://ctf.org/nfawareness)

### Become an *NF2-SWN* Accelerator

An *NF2-SWN* Accelerator is a community advocate who uses their voice to raise awareness and share information about CTF's commitment to expand the drug pipeline and accelerate treatments for *NF2-SWN*. If you are interested in becoming an *NF2-SWN* Accelerator, email "I want to be an *NF2-SWN* Accelerator" to [info@ctf.org](mailto:info@ctf.org)

### CTF Europe

Children's Tumor Foundation Europe, which launched in 2018, has been hard at work driving research, expanding knowledge, and advancing care for the over 250,000 Europeans living with NF. Find out more at [ctfeurope.org](https://ctfeurope.org)

## Stay Informed

Visit the CTF website at [ctf.org](https://ctf.org) to stay informed about NF research, find a calendar of events, or sign up for our newsletter or email list.

## Get Social

In addition to in-person events around the country, you can connect with NF patients and families on any of the Children's Tumor Foundation social media channels.

**Facebook:** [facebook.com/childrenstumor](https://facebook.com/childrenstumor)

**X:** [twitter.com/childrenstumor](https://twitter.com/childrenstumor)

**Instagram:** [instagram.com/childrenstumor](https://instagram.com/childrenstumor)

**YouTube:** [youtube.com/childrenstumor](https://youtube.com/childrenstumor)

**LinkedIn:** [linkedin.com/company/children's-tumor-foundation](https://linkedin.com/company/children's-tumor-foundation)

**TikTok:** [tiktok.com/childrenstumor](https://tiktok.com/childrenstumor)

## Form Connections

Reach out to CTF to see if there is a contact or event in your area. We have many volunteers and staff members across the country who are available and want to help support you. To speak to someone directly, email us at [info@ctf.org](mailto:info@ctf.org) or call **1-800-323-7938**.

## Remember that you are not your diagnosis.

Yes, it is a major part of your journey. Yes, it is very difficult. However, remember that you are a unique individual with talents, skills, personal strengths, and resources—even if there are many challenges to living with *NF2-SWN*. The world can be a better place because of you in so many ways.

## References

Plotkin, S. R., Messiaen, L., Legius, E., et al. (2022). Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation. *Genetics in Medicine*, 24(9), 1967-1977. doi: <https://doi.org/10.1016/j.gim.2022.05.007>

Evans DGR, et al. (2017). Cancer and CNS Tumor Surveillance in Pediatric Neurofibromatosis 2 and Related Disorders. *Clin Cancer Res*, 23(12), e54-e61. doi: 10.1158/1078-0432.CCR-17-0590. PMID: 28620005.

Ruggieri, M., Praticò, A. D., & Evans, D. G. (2015). Diagnosis, Management, and Therapeutic Options in Childhood Neurofibromatosis Type 2 and Related Forms. *Semin Pediatr Neurol*, 22(4), 240-258. <https://doi.org/10.1016/j.spen.2015.10.008>


Plotkin SR, Blakeley JO, Evans DG, et al. (2013). Update from the 2011 International Schwannomatosis Workshop: From genetics to diagnostic criteria. *Am J Med Genet A*, 161A(3), 405-416. doi: 10.1002/ajmg.a.35760. PMID: 23401320; PMCID: PMC4020435.

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This patient brochure was prepared by the Children's Tumor Foundation and approved by the 2023 CTF Clinical Care Advisory Board.

*The photographs of individuals living with NF2-SWN throughout this brochure were captured by photographer and NF Dad Craig Warga.*



We encourage families to use only reputable websites and not perform general online searches, which may be inaccurate or show worst-case scenarios. In addition, we advise against seeking personal medical advice from social media platforms. Please talk to your healthcare provider about specific questions regarding your or your family member's health or complications of *NF2*-related schwannomatosis.



**BORN**  
**A FIGHTER.**

*Leanna, who  
lives with  
NF2-SWN*

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Foundation  
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**CHILDREN'S  
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THROUGH RESEARCH

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