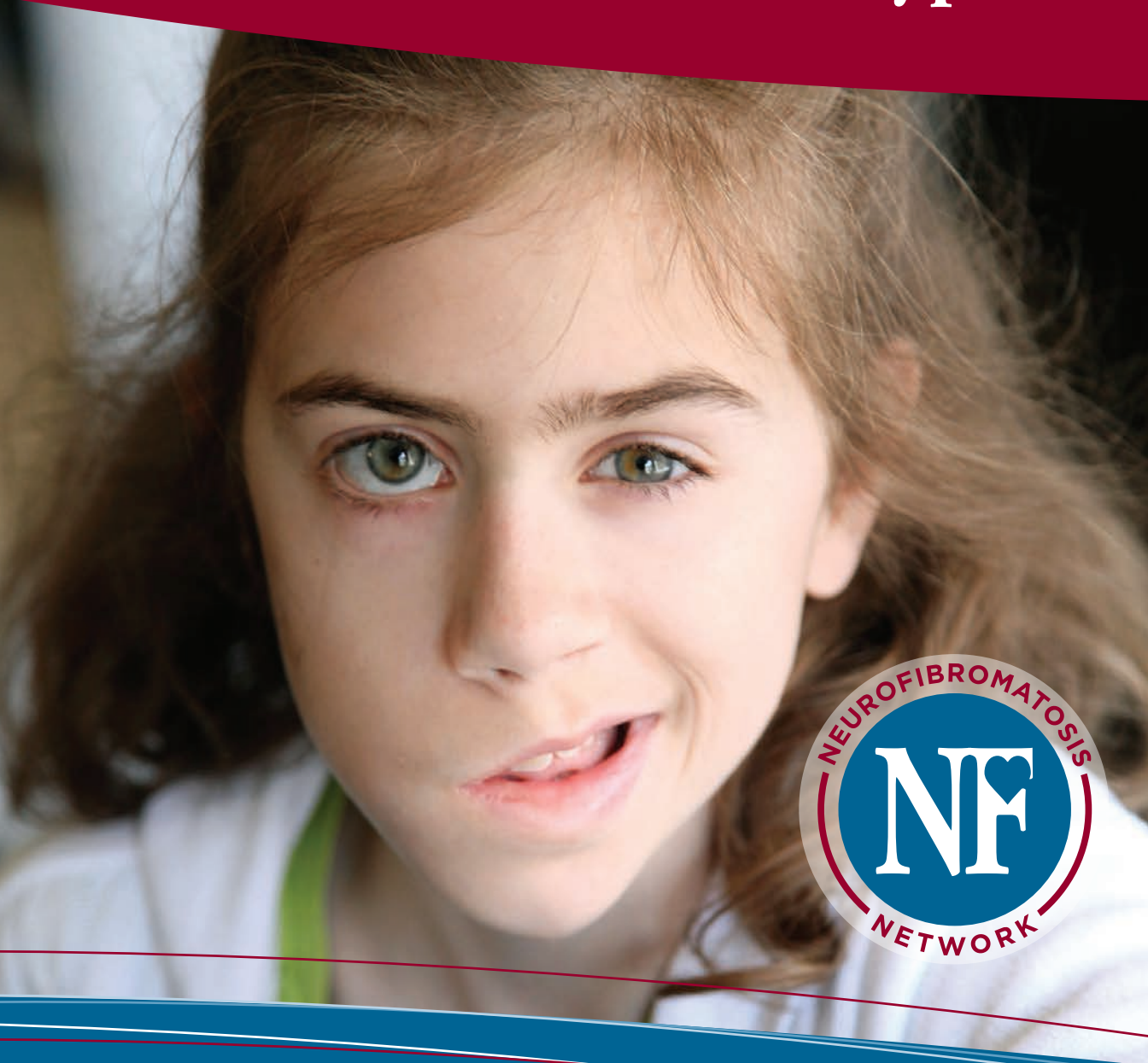


Understanding Neurofibromatosis Type 2



AN INTRODUCTION FOR PATIENTS AND PARENTS

John and Linda Manth
Scott Plotkin, MD, Ph.D.

**To Leah, the Sexton Family and
everyone affected by NF2.**

*“We cannot change the cards we are dealt,
just how we play our hand.”*

(The Last Lecture, by Randy Pausch)

Our hope someday is that there will be no need for
this booklet.

**To the researchers and doctors who have dedicated
their professions to finding treatments for NF2:
We did not choose NF2, but we are forever grateful
you have.**



Project Leader: Kim Bischoff
Layout and editing: Kelly Walsh-Curtis

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AN INTRODUCTION FOR PATIENTS AND PARENTS

Introduction

The purpose of this book is to assist NF2 patients and their family members to gain information and to know that you are not alone. As you begin your NF2 journey you may have feelings similar to others before you. You are overwhelmed, wishing it would all go away, and most of all scared. Suddenly all the hopes and dreams you have for yourself/family member may feel like they are being taken away. NF2 research continues to advance and scientists are beginning to piece together the NF2 puzzle. We now have hope that surgery will not be the only treatment for NF2 tumors, and that research may lead to salvaging hearing, the life-altering sense typically robbed by those affected with NF2. Hopefully, you will be glad you read this booklet, more informed at the end, but most of all feeling optimistic that you are not alone and that together we can make a difference and improve the lives of those affected by NF2.

What are the other forms of Neurofibromatosis?

Neurofibromatosis 1 (NF1): also known as von Recklinghausen NF or Peripheral NF. NF1 occurs in 1:3,000 births, is characterized by multiple cafe-au-lait spots and neurofibromas on or under the skin. Deformation of bones and curvature of the spine (scoliosis) may also occur. Occasionally, tumors may develop in the brain, on cranial nerves, or on the spinal cord. About 50% of people with NF also have learning disabilities.

Schwannomatosis: a rare form of NF that has only recently been recognized and appears to affect around 1:40,000 individuals. It is less well understood than NF1 and NF2, and features may vary greatly between patients.

*...together we can make a difference
and improve the lives of those
affected by NF2...*

What is NF2?

Neurofibromatosis type 2 is a genetic condition that causes a predisposition to develop bilateral (affecting both sides) vestibular schwannomas (tumors on the 8th cranial nerve, which affect hearing and balance) and other tumors on any nerves in the body. Patients are born with NF2; it is not acquired during life.

How is NF2 different from unilateral vestibular schwannoma (acoustic neuromas)?

Unilateral (one-sided) vestibular schwannomas are common in the general population and account for about 7-9% of all brain tumors. The average age at presentation for a unilateral vestibular schwannomas is around 55 years. In contrast, bilateral vestibular schwannomas are characteristic of NF2. NF2 patients typically present in the late teens or early 20s with symptoms related to vestibular schwannomas. Some patients with NF2 can have unilateral vestibular schwannomas plus other features of NF2 such as meningiomas, non-vestibular schwannomas or cataracts.

How common is NF2?

Neurofibromatosis 2 is an uncommon disorder thought to occur in 1 in 25,000 live births. Although it is a genetic disorder, many patients with NF2 lack a family history. This occurs in about half of patients who are

the first person in their family to have NF2.

What are the presenting symptoms?

The typical presenting symptoms for adult NF2 patients include hearing loss, tinnitus (“ringing” in the ear), and balance problems. In pediatric NF2 patients, presenting symptoms may also include skin tumors, visual problems, spinal cord compression, and seizures.

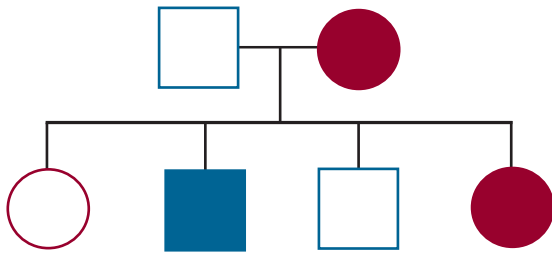
How is a diagnosis of NF2 confirmed?

A diagnosis of NF2 is confirmed by clinical and radiologic evaluation. There are multiple criteria that physicians use including the National Institutes of Health (NIH) criteria and the Manchester criteria. Your physician can provide details about the individual criteria. Essentially, three groups of patients meet these criteria. The first group of patients includes those with bilateral vestibular schwannomas on MRI scans. The second group of patients includes those with a family history of NF2 in a first degree relative (eg., a parent or brother/sister) and a personal history of vestibular schwannoma (either unilateral or bilateral). The third group of patients includes those with no family history of NF2 but with a unilateral vestibular schwannoma and at least 2 other clinical signs of NF2 that may include meningioma, neurofibroma, ependymoma, or juvenile cataracts.

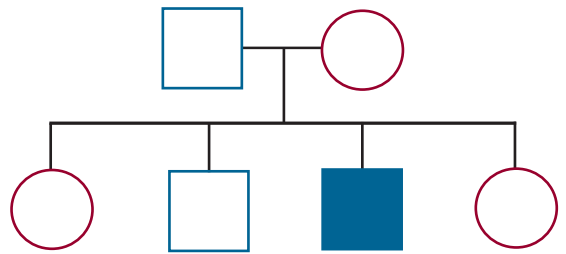
Why do I have NF2?

NF2 presents in two different ways. In about half of the patients it is inherited from a parent, the term familial is used in these cases. The other half of patients have no family history and the term sporadic is used. A genetic mutation occurs shortly after fertilization, resulting in the NF2 gene alteration. Some sporadic cases present as mosaic NF2, a genetic term that means not all of the cells in their body have the NF2 gene alteration. Patients with mosaic NF2 may have milder symptoms compared to others.

Key: squares = men; circles = women; filled in = affected with NF2; unfilled = unaffected



Typical family tree for someone who inherits NF2, each generation may be affected by familial NF2.



Typical family tree for someone who is the first in their family with sporadic NF2.

If I have NF2, what about my children?

The transmission pattern for NF2 is autosomal dominant. Meaning, on average, each pregnancy carries a 50% risk of passing the NF2 gene onto your children, whether the father or mother has NF2. The one exception to this may be mosaic patients, whose risk may be less than 50%. For all those affected by NF2, a genetic specialist can help estimate the risks based on your personal medical history.



NF2 is passed by autosomal dominance in about half the cases. If one parent has NF2, just like tossing a coin, each pregnancy carries a 50% risk of NF2 passing onto the child.



NF2 occurs *sporadically* in about half the cases, there is no family history, they are the first in the family to be diagnosed.

What tests are important for a patient with a new diagnosis of NF2?

Patients with a new diagnosis of NF2 should undergo an extent-of-disease evaluation. The goal of this evaluation is to understand what manifestations of NF2 an individual patient has. The evaluation may include an MRI scan of the brain with contrast and with fine cuts (3 mm slices) through the internal auditory canal, an MRI scan of the spine, a hearing test (including measurement of pure tones thresholds and word recognition score), and an ophthalmologic evaluation. In some individuals, evaluation of swallowing or voice quality is indicated. In some patients (those considering starting a family), genetic counseling is also advisable.

What's the deal with vestibular schwannomas?

Vestibular schwannomas are the hallmark tumor of NF2. These tumors are associated with hearing loss that can occur suddenly or gradually over time.

Sudden hearing loss is defined as a decrease in hearing that occurs in less than 72 hours. This type of hearing loss is usually treated with steroid medications taken by mouth and usually recovers with treatment. When patients experience sudden hearing loss, they should contact their medical team immediately for treatment.

Chronic hearing loss is defined as a decline in hearing over time (months to years). Surprisingly, there is a poor correlation between tumor size and hearing loss in patients with NF2. In practice, patients with large tumors may have good hearing and patients with small tumors may be deaf. Surgery is the mainstay of treatment for vestibular schwannomas, but medications are currently being studied as a treatment option. Surgeries that include cochlear nerve implants and auditory brain stem implants maybe an option for hearing assistance.

Potential complications of surgery include: Complete hearing loss, facial weakness, hoarseness, difficulty swallowing, and headache. It is important for NF2 patients to consult with experienced surgeons when they are considering surgery for their vestibular schwannomas. Studies have shown that medical centers that perform many surgeries have better outcomes than centers that perform only a few surgeries.

Facial weakness is associated with a reduced quality of life in a patient with NF2. Facial weakness can involve the upper face (forehead and eyes), mid-face (cheeks and nose), and lower face (mouth and chin). There are surgical and non-surgical procedures that can improve this problem.

Patients with upper facial weakness usually have difficulty completely closing their eyes. If left untreated, this can result in scarring of the surface of the eye (cornea) and ultimately blindness. For this reason, treatment of facial weakness is essential for NF2 patients. Patients with a “facial droop” are often self-conscious about their appearance. Dealing with issues of self-esteem is important in maintaining mental health for NF2 patients.

Difficulty with swallowing can be a major problem for NF2 patients after surgery. The major concern with swallowing problems is that food or liquids can travel into the lungs and cause infection or inflammation. Thus, it is helpful for centers to evaluate vocal cord function prior to surgery as a baseline.

Over the past 20 years, radiation therapy has become more popular for treatment of vestibular schwannomas in NF2. Radiation is effective in controlling the size of vestibular schwannomas. However, radiation is not as effective in maintaining hearing function in that ear. The majority of patients treated with radiation therapy experience significant hearing loss over the months and years following treatment. In addition, there appears to be a slight increase in risk of malignancy (cancer) in the treated area after radiation. For this reason, many (but not all) clinicians avoid radiation in chil-

dren with NF2 unless all other options have been considered.

Medical researchers are actively looking for new treatments for NF2-related vestibular schwannomas. Currently, clinical trials are underway to identify new medicines to treat these tumors.

What's the deal with meningiomas?

Meningiomas are benign (non-cancerous) tumors of the covering of the brain and spinal cord. About 50% of NF2 patients have meningiomas of the brain and another 50% of patients have meningiomas of the spine. These tumors cause symptoms when they press on the underlying brain or spinal cord. Typically, the growth of these tumors is slow and surgery can be planned in advance. The symptoms caused by meningiomas correspond to the part of the brain or spinal cord that they compress.

In general, the treatment for meningiomas is surgical removal by an experienced surgeon. Radiation treatment may be indicated for tumors that recur after treatment, for aggressive meningiomas that are not completely removed by surgery, and for tumors that are not surgically accessible.

Medical researchers are actively looking for new treatments for NF2-related meningiomas. Currently, clinical trials to identify new medicines to treat these tumors are underway.

What about spinal tumors: ependymomas, schwannomas, and meningiomas?

Spinal tumors are common in NF2 patients and can include ependymomas, schwannomas, or meningiomas. Ependymomas are tumors that develop from cells within the spinal cord; meningiomas and schwannomas are tumors that develop outside the spinal cord (but cause symptoms by compressing the spinal cord). The number of spinal tumors in NF2 patients ranges considerably. Some NF2 patients may have no tumors while others may have multiple tumors that affect every part of the spine. It is important to distinguish between tumors that are identified on MRI scan only and those that cause symptoms. Surgery is the mainstay of treatment for spinal tumors that cause symptoms; surgery is not usually recommended for asymptomatic tumors (although there are exceptions). In patients with multiple spinal tumors, it may be impossible to determine which tumor is causing the symptoms. In other patients, tumors may significantly compress the spinal cord but may not cause any symptoms. Thus, it is important to work with your physician to decide whether surgery is appropriate for a given spinal tumor.

Complications of surgery for the spinal cord include walking problems, muscle weakness, paralysis, problems with the bladder, problems with bowel movements, and pain. As with other tumors in NF2, finding an experienced surgeon is important when considering surgery for these tumors.

What about peripheral schwannomas?

Peripheral schwannomas are defined as tumors that occur outside of the brain and spinal cord. These can affect any nerve in the body including the arms, legs, torso, and skin. Surgery is the typical treatment for these tumors when they are symptomatic and growing. In general, most physicians observe tumors that are present but not causing symptoms or growing. Recently, there has been interest in using whole body MRI scans to evaluate these tumors but this technique remains investigational in 2012.

What about vision?

Maintaining good vision is a high priority for NF2 patients since they may experience significant hearing loss. Vision loss can either be congenital (present since birth) or acquired (develops in childhood or adulthood).

Non-tumor causes of vision loss include cataracts and benign growths of the retina (the seeing part of the eye). As noted earlier, upper facial weakness after surgery for vestibular schwannomas is a common cause of visual loss. In these instances, repeated scratches to the eyes surface can lead to vision loss and even blindness. NF2 patients with facial weakness should visit their ophthalmologist regularly to maintain eye health. Minor surgical procedures such as placement of a gold or platinum weight can help correct eyelid weakness.

What about problems with voice quality and swallowing?

Hoarseness is a problem that can develop over time (as tumors grow) or can occur suddenly (as a result from surgery). Hoarseness usually reflects weakness of one or both vocal cords in the throat. Weakness of the vocal cords may be associated with difficulty in swallowing. Examples of this include coughing during eating or drinking. The vocal cords are usually examined by directly looking at them with a scope in the physician's office.

Hoarseness can be treated in some patients through minor surgical procedures. For example, experienced surgeons can inject fillers beneath the vocal cords to improve voice quality. In some cases, permanent implants can also improve vocal cord function and improve speech.

Swallowing problems are typically identified on a swallow study (for example, a barium swallow). Speech therapists may be able to help patients improve their swallowing through exercises and education. In some cases, modification of a diet can reduce the risk from inefficient swallowing.

What about pain and/or neuropathy?

Pain is not a common problem in NF2 although it does occur in a minority of patients. When it occurs, it is usually associated either with surgery or with neuropathy. Neuropathy is a condition characterized by damage to the nerves in the body. Multiple medications have been approved for treatment of neuropathic pain, but these medications may not be fully effective (in some cases). Patients with significant pain should be referred to a pain specialist for a comprehensive evaluation.

Once Diagnosed:

What comes next?

What physicians do you need?

This depends on the type of tumors and the age of the patient. First, be clear not every doctor knows about NF2 or how to appropriately treat NF2. For some specialists you may be able to see someone in your home town, or close by. For other specialists you will need someone familiar with NF2 tumors, usually found in an NF clinic, which are often in large cities. For example, hearing aids may be effective for patients early in their hearing loss. Finding an audiologist and hearing aid specialist can probably be done in your local area. If your VS needs surgical intervention, you need an NF2-specialized team of doctors.

Good communication with your doctor is important. Patients who are deaf/hard of hearing should contact the hard of hearing office at the hospital to set up communication arrangements. Some NF Clinics use the CART (Communication Access Real Time Translation) which provides captioning services.

As for the pediatric population, there are NF clinics associated with children's hospitals. You will most likely have more than one physician involved in your care, and one will hopefully act as the coordinator. Clinic or not, you are your own team coordinator for you or your child's care.

This is a list of specialists that may be involved in the care of an NF2 patient:

Audiologist
Geneticist
Genetic Counselor
Oncologist
Neurosurgeon
Neurologist
Ophthalmologist

Otolaryngologist
(ear, nose & throat specialist)
Physical Therapist
Plastic Surgeon
Psychologist/Counselor
Pulmonologist
Speech Therapist
Radiation Oncologist

Educating Yourself

Symposiums

A symposium is a medical conference for patients and their families who are diagnosed with a medical condition. There are several symposiums annually around the country. To find one near you, visit nfnetwork.org. Be prepared to meet people there with all the different manifestations of NF2. If you are early in your diagnosis and have no outward signs, this can be overwhelming. Please remember that all of these people are living with NF2 and learning about NF2. The NF2 community can be an incredible source of support and inspiration.

Web Sites/On-line Networking

The wonder of the Internet is that it can be an incredible source of information, as long as it is legitimate information. Use filtered websites and the links found in the NF community. At the end of the booklet, additional NF2 resources will be provided.

Drug and Natural History Studies

If you stay even remotely connected to one of the NF sites, you will become informed of upcoming drug studies and their criteria for inclusion. A natural history study tries to gain insight about a particular disease/diagnosis, to learn about it from the beginning in

the hopes of appropriately steering further studies into effective treatments. Frequently check www.clinicaltrials.gov, type “NF2” into the browser, and see what comes up to stay informed about any current NF2-related studies.

Becoming involved with the NF Network

The amount of involvement is up to you. You could become the president of the local group or volunteer for a one-time event. There are many different types of ongoing needs. The Network offers an incredible amount of support and information. This is a lifelong diagnosis and a great opportunity to make lifelong friends.

Tissue Donation

An option to consider is donating a tumor sample. The NF Network can help put you in touch with an appropriate research lab in your area. An incredible amount of valuable research and information can be gained by the labs with donated samples. It is a great way to contribute to the solution for solving the NF2 puzzle.

To become involved and learn about NF activities in your area, please visit the NF Network website or contact us at the address or number on the back of this brochure.

Fundraising

Since NF2 is a rare disorder, research is underfunded by pharmaceutical companies and the government. There is a great need for funding of basic laboratory research and clinical trials. You may want to consider raising money to go towards NF2 research. At a time when you feel there may not be anything to do, this gives you something to do with a valuable outcome. You also might be surprised what you, your family, and your community are capable of.

Advocacy

Since 1996, NF research has been funded by Congress through the Congressionally Directed Medical Research Programs (CDMRP-NFRP) from the Department of Defense (DOD), and through the National Institutes of



Contact the NF Network so we can help you get started at www.nfnetwork.org.

Health (NIH). Every year the NF community competes with other diagnoses for part of a pool of money available for medical research. It is important for NF organizations, patients, and families to advocate for our piece of the pie. There are many ways you can do this. The easiest way is to write your Senators and Representatives and ask for their support. The local and national Network chapters will provide assistance with this at the appropriate times of the year when the budgets are being determined. Some NF2 patients and families meet with the local staffers in their home towns and others travel to Washington, D.C. and meet with their representatives face-to-face.

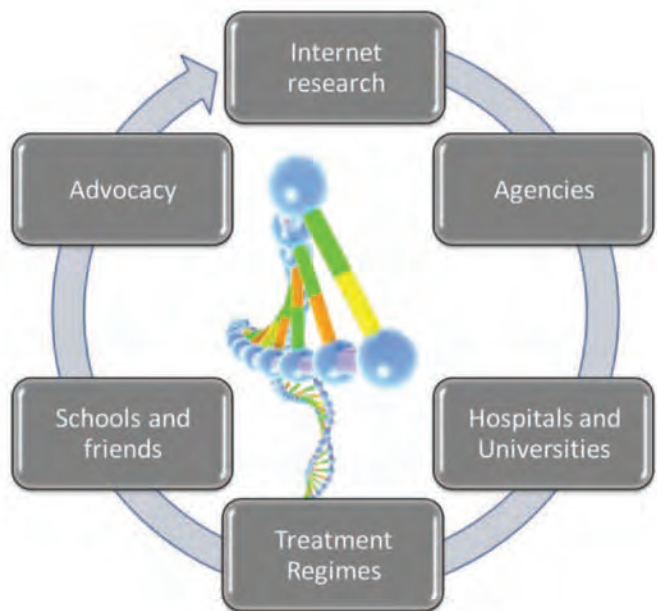
Helpful Ways to Organize

NF2 is a lifelong diagnosis and it can be very difficult to keep all the testing, reports and specialty physician visits organized. Some doctors prefer MRI scans be done on the same machine for consistency and easier comparison. Get copies/CD's of all physician reports, MRI's/CTscans/X-Rays, operative reports and pathology reports. Actually any report, as they are all important. Get a 3-ring binder with tab dividers and figure out a system that works for you. You cannot imagine how invaluable getting organized

from the beginning will be. Bring the binder to every physician visit and let them copy whatever reports are needed, and you have all the medical information at your fingertips that anyone would ever need. As health care becomes more digital, this will change with time. Have a stenographer's pad at every visit where you can write key words the doctors say, because you can only process so much in some visits. It is helpful to write any questions you may have for the specialist you are seeing before the appointment so that you make sure you cover everything.

A Patient/Parent Cycle

- Upon discovery, they rush to find information and hope
- They eventually find the right agencies to partner with
- As a result they find the right resources
- They begin to implement the correct regimes
- They are now the local experts
- They work to change the cycle



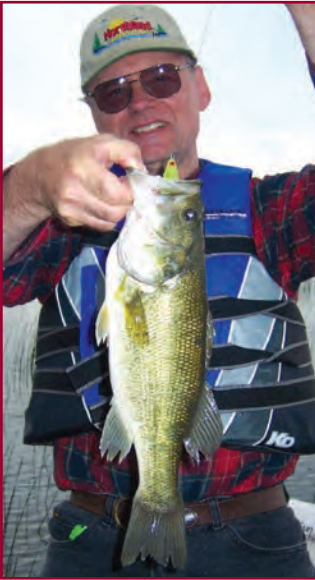
*The following 5 personal stories
are from individuals with NF2
who are all living their lives
to the fullest.*



*This is just a snapshot of all the
wonderful people and their
families whose community
you are now a part of.*

Personal Stories

Steve Reason



Steve Reason is 60 years old, married with two adult children, and living with NF2. He and his wife, Diane, reside in a small town in rural western IL. His journey with NF2 began in 1997 after consulting an ENT

for tinnitus, vertigo, and hearing loss in his right ear. Subsequent to finding a 2.1cm tumor on his right acoustic nerve that was surgically removed in 1997 at age 45, he was then diagnosed with NF2 six years later at age 51 when a second acoustic nerve tumor (vestibular schwannoma) was discovered in his only remaining hearing ear on the left side. An attempt to place a cochlear implant was made at the time of surgery but the CI failed leaving him profoundly deaf along with balance and vision problems.

Because he generally experienced overall good health for four decades prior to NF2, his life was basically turned upside down when he found himself

suddenly being thrust into a world of deafness without any established coping skills or mechanisms for communicating in place. That, along with mobility issues forced him to sell his business and reorganize his priorities. By the grace of God, buoyed by a strong faith and the amazing support of a loving family and friends, life for him is good once again!

Motivated to do what he can to help research find a cure, he enrolled in the NF2 Natural History Study at NIH in April, 2009 and is currently involved in fundraising, support, and advocacy through his local NF Network organization, serving on a committee dedicated solely to NF2 issues called NF2ACT! The networking and friendships established through these associations have given his life with NF2 an added dimension of meaning and purpose. His hope for the future is found in encouraging others to not give up.



Personal Stories

Leah Manth



Leah Manth lives in a suburb of Buffalo, NY and was only 7 years old when she was diagnosed with Neurofibromatosis Type 2. Since then she has endured surgeries on her spine and brain, developed facial paralysis and a partial hearing loss. Despite all she has been through Leah has an extremely positive outlook on life and she is an inspiration for anyone who meets her.

Her family decided early on to be part of the solution towards understanding and treating NF2. The first decision they made was to participate in various fundraising efforts to donate money to scientists who are trying to understand the basic science of NF2, which will then hopefully spark further research

and answers. Leah also enrolled in the NF2 Natural History Study initiated by the National Institute of Health (NIH) in 2009, where she visits twice a year. At NIH patients connect with experts on all the different aspects of NF2's effects, and also connect with others diagnosed and living with NF2 from all walks of life. We have met wonderful people and made genuine friendships on our NF journey, and we would never have met if not for NF. Leah and her family also lobby Congress to help continue funding at the federal level for all who may benefit from NF research and its effects on the body.

Leah is still the intelligent and inquisitive young lady that she always has been, who loves school, learning and life. She also enjoys all types of sports, but her favorite is basketball and anything outdoors like boating, fishing and skiing.



Leah is a child with NF2 and this affects her life, but she doesn't let it define who she is.

She is determined, along with her family and friends, to help discover reasonable treatments for NF2. For more information on Leah you can visit **www.leahmanth.com**.

Personal Stories

Ali Didier



June 15th, 2007 is the day that changed my life forever. I was 10 years old and was told I have NF2. The tumor in my right ear was growing with little chance of saving my hearing there.

I thought, How could this happen to ME!?

In 2008 I had my first surgery and sadly lost my hearing. In 2009 I had another surgery to remove a tumor behind my eye. Meanwhile, I enrolled in a 5-year NF Natural History study at the National Institute of Health (NIH). Every six months I fly from Chicago to DC for a week and have MRI's and other tests and meet with doctors. Soon the hearing in my left ear began to decrease and I had to get a hearing aid. It is too risky to remove the tumor in my left ear, so in mid-2011, I enrolled at NIH in an 18 month-long chemo study, the youngest of 14 patients.

Each year, our team Ali's Gaitors walks in the annual Great Steps for NF walk in Naperville, IL to help raise money for research. My mom's friends also started an NF walk in her hometown Effingham, IL. Having NF2 has introduced me to two whole new communities. People in the Deaf community have inspired me to be proud to be deaf. It also opened my eyes to the amazing people with NF, who are really supportive and kind and I am so glad I met them.

I also had my wish granted by the wonderful Make-A-Wish foundation: a trip to all the parks in Orlando, FL, a great week I will remember forever. My advice to anyone who is diagnosed with NF is no matter what, be happy, do what you love, and live your life to the fullest, I know I am.



Personal Stories

Matt Hay



Matt is a 35-year-old, married father of three, working for a marketing and advertising company in Chicago. He was diagnosed with NF2 during his sophomore year at Indiana University. Over the last 15 years, Matt has dealt with deafness, facial paralysis, vision problems and balance issues, resulting from the disease as well as long recoveries from a spinal surgery, two brain surgeries and six eye surgeries.

Matt deals with NF2 by focusing on what he can do rather than what he cannot, leading him to get involved with NF walks and eventually fundraising for NF causes via endurance events. He has participated in a number of runs, including the first of several marathons and, most recently, an Ironman distance triathlon.

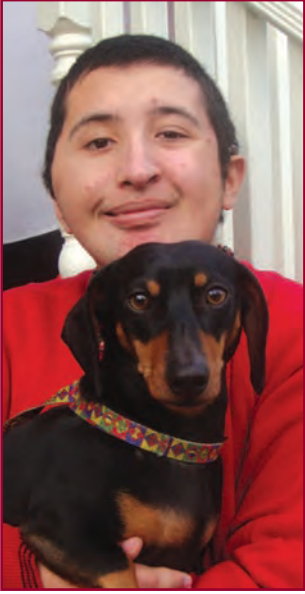
Matt also completed his MBA from the IU School of Business in 2010. The latter of these two accomplishments were things he once thought would forever be beyond his reach due to the obvious physical challenges brought on by NF2.

Matt is grateful for the awareness and fundraising work that the NF Network does for those with NF. His involvement with the local NF Network organizations and the physicians he has met at those events led him to learning about the auditory brainstem implant (ABI) that allows him to understand speech with the aid of lip reading and also learning about the pre-implantation genetic diagnosis (PGD) procedure that ensured he would not pass NF along to his children.

By staying involved with the NF community and taking part in walks, runs and other events, Matt is able to maintain certain aspects of his health that are within his control. He also appreciates the ‘upward spiral’ that can result both mentally and physically by getting involved with such a supportive community of caring people.

Personal Stories

Stephan Zepeda



In 1998 at the young age of 3, Stephan Zepeda was diagnosed with Neurofibromatosis (NF) Type 2. Stephan has endured numerous trips to the hospital for countless appointments, MRI scans, audiology tests and medication infu-

sions. In addition, Stephan has had numerous surgical procedures ranging from brain surgeries to remove tumors, procedures involving tumor decompression near the brain stem, to facial nerve reanimation to help correct facial paralysis.

In addition to spending countless hours in the hospital, NF has also robbed Stephan of his ability to hear simple sounds that many people so often take for granted. He has great difficulty communicating on a daily basis. Social isolation is a constant struggle.

NF has caused Stephan's coordination to deteriorate to the point where he has frequent falls. His vocal cords are

now showing signs of paralysis which causes labored breathing and voice hoarseness.

The disorder has also made us realize how fortunate and blessed we are. We have come to know so many great people and organizations as well. Also, doctors, nurses and other hospital staff who have dedicated their lives to help people like Stephan. Organizations such as the NF Network who continue to work tirelessly to raise funds to benefit people like Stephan. The Make-A-Wish Foundation who enabled Stephan to experience "the best day of my life" by visiting his boyhood hero Mr. David Ortiz.

Stephan is currently entering his senior year in high school and is preparing for the next phase of his life; a college education. Given all the obstacles in Stephan's life, he remains funny, witty, outgoing and a courageous young man. He enjoys his video games, surfing the web on his Ipod and an occasional sleepover with a friend. This disease has taught all of us not to take life for granted and to take each day one day at a time and to appreciate the simple things in life. As a family, we remain positive that in time there will be a cure or an effective treatment which will enable Stephan to lead a long, healthy life.

Additional Resources

NF Network is the leading national organization advocating for federal funding for NF research and the development of local NF organizations through many activities. The NF Network website will help you contact your local NF organization, find an NF doctor in your area and keep you up-to-date on advocacy efforts on federal funding for NF research. To stay current on NF information sign up for the email list at **www.nfnetwork.org**.

Trial Talk (TT) is a streamlined email group list created exclusively for NF2 patients (and/or family member with NF2), who are interested in finding a cure (or effective treatment) for NF2. It provides a confidential forum to discuss or participate in clinical trials.

NF2Trialtalk-subscribe@yahoogroups.com

ClinicalTrials.gov is a registry and results database of federally and privately supported clinical trials conducted in the United States and around the world. ClinicalTrials.gov gives you information about a trial's purpose, who may participate, locations, and phone numbers for more details. ClinicalTrials.gov currently contains 128,269 trials sponsored by the National Institutes of Health, other federal agencies, and private industry. Keywords: NF2, Neurofibromatosis Type 2

Advocure NF2 is an international coalition committed to ensuring the rapid translation in the advancement of NF2 research into targeted therapies for the NF community. Advocure will monitor relevant research and drug developments worldwide and lobby to achieve our goal of expediting much needed systemic therapy for this devastating CNS disorder.

advocureNF2.org

NF2 Crew is an online-based support community for patients and family members (or loved ones) with Neurofibromatosis Type 2.

Nf2crew.org.

Glossary of Terms

Acoustic Neuroma (now called **vestibular schwannoma**) - are benign tumors of the eighth cranial nerve (vestibulocochlear nerve).

Auditory Brainstem Implant (ABI) - are devices that can be implanted during surgery for removal of vestibular schwannomas. These implants can transmit some auditory sensations to the brain.

Audiometry - Measures how loud a noise must be for a patient to hear it and measures how well a person can understand spoken language.

Autosomal Dominant - is a term used to describe how a genetic condition is passed from parent to child. Conditions that are passed in autosomal dominant fashion occur in 50% of offspring of affected parents.

Cancer - is a type of tumor (growth) characterized by continued growth and may spread to distant portions of the body (metastasisize).

Cataract - is a clouding of the lens of the eye. Senile cataracts are those that occur later in life and are usually caused by ultraviolet radiation (sunlight). Juvenile cataracts occur early in life and often occur in the setting of NF2.

Cavernous Sinus - is the region of the skull base that houses the blood supply and many cranial nerves.

Chromosome - is a genetic structure that contains genes. Humans have 22 pairs of non-sex chromosomes plus a pair of sex chromosomes, X and Y from each parent. The NF2 gene is located on chromosome 22.

Cochlear Implant (CI) - is a type of hardware that is implanted into the inner ear. These implants can transmit auditory sensations to the inner ear.

Computer Assisted Tomography - CT scans are a form of x-rays that can create three-dimensional images of the brain and other body parts.

Cyber Knife - is a trademarked term that applies to a specific type of focused radiation. Other equivalent types of radiation include Gamma knife, LINAC (linear accelerator) radiation, and proton radiation.

Eighth Cranial Nerve - has two branches including the acoustic nerve and the vestibular nerve. The acoustic nerve transmits sound from the environment to the brain. The vestibular nerve transmits balance signals from the environment to the brain.

Electromyogram (EMG) - is a test of the muscles and nerves to determine if they are functioning normally.

Ependymoma - are benign (non-cancerous) tumors that arise from the normal cells within the spinal cord. In NF2 patients, they most commonly occur at the junction of the brain and spinal cord or in the spinal cord.

Fifth Cranial Nerve (trigeminal nerve) - transmits sensations from the face to the brain.

Gamma Knife - is a trademarked term that applies to a specific type of focused radiation. Other equivalent types of radiation include Cyber knife, LINAC (linear accelerator) radiation and proton radiation.

Gene - a segment of DNA within chromosomes that transmits information about specific traits (for example hair color).

Magnetic Resonance Imaging - MRI is a type of imaging in which 3-dimensional images are produced by magnets (without radiation). These images are typically high quality, which is useful for viewing the brain or spinal cord.

Meningioma - are benign (non-cancerous) tumors of the covering of the brain or spinal cord.

Neuropathy - is a neurologic condition in which nerve cells in the body do not function normally. This is typically experienced by patients as either numbness, tingling, or pain.

Radiation Therapy - is a medical procedure in which focused radiation is provided to a specific part of the body in hopes of treating tumors. Radiation can be provided in one day (radiosurgery) or over multiple days (radiotherapy). The dose of radiation is measured in Grays.

Schwannoma - are a benign (non-cancerous) tumor arising from the covering of a nerve.

Seventh Cranial Nerve (facial nerve) - transmits information from the brain to the muscles of the face. Abnormalities of the seventh cranial nerve leads to facial weakness.

Third Cranial Nerve (oculomotor nerve) - transmits information from the brain to the muscles of the eye. Abnormalities of the third cranial nerve leads to double vision and weakness of eye movement.

Tinnitus - is the perception of sound, often described as a ringing sound in the ear, when no actual sound is present in the environment.

Tumor - is an abnormal growth in the body. Tumors can either be benign (non-cancerous) or malignant (cancerous).

Vestibular Schwannoma (previously called acoustic neuroma) - are benign tumors of the eighth cranial nerve (vestibulocochlear nerve), which affects hearing and balance.

A portrait of a young woman with long brown hair and glasses, smiling. She is wearing a light green shirt. The background is slightly blurred, showing a plant on the left and wooden cabinets on the right. The entire image is framed by a dark red border.

**This booklet is dedicated to
Ashley Sexton.**

**The generosity and support of her family and
friends made this project possible.**

**“Having NF2 has changed me because
it is forever a part of me.”**



www.nfnetwork.org

Contact us at:

NF Network

213 S. Wheaton Ave.

Wheaton, IL 60187

630-510-1115

Toll Free 1-800-942-6825

admin@nfnetwork.org