Neurofibromatosis Type II (NF2) is a rare condition that results in slow growing noncancerous, but still dangerous tumors, primarily in the Central Nerve System; in the brain and along the spinal cord. These tumors can result in different forms of nerve, brain and body damage. Damage development depends on actual location of growth for each individual tumor. Many people with NF2 do not always cope with the same issues. Eye issues are also a common occurrence; some are unrelated to tumor nerve damage, like Juvenile Cataracts.

There is no treatment to stop the growth of all tumors that are the result of NF2. Many people with NF2 undergo multiple cranial and spinal surgeries in their lifespan. Tumors are monitored with frequent MRIs throughout their lives.

While NF2 is genetic, many instances of it are people born with it as a result of spontaneous mutation and have no family history of it. Birth with spontaneous mutation makes it hard to get necessary treatment to help manage issues since with earlier diagnosis symptoms can be managed to help reduce the physical impact of NF2. Earlier diagnosis allows for potential minor medical interventions to help delay some of the worst potential symptoms and issues.

Neurofibromatosis Type II (NF2), also known by the name "MISME Syndrome" (Multiple Inherited Schwannomas, Meningiomas and Ependymomas), or Bilateral Acoustic NF (BAN), is a genetic Autosomal Dominant condition. It affects an individual throughout the course of their life. The mutated or missing gene that results in NF2, a tumor suppressor gene, should create a protein known as **MERLIN (Moesin-Ezrin-Radixin-Like Protein)**. When the Merlin Protein is nonfunctional, tumor development occurs.

Tumor growth is due to different issues, but exactly how NF2 effects each individual can vary depending on an individual's exact mutation type. Other variables might also be responsible and additional research is necessary to understand additional causes of increased growth.

NF2 Tumors

Tumor formation types can develop, including some combination of the following 5 categories of tumor growth:

- Bilateral Vestibular Schwannoma
- Additional Brain Tumors
- Spine Tumors
- Peripheral Nerve Schwannoma
- Skin Surface: Schwannoma and Neurofibroma

Diagnosis

Criteria of NF2 Diagnosis the Manchester Criteria

As of 1997 the diagnostic criteria for NF2, known as the Manchester Criteria, requires one of the following three conditions to apply:

- Bilateral Vestibular Schwannoma
- 1 or more 1st degree relative with NF2 + unilateral Vestibular Schwannoma at <30 years
- 2 of the following: Meningioma, Glioma, Schwannoma, Juvenile Posterior Lenticular Opacities

Diagnosis Methods

1. Neurologist Examination
   - Physical Exam
   - MRI with and without Gadolinium image contrast, brain and spinal cord.

2. Neuro-Ophthalmologist
   Eye Exam for a variety of NF2 specific eye issues

3. Otologic Examination done by an ENT Doctor (Ear Nose Throat), also known as an Otolaryngology
   - Audiometry Hearing Test - Speech Recognition/Discrimination Tests
   - ABR - Auditory Brain Stem Response
   - MRI - Magnetic Resonance Imaging of with and without Gadolinium of the brain and spinal cord.

4. Dermatologist Examination

5. Geneticist - Genetic Test

NF2 Information & Services

NF2 Information and Services is dedicated to: education of NF2 health issues and treatments, with charitable funds to go to education, scientific advancements and support for people with NF2.

Learn more about NF2 at: www.nf2is.org

Neurofibromatosis Type 2

Basic Treatment Guide:

Diagnosis, Management
& Who to See

Doctors / NF2 Specialists

People with NF2 need to see specific specialists for different issues, finding a treatment location where the NF Clinic Coordinator or Administrator can help plan out MRI's and doctor visits with Interpreters helps. Otherwise the only doctors an individual with NF2 might see yearly are; Neuro-Oncologists, Neuro-Ophthalmologist and Otolaryngologist also known as an ENT.

It is helpful for treatments to have a hospital that has NF2 specialists for all the doctors needed to be seen. It helps the team plan treatments better based on all of your issues and can be reviewed by all of your doctors when needed.

Doctors that a person with NF2 might need to see include:

- Neuro-Oncologists
- Neuro-Ophthalmologist
- Geneticist
- Brain & Spine Neurosurgeons (Microsurgery)
- Brain & Spine Neurosurgeons (Radiosurgery)
- ENT (Ear Nose and Throat Doctor) also known as an Otolaryngologist
- Audiologist
- Dermatologist
- Plastic Surgeon.

Doctor Types

- **Neuro-Oncologists:** For regular Neurological checkups, the proper doctor to see is a Neuro-Oncologist, not a Neurologist. This is a special class of doctor who understands the results of tumor development and potential tumor suppressors an individual might qualify for.
- **Neuro-Ophthalmologist:** Many NF2 issues can be identified in a Neuro-Ophthalmologist exam. Since many eye issues that develop at early ages can result in vision loss these visits should be done regularly. A Neuro-Ophthalmologist would be able to review MRI scans and can even see tumor pressure and other issues that Ophthalmologist would not see, or know to look for, in a regular eye exam.
- **Geneticist:** A geneticist does not need to be a specialist in NF2, but be able to explain how Autosomal Dominant conditions might affect a family. The geneticist would also know what means would work in obtaining an accurate diagnosis. Genetic tests themselves are generally done in different locations in each country.
- **Brain & Spine Neurosurgeons (Microsurgery):** A doctor that specializes in brain and another in spine, specifically for NF2 since NF2 tumors are unique in surgical removal.
- **Brain & Spine Neurosurgeons (Radiosurgery):** Surgeons that know long term consequences of NF2 and Radiosurgery.
- **Otolaryngologist aka ENT (Ear Nose and Throat Doctor):** A doctor capable of ABI surgery with help from another surgeon.
- **Audiologist:** Audiologists are specialists who understand how a wide variety of hearing implants work. This includes Cochlear Implants (CI), but not always all Cochlea Devices. With Neurofibromatosis Type 2, the Auditory Brainstem Implants (ABI) are the Cochlea Devices more commonly require, therefore Audiologists in NF2 Treatment Centers Need to know how to MAP, aka tune, ABI's as well as CI's.
- **Dermatologist:** Someone that would know when a tumor can be removed surgically in office or needs closer examination before removal if a tumor is attached to a nerve.
- **Plastic Surgeon:** Trained in Facial Reanimation methods and eye springs.

NF2 Issues

Some Common Eye Vision Issues

- Juvenile Cataracts - PSC and CC
- Combined Hamartoma of the Retina & Retinal Pigment Epithelium
- Retinal Detachment
- Congenital Glaucoma
- Diplopia (Double Vision)
- Retinal Microaneurysms
- Opric Disc Glioma
- Epiretinal Membranes
- Papilledema
- Nystagmus and Oscillopsia
- Dry Eyes / Excessive Tearing

Tumor Nerve Damage

- Hearing Loss
- Facial Nerve Damage
- Brain and Cranial Nerve Damage
- Swallowing Difficulties
- Loss of Mobility
    - Spinal Cord Compression - Spine Damage
    - Balance Issues
    - Peripheral Neuropathy
    - Muscle Wasting
    - Drop Foot / Foot Drop9.5

Some Additional Concerns

- Café-au-lait Spots – typically less than 5
- Skeletal Abnormalities – following spine surgery or spine tumor growth
- Anxiety and Depression
- Nerve Pain
- Migraines

Location Recommendation

With Neurofibromatosis Type 2, a condition that results in the need to see multiple doctors following diagnosis, it is important to pick one hospital for all medical needs other than the care needed from a General Practitioner or Pediatrician.

The importance of having all doctors who treat an individual at one hospital cannot be stressed enough. Most large hospitals have gone to a computer system. 'Patient Portals' of record keeping so that any physician on and individual's case has access to all records. This information can be accessed by any doctor in the hospital. This prevents such things as double testing but more importantly any one doctor on a case is less likely to miss a crucial piece of information.

Neuro-Ophthalmologist can access recent MRIs and surgeons can access balance information as well as current MRIs. In fact the longer an individual is seen by the same team of doctors in the same Center or Hospital, the longer their histories become and MRIs that are five years apart for example can be compared to see growth or stability over a longer period of time. This in and of itself is very helpful to clinicians reviewing tumor changes over time to determine treatment recommendations.

Tumors may not seem to be growing from year to year but when reviewed over a longer time, side by side with digital comparisons in mm, completely different result may become obvious.

When choosing a treatment center it is best that all of the types of doctors be available to a patient and that these doctors are well-versed in the NF2 literature. In addition, staying with an institute if possible can be important for the reasons cited above. However, changing locations for treatment is clearly necessary if an individual has a problem with the level of care received.

Most hospitals are also making their patient records available to the patient so an individual can become proactive in their care rather than just reactive. More and more "Patient Portals" are being developed. These can be used to help your doctors follow your care.