



Neurofibromatosis Type 2: An Introduction for patients

About Neurofibromatosis (NF)

NEUROFIBROMATOSIS (NF for short) is a collective name for a group of conditions that causes benign tumours in the nervous system. NF occurs all over the world and affects men and women equally.

There are two types of NF: NF1 and NF2.

In **Type 1** the tumours grow mainly on the skin and people with NF1 are at an increased risk of having other problems such as difficulties with learning, bone problems and nerve tumours inside the body. People with NF1 have a number (usually at least 6) of harmless flat brown marks on the skin called café au lait spots.

In **Type 2** the benign tumours grow on nerves within the body. Most people with NF2 develop tumours on both hearing nerves. Benign tumours may also develop on the nerve roots as they leave the spine and on the coverings of the brain. Some people with NF2 have a few benign tumours on the skin nerves but, in contrast to NF1, have few, if any, café au lait spots.

They are very different conditions and it is extremely unlikely that you would have both NF1 and NF2

This leaflet is just about NF2.

When you first find out about NF2 you may experience a variety of emotions and it is quite common to feel overwhelmed and confused. This information is an introduction to the condition.

What causes NF2?

NF2 is a genetic disorder that is caused by a **misprint in a single gene on Chromosome 22**. The misprinted gene will be present at birth but signs of the condition do not usually appear until the teenage years, twenties or later.

NF2 may be passed on from parent to child at the time of conception, or it may start in a family with no previous history of the disorder. A person who has NF2 has a 50% (or 1 in 2) chance of passing on the condition to each of his/her children.

About 50% of people with NF2 are the first person in the family to be affected. Someone with NF2 usually has a 50% chance of passing it onto their children. However, if you are the first person in the family to be affected, the risk can be lower than that. When people are first diagnosed, they are referred to a Clinical Geneticist or Genetic Counsellor to discuss the genetic risk.

What is NF2?

NF2 is a variable and unpredictable condition affecting people in different ways. The hallmark of NF2 is the development of **benign** tumours called vestibular schwannomas (formerly called Acoustic Neuromas) which grow on **both** hearing nerves. In addition to these, other benign tumours associated with the nervous system may occur, in particular:-

- * The lining of the brain (meningiomas)
- * The spine (meningiomas, schwannomas)
- * The skin (schwannomas)

NF2 can also cause some ocular (eye) changes such as cataracts. These are often present from an early age but may not cause any significant visual problems.

Each hearing nerve has two parts. The Cochlear nerve carries information about sound and the Vestibular Nerve carries information about balance to the brain.

Over time these tumours are likely to cause deafness. A tumour on one side may grow at a different rate to the one on the other side. The size of the tumours may bear little relationship to the degree of deafness. For example, a small tumour may produce deteriorating hearing whilst a large tumour may only cause minimal hearing loss.

How does NF2 affect you?

Most NF2 tumours are slow growing and may cause minimal problems for years. Although they are not malignant (not cancerous) their position may produce significant symptoms.

For the majority of people, the most common first symptoms of NF2 are:-

- ❖ Gradual hearing loss
- ❖ Tinnitus (ringing or roaring in the ears)
- ❖ Unsteadiness, particularly when walking on uneven ground or in the dark.

These symptoms are caused by tumours on the hearing nerves (vestibular schwannomas).

Other symptoms may relate directly to the pressure caused by tumours on the spine or on the lining of the brain. For example:-

- ❖ Headaches
- ❖ Change in vision
- ❖ Change in sensation, pain or weakness of an arm or leg.

While the problems in NF2 can be worrying, treatments are improving and support is available through the various doctors and therapists who you may be in contact with.

Management and Treatments

NF2 is a rare disorder. In 2010, services for patients with NF2 became part of the National Specialised Services for England. This service provides 4 designated centres of excellence, which would be responsible for overall management of NF2 patients in England. Each centre is linked to a number of 'satellite' centres. The aim being to provide, through a multidisciplinary approach, a more effective and cohesive healthcare system for patients with NF2. The four centres are based in London, Cambridge, Manchester and Oxford.

It is very important to have a detailed discussion with a specialist who is familiar with NF2, to find out about the various options that may be available and to consider the benefits and risks of any operation or treatment.

You will need regular examinations to check hearing and neurological function. Scans of the brain and spine, balance tests as well as eye examinations may be needed annually.

The timing of any surgery to remove tumours should be carefully considered. Not every tumour seen on scans needs to be removed. Some NF2 tumours may show very little growth over many years.

Treatments are based **mainly on the symptoms the patient describes**, alongside results of physical examinations, scans and hearing tests.

Operations to remove the tumours on the hearing nerves (vestibular schwannomas) should be performed by an Ear, Nose and Throat Surgeon and/or Neurosurgeon with special expertise in NF2.

The treatment of vestibular schwannomas will depend on:-

- ❖ The size of the tumours
- ❖ Whether they are pressing on the brain as well as on the hearing nerve
- ❖ How rapidly they are growing
- ❖ How much hearing loss the tumours have caused

Some people choose to have X-ray treatment rather than an operation. **The Gamma knife or stereotactic surgery** shrinks the vestibular schwannomas. **This treatment does have risks** and should only be undertaken after discussion with Doctors who are familiar with NF2 and its management.

Whichever option is chosen it is difficult to treat a vestibular schwannoma without causing damage to the hearing nerve and subsequent deafness. Sometimes damage to other nerves, which are close to the hearing nerve, is unavoidable. Most people who become deaf through NF2 learn to lip read very well.

Some people can now be offered an auditory brainstem Implant (ABI) to help with hearing after surgery. The ABI does not restore hearing but gives an awareness of certain environmental sounds and is an aid to lip-reading. ABI implant surgery is only available in Specialist NF2 Centres.

If it becomes necessary to have other NF2 tumours removed - on the spine, skin or lining of the brain, the operations are usually more straightforward and carry fewer risks.

Hearing Rehabilitation

Hearing Link is the national organisation that helps hearing impaired adults and their families to manage the impact that hearing loss can have on their lives. The organisation makes it easy for people to find information, support and ways to connect with others. It also brings people together so experiences can be shared and feedback can be gathered and passed on to service providers.

Hearing Link focuses on adults who have acquired hearing loss during their lifetime and who prefer to communicate through the spoken and written word rather than sign language. It delivers highly personalised services and specialises in helping people to adjust and manage the practical, emotional and social challenges that hearing loss can bring.

What is the NF2 programme?

The programme is a week-long residential rehabilitation service attended by up to 8 adults with hearing loss. Participants are also encouraged to bring along a family member, partner or close friend.

Known as Hearing Link's Intensive Rehabilitation Programme, the aim of the week is to help people adapt to hearing loss and to equip them with the skills and information to manage their changing health status. Through increased knowledge and understanding people with NF2 are helped to manage the emotional and practical challenges the diagnosis brings.

For more information or advice:-

For more information about NF2 ask your GP or specialist to refer you to a geneticist.

To find out if other family members have NF2: ask for a referral to a geneticist at a regional genetics centre.

Further information regarding hearing difficulties, including tinnitus and balance problems, is available on The Nerve Tumours UK Website: NerveTumours.org.uk

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