



THE NEUROFIBROMATOSIS
ASSOCIATION

Neurofibromatosis Type 2 Information 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 main types of Nf – Nf1 and Nf2.

In **Type 1** the tumours grow mainly on small nerves in the skin and people with Nf1 are at 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 nerves running down 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 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. More detailed information can be requested from The Neurofibromatosis Association.

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.

WHAT IS Nf2?

Nf2 is variable and unpredictable affecting different people in different ways. Almost everybody who has Nf2 develops benign tumours called vestibular Schwannomas (formerly called acoustic neuromas) which grow on both hearing nerves. Each of these nerves 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. Other **benign** tumours associated with nerves inside the body **may** occur, in particular:

- Of the lining of the brain (meningiomas)
- On the spine (meningiomas, schwannoma)
- On the skin (schwannomas)

Cataracts are often present from an early age but may not cause significant problems with vision.

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.

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.

WHAT TREATMENTS ARE AVAILABLE FOR Nf2?

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 Schwannoma 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 give a return of hearing but gives an awareness of certain environmental sounds and is an aid to lipreading. This new treatment 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.

FOR MORE INFORMATION OR ADVICE

QUESTION

ANSWER

For more information about Nf2.

To find out if other family members have Nf2 or you want a blood test to look for the Nf2 gene.

For advice about tinnitus and learning different ways of communicating.

For advice and help with communication aids eg using the telephone, pagers, alarm systems.

For advice about how to minimise balance difficulties.

To find out about exercises for facial weakness.

For help with speech and eating difficulties.

For support in adjusting to hearing loss and change of life issues.

The NfA has a more detailed booklet. Ask your GP or specialist to refer you to a Geneticist.

Ask for a referral to a Geneticist at a Regional Genetics Centre.

Ask the Hearing Therapist at your local Audiology Department.

Ask your local Social Service Department for a meeting with a Specialist Worker for the Deaf.

Ask for a referral to a Physiotherapist at your local hospital. Some Hearing Therapists also specialise in balance problems.

A Physiotherapist can help.

Talk to a Speech and Language Therapist at your local hospital

Ask your Hearing Therapist or Nf Specialist Advisor about The Link Centre, Eastbourne.

THE NEUROFIBROMATOSIS ASSOCIATION

The Neurofibromatosis Association (NfA) has the following aims:

- To provide accessible, up to date **information** on Neurofibromatosis – not only to those who are affected and to their families, but also to doctors, therapists and other professionals.
- To maintain a network of professional Neurofibromatosis Support Co-ordinators – who are able to **support** those who are affected and their families.
- To promote **awareness** and understanding of the problems encountered in the disorder and to improve clinical care.
- To raise funds to further **research** into improved treatments and, ultimately, to find a cure for the disorder.

The Nf. Association provides opportunities for people with Nf2 and their families to meet others and spend time together. Events include weekend breaks where people can also get information from experts, and social get-togethers in informal settings around the country.

Recognising that Nf1 and Nf2 are two distinct conditions, all Nf2 members of the NfA receive a newsletter designed specifically for people affected by Nf2, as well as the quarterly Nf Association Newsletter which is sent to all members.

The NfA can put you in touch with an Nf Specialist Advisor in your region. They are there to provide support, information and advice not only for those who have Nf2 but also for the family and professionals concerned with their care.

We are grateful to the following people for their help in compiling this booklet:

Professor Gareth Evans
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Guys and St. Thomas` Hospital, London

Dr Susan Husan
Consultant in Clinical Genetics
Churchill Hospital, Oxford

Dr Elizabeth Rosser
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Institute of Child Health, London

Staff and Management Committee of the
Neurofibromatosis Association

and particularly to the 10 patients who gave their valuable comments and contributions.

Produced
with financial
support from



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For more information and a full list of publications please contact:

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