



# The Neurofibromatosis Association

Registered Charity No 1078790



## 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 patches.

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 patches.

*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 and a larger print version of both books is available.**

## 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 a variable and unpredictable condition 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 from 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, schwannomas)
- 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 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 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

<b>For more information about Nf2.</b>	<b>The Nf Association has a more detailed booklet. Ask your GP or specialist to refer you to a Geneticist.</b>
To find out if other family members have Nf2 or if you want a blood test to look for the Nf2 gene.	Ask for a referral to a Geneticist at a Regional Genetics Centre.
<b>For advice about tinnitus and learning different ways of communicating.</b>	<b>Ask the Hearing Therapist at your local Audiology Department.</b>
For advice and help with communication aids, eg using the telephone, pagers, alarm systems.	Ask your local Social Service Department for a meeting with a Specialist Worker for the Deaf.
<b>For advice about how to minimise balance difficulties.</b>	<b>Ask for a referral to a Physiotherapist at your local hospital. Some Hearing Therapists also specialise in balance problems.</b>
To find out about exercises for facial weakness.	A Physiotherapist can help.
<b>For help with speech and eating difficulties.</b>	<b>Talk to a Speech and Language Therapist at your local hospital</b>
For support in adjusting to hearing loss and change of life issues.	Ask your Hearing Therapist or Nf Specialist Advisor about The Link Centre, Eastbourne.

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

First published by the Neurofibromatosis Association 2001

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

Professor Gareth Evans  
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now Hon Consultant in Clinical Genetics  
St Mary's Hospital, Manchester

Dr Elizabeth Rosser  
Consultant in Clinical Genetics  
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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Registered Charity Number 1078790