

# Neurofibromatosis type 1

**This leaflet offers information about neurofibromatosis 1 (NF1), how it is diagnosed, and how it might affect you. If you have any questions or concerns, please speak to the clinical genetics department (contact details listed at the end of this leaflet).**

## What is neurofibromatosis type 1?

Neurofibromatosis type 1 (NF1) is an inherited condition, affecting one person in every 3000–3500 in Britain. It is completely separate to NF2 (neurofibromatosis type 2), which is less common, affecting one in every 35,000 people.

Although NF1 is something you are born with, some symptoms develop gradually over a number of years. The severity of the condition can vary from person to person.

Certain features are present in almost everyone with NF1. These include:

- Flat, coffee-coloured patches on the skin known as "café au lait spots". They usually begin to appear during the first year of life and gradually grow in size and number. Many people have café au lait patches, but individuals with NF1 tend to have six or more, which can vary in size from 0.5cm in children/1.5cm in adults to several centimetres in diameter.
- Freckling around the neck, under the armpit or in the groin.
- Small lumps on the skin known as neurofibromas. These usually begin to appear during adolescence and tend to increase in number throughout life. The number of neurofibromas varies widely in people with NF1, from only a few to several hundred. Sometimes patients may have much larger, more spread-out lumps known as "plexiform neurofibromas". These may be present at birth or appear during early childhood.
- Small, harmless lumps on the iris (the coloured part of the eye) known as "Lisch nodules". A special eye examination is needed to detect these.

## How is NF1 diagnosed?

In adults, the diagnosis of NF1 is normally clear-cut. Initially, children with NF1 may only have café au lait spots, but other features can appear over time. For this reason, children cannot be confidently diagnosed with NF1 before the age of three.

NF1 is caused by a change (mutation) in a gene. In the past, genetic testing for NF1 was not performed routinely because gene changes were difficult to detect. Now, although diagnosis of NF1 is usually based on clinical examination, gene testing may be considered helpful in certain circumstances. Advice may be obtained from local genetics centres or national neurofibromatosis centres.

## How is NF1 inherited?

Each child of a person with NF1 has a one in two chance of inheriting the condition. The symptoms of NF1 can sometimes be very mild, and people with NF1 may not realise that they carry the mutation until other family members have been diagnosed.

For around half of people with NF1, no signs of the condition are found in either of their parents, making it most likely that the gene mutation has occurred for the first time in the person with NF1. In this situation, parents without NF1 have a very low chance of having another child with NF1.

Anyone considering starting a family can be seen by their local genetics service to discuss the options that are available to them. This may include pre-implantation genetic diagnosis (PGD) and/or testing during pregnancy.

## **What medical problems are associated with NF1?**

Many people with NF1 do not experience problems as a result of the condition. However, there are a number of complications that people with NF1 may develop. Unfortunately, it is difficult to predict whether or not these problems will occur. All patients should have a check-up with their doctor once a year. Your GP may wish to refer you to one of the national centres for neurofibromatosis in London (located at Guy's and St Thomas') or Manchester (located at Central Manchester University Hospitals).

Below is a list of some of the health issues associated with NF1:

### **High blood pressure (hypertension)**

High blood pressure is common in NF1, particularly in adulthood. In most cases, no underlying cause is found. Very rarely it is associated with narrowing of the artery to the kidney (renal artery stenosis) or a tumour of the adrenal gland (phaeochromocytoma), which may require a combination of medical and surgical treatment.

### **Learning difficulties**

Many people with NF1 also have learning difficulties and may require extra support at school and in the work place. Hyperactivity, autism spectrum disorder and clumsiness also seem to be more common in people with NF1. Only a small percentage of people with NF1 have severe learning difficulties.

### **Tumours**

Neurofibromas are small tumours which grow on nerve coverings. They are usually benign (non-cancerous) and do not spread to other parts of the body or cause symptoms.

A small number of individuals with NF1 will develop malignant (cancerous) tumours. It is important to realise that the risk of developing a cancer that is definitely related to NF1 is low. However, if you notice a lump growing rapidly or becoming unusually painful or hard you should consult your doctor and ask to be referred to one of the national centres for neurofibromatosis.

Neurofibromas occasionally arise from the nerve roots of the spinal cord, causing nerve compression and pain, weakness or numbness in the legs or arms. These may require an operation and should be monitored by specialist teams.

Tumours of the optic nerve may not cause any symptoms at all, but can sometimes cause a squint, reduced vision, double vision or blurred vision. If this kind of tumour is found, it needs to be monitored by an ophthalmologist (eye doctor), and treatment is sometimes required. Children with NF1 should have their vision checked yearly.

### **Bone problems**

Vitamin D, which regulates the amount of calcium in the body, may be low in people with NF1, and this can contribute to bone problems.

NF1 can sometimes be associated with curvature of the spine (scoliosis) which may occasionally need surgery. Rarely, children are born with, or develop, bowing of the bones in the lower leg or arm. A balanced diet containing calcium is helpful to maintain bone health.

## What treatments are available for NF1?

There is currently no cure for NF1, but you will be monitored regularly for further problems, and treated for these if they develop. This might involve:

- surgery to remove tumours and improve bone abnormalities
- medication to control secondary conditions like high blood pressure
- therapy for behavioural problems.

In many cases, careful monitoring and treatment can help people with NF1 to live a full life.

## Is there anything I can do to help myself?

Women are usually encouraged to take part in the National Breast Screening Programme from the age of 50, however, there is evidence that women who have NF1 are at moderately increased risk of developing breast cancer and are therefore eligible for earlier annual mammograms (between the ages of 40 and 49). If you would like to make an appointment for early screening, this can be arranged through your GP.

### Contact us

If you have any questions or concerns, please contact your **local neurofibromatosis team** or speak to the **neurology team** on **020 7188 3966**. Alternatively, you can visit our website at: [www.guysandstthomas.nhs.uk/services/managednetworks/genetics](http://www.guysandstthomas.nhs.uk/services/managednetworks/genetics)

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit [www.guysandstthomas.nhs.uk/leaflets](http://www.guysandstthomas.nhs.uk/leaflets)

## Further information:

### The Neuro Foundation

The Neuro Foundation provide information, support, advice for people with neurofibromatosis and their families/carers.

**t:** 020 8439 1234    **e:** [info@nfauk.org](mailto:info@nfauk.org)    **w:** [www.nfauk.org](http://www.nfauk.org)

### Pharmacy Medicines Helpline

If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.

**t:** 020 7188 8748 9am to 5pm, Monday to Friday

### Patient Advice and Liaison Service (PALS)

To make comments or raise concerns about the Trust's services, please contact PALS. Ask a member of staff to direct you to the PALS office or:

**e:** 020 7188 8801 at St Thomas'    **t:** 020 7188 8803 at Guy's    **e:** [pals@gstt.nhs.uk](mailto:pals@gstt.nhs.uk)

### Language Support Services

If you need an interpreter or information about your care in a different language or format, please get in touch using the following contact details.

**t:** 020 7188 8815    **fax:** 020 7188 5953

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