

Neurofibromatosis (Nf)

WHAT IS Nf?

Neurofibromatosis (pronounced new-roe-fye-broe-mer-toe-sis) is a genetic disorder which causes tumours to form on nerve tissue anywhere in the body. There are two main types, Nf 1 and Nf 2.

HOW IS Nf PASSED ON?

The pattern of inheritance for both types of Nf is autosomal dominant. Each and every child of an affected parent has a 50% (1 in 2) chance of inheriting the faulty gene and of having Nf. However, 50% of people with Nf have no family history of the condition. They are probably affected because of a spontaneous genetic mutation which took place in the egg or sperm before fertilisation.

A PERSON WITH Nf1 MAY BE AFFECTED IN SOME, OR ALL, OF THE FOLLOWING WAYS:

- Early signs of Nf1 include six or more coffee-coloured (cafe-au-lait) marks on the skin before a child is five years old, freckling in the armpits and neurofibromas (pronounced new-roe-fye-broe-mers), which are lumps and bumps on or just below the surface of the skin.
- large tumours (plexiform neurofibromas) can form on the skin. These tumours are usually not harmful in themselves but can severely affect a person's appearance, particularly if they are on the face.
- Tumours pressing on the nerves in the eye, the ear or internally on the spine or the brain can affect sight and hearing, speech and even lead to paralysis and early death.
- A person with (Nf1 may have learning difficulties and behavioural problems. An increased risk of epilepsy is also likely.
- High blood pressure is associated with Nf1.
- Some bones may be affected — the spine may be curved (scoliosis) and the long bones below the knee and below the elbow maybe malformed.

OTHER INFORMATION

- Nf1 affects about 1 in every 2,500 people worldwide. There are more than 23,000 people with Nf1 in the UK alone.
- Nf2 is less common, affecting 1 in every 35,000 people worldwide. It nearly always involves hearing loss due to tumours on nerves in both ears. Diagnosis can be difficult, but the effects of the tumours in the body are always serious and may be life-threatening.
- Living with Nf can be hard for people whose appearance is severely affected. Tumours on the skin tend to start to appear around puberty and may increase in number throughout life, so some adults with Nf will have many swellings of all shapes and sizes anywhere on their bodies. Some tumours can be removed surgically, but this leaves scarring and is not practical for all swellings.
- If you are interested in finding out more about Nf, you can write (enclosing an A5 stamped addressed envelope) to: The Neurofibromatosis Association, Quayside House, 38 High street, Kingston-upon-Thames, KT1 1HL.
- Telephone: 020 8439 1234. Fax: 020 8439 1200. Minicom: 020 8481 0492