NEUROFIBROMATOSIS TYPE 1 (NF1)

WHAT IS NEUROFIBROMATOSIS (NF)?

NF is an inherited condition which occurs in two completely separate forms. NF Type 1 (NF1) is the most common, affecting about 1 person in every 4000 in Britain. NF Type 2 is much rarer, affecting about 1 in 35 000 people.

HOW IS NF1 DIAGNOSED?

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

- Flat, coffee-coloured patches on the skin known as ***café-au-lait*** spots. These café-au-lait spots usually begin to appear during the first year of life and gradually grow in size and number. Individuals with NF1 usually have many (at least 6) café-au-lait spots which can vary in size from one to several centimetres in diameter.
- ***Freckling*** in unusual places such as the ***armpit or the groin.***
- Small lumps on the skin known as ***neurofibromas***. They 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 diffuse 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 test is needed to detect these.

In adults the diagnosis of NF1 is normally clear-cut. Initially, children with NF1 may only have café-au-lait spots but other features usually appear over time. From this reason diagnosis cannot be excluded with confidence before 5 years.

NF1 is caused by a change ("mutation") in a gene. In the past, genetic testing for NF1 has not been performed routinely because genes changes have been difficult to detect. The diagnosis is therefore based on clinical examination. However gene testing may now be helpful in certain circumstances.

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HOW IS NF1 INHERITED?

Each child of person with NF1 has a 50:50 (1 in 2) chance of inheriting the condition. Since the symptoms can sometimes be very mild, affected individuals may not realise that they carry the gene change until the diagnosis is made in other family members.

For 25-50% of people with NF1, no signs of the condition are found in either of their parents, making it most likely that the gene change has occurred for the first time in the affected person. In this situation, the unaffected parents have a very low chance of having another child with NF1.

In families where more than one individual is affected, it may sometimes be possible to offer testing in pregnancy. Couples wishing to consider this should be referred to a clinical geneticist before pregnancy.

WHAT MEDICAL PROBLEMS ARE ASSOCIATED WITH NF1?

The majority of people with NF1 do not experience problems as a result of the condition. However, about one third will develop one or more complications during their lifetime. Unfortunately, it is difficult to predict in which patients these problems will occur. Since the complications of NF1 respond best to early treatment, all patients should be seen regularly by a doctor once a year for a check-up.
You should also consult your doctor if you are worried about new or unexplained symptoms.

HIGH BLOOD PRESSURE (HYPERTENSION)

High blood pressure is frequent 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). Both these problems can be treated by an operation.

LEARNING DIFFICULTIES

About one third of children with NF1 require extra help at school (compared to 8-9% of children in the general population). Hyperactivity and clumsiness also seem to be more common in children with NF1. Only a very small number (1-2%) of people with NF1 have severe learning difficulties.

TUMOURS

Tumours growing in and around the brain and spinal cord are an uncommon complication of NF1. They are usually benign (not cancerous) and do not spread to other parts of the body. Tumours of the optic nerve may not cause any symptoms at all but can sometimes cause a squint, double vision or blurred vision. If such a tumour is found, it needs to be monitored by an ophthalmologist (eye doctor), and treatment is sometimes required.

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 require an operation.

A small number of individuals with NF1 will develop malignant (cancerous) tumours. However, it is important to realise that the risk of developing a cancer which is definitely related to NF1 is low- only around 5% (1 chance in 20). If you notice a lump growing rapidly or becoming unusually painful you should consult your doctor.

BONE PROBLEMS

NF1 can sometimes be associated with curvature of the spine (scoliosis) which usually develops in the early teens. Occasionally children are or with, or develop, bowing of the bones in the lower leg.

LOCAL CONTACTS

Your Local Genetics Services:

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