

## **Neurofibromatosis Type 1 - An Information Leaflet for Health Professionals**

### **Introduction**

Neurofibromatosis type 1 (NF1) is a genetic condition that affects about 1 in 4000 people. The characteristic findings of NF1 are café-au-lait spots, neurofibromas and Lisch nodules; other manifestations include optic gliomas, plexiform neurofibromas and learning disabilities. NF1 is sometimes referred to as von Recklinghausen or “peripheral” neurofibromatosis. It is a separate disorder to NF2.

### **Diagnostic criteria for NF1**

NF1 should be considered if 2 or more of the following are present in an individual:

- At least 5 café-au-lait macules >15mm diameter if post pubertal. (May be fewer in children <5yrs)
- 2 or more neurofibromas of any type or 1 plexiform neurofibroma
- Multiple freckles in the axillary or inguinal regions
- Optic nerve glioma
- 2 or more iris Lisch nodules seen on slit lamp
- Sphenoid wing dysplasia or congenital bowing or thinning of long bone cortex with or without pseudarthrosis
- A first degree relative with NF1 by the above criteria
- Genetic testing for NF1 is not always helpful.
- The diagnosis should be considered in individuals who do not quite meet the criteria above and monitoring or further investigations may be appropriate for them

### **Genetics**

Inheritance is autosomal dominant, but about half of cases will be new mutations and so have no family history of NF1. The gene has been found but mutations can only be identified in about half of cases.

### **Major complications of NF1**

Complications of NF1 are numerous. Approximately 30% of people with NF1 will have at least one complication. Serious, or life-threatening, complications affect 5-10% of individuals with NF1. Everyone with NF1 has to be considered at risk of the various complications, although individually they are quite rare. The most common serious complications include:

**Central nervous system:** Learning disabilities, optic gliomas, ,

**Peripheral nervous system:** Peripheral nerve compression, , and large plexiform neurofibromas

**Cardiovascular:** Hypertension (renal artery stenosis/\_phaeochromocytoma/essential)

**Endocrine:** Pheochromocytoma

**Musculoskeletal:** Scoliosis, short stature, congenital bowing or pseudarthrosis of long bones, usually tibia

## **Screening children**

Any child affected with, or possibly affected with NF1 should be carefully examined at least once a year with particular attention paid to the following:

- Blood pressure measurement
- Visual field assessment until 5 years
- Examination of the spine for scoliosis
- Skin examination for plexiform neurofibromas
- Monitoring of school performance (or development if preschool)
- Height and OFC
- Routine brain imaging is not recommended but there should be a low threshold for investigating unusual signs or symptoms

## **Screening adults**

Annual examination is recommended with concentration on the skin, CNS and blood pressure. Any unusual or persistent symptoms should be considered in relation to NF1 and referral to, or discussion with, someone with patient expertise in the condition may be helpful.

Affected adults should be made aware of the 50% chance of passing the condition on to any child they may have, and that a child could have more or less severe problems than themselves.

## **Support group**

There is a family support group for NF called the Neurofibromatosis Association (NfA). It provides a regular newsletter and information leaflets and there are often local groups which meet for support advice and fund-raising. The NfA can be contacted at:

Quayside House  
38 High Street  
Kingston on Thames  
Surrey KT1 1HL  
Tel: 020 8439 1234  
Email: [info@nfauk.org](mailto:info@nfauk.org)  
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