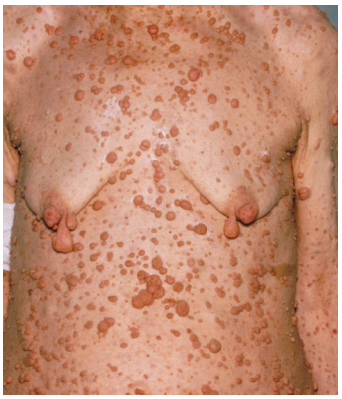

The physical and psychological implications of neurofibromatosis

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ABSTRACT **Tower, R. (2004) The physical and psychological implications of neurofibromatosis. *Nursing Times;* 100: 27, 34–36.**

Neurofibromatosis is a common genetic condition that causes nerves to develop non-malignant swellings (neurofibromas). These can occur on any area of the body, and can result in a wide range of symptoms involving the neurological, cognitive, orthopaedic, renal and endocrine systems. The severity of the condition is highly variable, and an individual approach is necessary to address the specific symptoms experienced by each patient. Nurses can address many aspects of neurofibromatosis including screening, medical complications, learning difficulties, and the impact of disfigurement. With knowledge of the broader implications of a diagnosis nurses can significantly improve patients' experience of the health service and strengthen their coping skills.

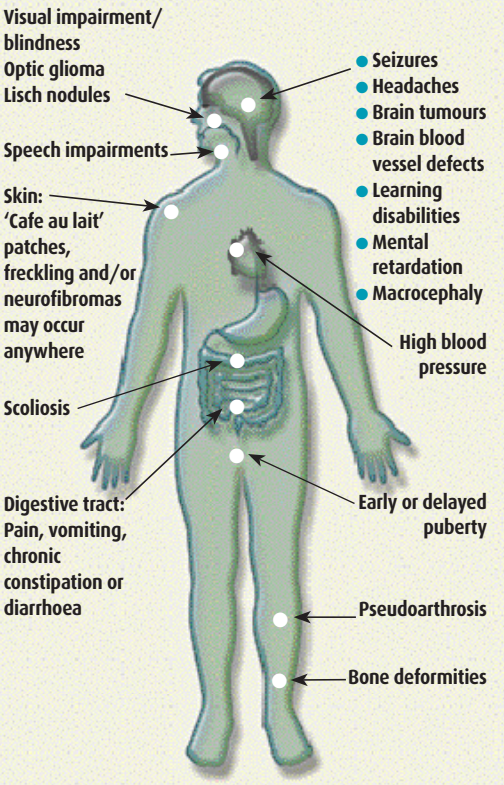
Neurofibromatosis (NF) is one of the most common known genetic disorders, and encompasses at least two diseases: NF1 (previously known as Von-Recklinghausen's disease) and the much rarer NF2 (previously known as central or bilateral acoustic NF). The incidence of NF1 at birth is one in 2,500–3,000 (Huson et al, 1988). It causes nerves in any part of the body to develop non-malignant swellings (neurofibromas), which can appear in any tissue where the affected nerve is lying. NF2, which has a prevalence of one in 33,000–40,000 births (Evans et al, 1992), is characterised by tumours on the eighth cranial nerve and predominantly leads to deafness.

The clinical manifestations of NF1 can vary widely (Fig 1) and depend on where the neurofibromas are located. It can cause numerous social, educational, and medical problems involving the neurological, cognitive, orthopaedic, renal, and endocrine systems. The main symptoms are:

- 'Cafe au lait' patches (macules) and freckles on the skin, as well as small, jelly-like and purplish swellings, which are not usually painful but can itch and catch on clothing and other objects;
- Swellings in major nerves that can cause tingling and other odd sensations if pressure is exerted on the area supplied by the nerve;
- Deafness, tinnitus, and vertigo if the inner ear is affected.

About one-third of people with NF1 have complications such as mild learning disabilities, nerve root compression, intestinal bleeding and obstruction, joint problems and back deformity, and hypertension. Again, complications depend on the location of the neurofibromas.

FIG 1. CLINICAL MANIFESTATIONS OF NF1 (NEUROFIBROMATOSIS INC, 2001)



Fifty per cent of people with NF1 inherit it from a parent, while the others have no family history and are described as new genetic mutations. There is a 50 per cent risk of NF1 being passed from parent to child but there is no clear familial link in the severity of the condition, so a parent can have mild symptoms while a child inheriting it may develop serious complications or vice versa. These inheritance patterns create difficulties when advising prospective parents, as antenatal genetic testing can confirm or exclude the condition but not its severity or likely complications. It can be difficult to advise the parents of children with new genetic mutations, as they have no experience of the condition themselves.

Diagnosis

Diagnosis of NF1 requires two or more of the following (Ferner et al, 2003; Gutmann et al, 1997):

- Six or more 'cafe au lait' macules >0.5cm in children

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and 1.5cm in adults (Fig 2);
- Two or more cutaneous or subcutaneous neurofibromas (Fig 3) or one plexiform neurofibroma (flat, diffuse lesion, Fig 4);
- Axillary or groin freckling (this is an unusual location for freckling);
- Optic pathway glioma (an abnormal thickening of the optic nerve);
- Two or more Lisch nodules (iris lesions);
- Bony dysplasia (sphenoid wing dysplasia causing proptosis, bowing of a long bone);
- A first-degree relative with NF1.

Although diagnosis is usually made on a clinical basis, it may be aided by a number of tests that are used to assess for potential complications (Box 1). Since neurofibromatosis can present in an extremely mild form, it is not uncommon for it to be diagnosed in a parent after their child has been diagnosed.

Neurofibromatosis in childhood

Once a child is diagnosed with NF1, the condition requires careful monitoring for complications. One of the most serious childhood complications, particularly before the age of seven, is an optic pathway glioma (Listernick et al, 1999), which can permanently damage vision. This is usually a unilateral problem although it occasionally affects both eyes.

Parents should be made aware of the need to monitor their child's vision and annual visual acuity checks should be maintained. Any reduction or change in vision should be investigated thoroughly. The decision to treat an optic glioma is based on visual loss and progressive change on MRI scan. In children it usually involves chemotherapy, which may obviate the need for radiotherapy and avoid possible longer-term consequences (Packer et al, 1997).

Monitoring for orthopaedic complications is also necessary as NF1 can cause bony dysplasia such as scoliosis or bowing of the tibia (with associated risk of fracture and non-union – known as pseudoarthrosis). Sphenoid wing dysplasia – a poorly formed or missing bone forming the back of the orbit – may cause bulging or downward displacement of the eye (proptosis). Children therefore require annual assessment of the skeleton and spine and parents should be advised to report any abnormalities.

Developmental delay occurs frequently in people with NF1. For example many children take longer to develop speech and walking, while learning disabilities or cognitive impairment are also common (Ferner et al, 1996). Although mild in most cases, these can result in:
- Immaturity;
- Concentration difficulties;
- Organisational problems;
- Difficulty with reading, writing, and calculating;
- Short-term memory deficit;
- Processing difficulties.

There may be some difficulty with flexibility in thought and action, problem solving, speech organisation, strategic planning and the child's regulation of her or his own behaviour. These difficulties are described as 'executive skill dysfunctions' (Ozonoff, 1999). Social deficits are also possible, leading to poor communication and a limited understanding of danger, while maintaining friendships can be difficult for children.

Gross and fine motor skills can be affected and this can have an impact on the child's ability to manipulate objects such as scissors and pens, and make it difficult to participate in many sports. Occupational therapy and physiotherapy can often be helpful with motor functions.

Short stature in children with NF1 is common, while the onset of puberty can be early or delayed and they may need a referral to an endocrinologist. Blood pressure should be checked at least annually as renal artery stenosis or, rarely, phaeochromocytoma (lesion of the adrenal gland) can result in hypertension. Plexiform neurofibromas in children should also be monitored.

Neurofibromatosis in adulthood

Dermal neurofibromas

The most distressing symptom for adults is the cosmetic impact of dermal neurofibromas (Wolkenstein et al, 2001). Although these grow from peripheral nerves, they are not a direct extension of nerve tissue and are not generally painful. Subdermal neurofibromas are less common and have less cosmetic impact but they can be painful if knocked.

Dermal neurofibromas can first appear as purple

FIGS 2–4. VISIBLE SIGNS OF NEUROFIBROMATOSIS

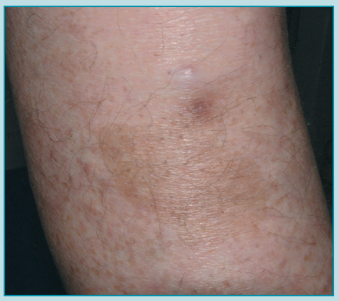

Fig 2. Cafe au lait lesions

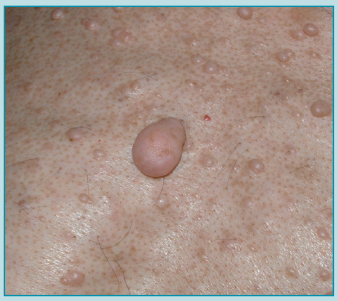

Fig 3. Cutaneous lesions

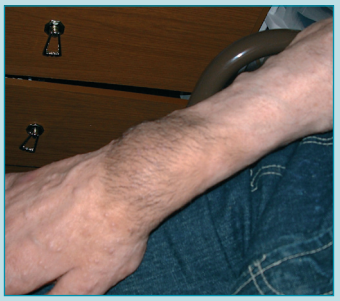

Fig 4. A plexiform neurofibroma

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bruise-like marks under the skin, which in most cases grow into round lumps that are soft to the touch. They often start to appear after puberty, although they can be seen in children. Developing skin abnormalities during adolescence can have a huge psychological impact, regardless of the number of lesions, particularly since people with NF1 experience discrimination because they look different and other people may fear that the lumps indicate a contagious disease. It is vital that cosmetic issues are addressed sensitively.

These neurofibromas grow at varying rates, and while some people develop hundreds over their lifetime, others may develop very few. Neurofibromas often increase in both size and number during pregnancy. While they can cause itching, especially during hot weather, this can be alleviated by the use of emollients but not generally by antihistamines.

Neurofibromas can be removed harmlessly, although there is a risk of scarring. They are generally removed by conventional surgery under local anaesthetic, although laser surgery is sometimes used. However, it is important for individuals to try to develop a degree of acceptance of the lumps, as it is often unrealistic to consider removing all of them. Priority is given to visible lumps, such as those on the hands and face, and those that cause discomfort, for example because they are situated under a bra strap or belt.

Plexiform neurofibromas

Plexiform neurofibromas are far more complex than the dermal variety and are mainly situated upon and under the skin, although they can also occur internally. They are often characterised by a larger 'cafe au lait' mark on the surface of the skin – sometimes with unusual hair growth.

These lesions are difficult to remove as they are highly vascular and cause a thickening of several nerves rather than involving only small peripheral nerves. They may track along nerves for some distance underneath the skin and MRI scanning is necessary to establish the extent of the problem before surgery is considered.

Plexiform neurofibromas can, in rare cases, impinge on the spinal nerve roots or spinal cord, with serious neurological consequences. They can also cause deformity by distorting the skin with significant growth and bony dysplasia. Surgery on plexiform neurofibromas often leads to complications and is only indicated with very careful consideration.

Both adults and children with NF1 are at increased risk of malignancy (Zoller et al, 1997), and rapid growth of any internal lump or change in texture and persistent pain should be investigated thoroughly. Patients should also be advised to report any change in function such as persistent numbness, sphincter disturbance (especially bladder function changes), pins and needles, or weakness.

Management and patient support

The diverse nature of the complications seen in patients with NF1 means that they may be treated by nurses in any specialty. It is important that nurses understand both the psychological and physical effects of the condition, and be sensitive to patients' needs. The condition can have a major impact on patients' lives and they are often extremely vulnerable. Advocacy and sensitive handling of issues related to learning difficulties and cosmetic problems are vital.

BOX 1. DIAGNOSTIC TESTS

- Slit lamp ophthalmic examination (most commonly used test)
- Roentgenography
- Computerised tomography
- Magnetic resonance imaging
- Electroencephalography
- Biopsy and histological examination of cutaneous lesions
- Development and neuropsychiatric testing
- Genetic testing

Acting as a 'key worker' can be a valuable role in managing and effectively supporting people with this complex condition. Nurses need to ensure that patients understand what the lumps are and that they are harmless. Many patients know little about the condition and it can often come as a great relief to them to be able to talk about it and obtain accurate information.

Even if only one member has the condition, NF affects whole families and nurses need to be aware of its impact and offer the support they need. Accurate information on the condition, screening, and management improves patients' and family members' understanding and can help them to cope.

Social workers can offer support with a variety of social issues from housing to work-related problems, as well as advising on how to access services and entitlement to benefit. They may also be able to offer representation and advocacy services in situations where patients struggle to assert their rights.

The Neurofibromatosis Association employs specialist advisers across the UK to offer specific support, particularly to parents. These professionals operate across working boundaries with inpatients, outpatients, schools, social services, housing, and voluntary organisations to help people with this condition improve their situation. For example their role involves direct liaison with schools, providing information and talking to teachers to improve the support children receive if they have associated learning problems. Community support services are crucial to help parents manage difficult behaviour in children.

Conclusion

Many people with NF1 have negative life experiences related to their health and to the reactions of others. They therefore require sensitive support to help them cope with the practical and social implications of their condition. They may also find it helpful to be put in contact with the Neurofibromatosis Association, which produces literature on the condition and can put patients in contact with specialist advisers. ■