



## **Bone Changes in Neurofibromatosis Type 1**

### **Introduction**

A proportion of people with NF1 develop orthopaedic changes affecting bones. Some of these are age-related and may be first identified during early childhood. The orthopaedic (bone) manifestations of NF1 fall into three main categories:

- **scoliosis**
- **congenital (present at birth) pseudarthrosis**
- **disorders of bone growth**

In this information sheet we shall examine each of these problems in turn.

### **SCOLIOSIS**

#### **What is Scoliosis?**

Scoliosis is derived from a Greek word meaning a sideways curvature of the spine. It is present in about 6% of people with NF1 and can be so mild as not to be noticed or severe enough to be obvious and painful. In the most severe form there can be considerable forward bend as well as a sideways component. If this is predominant in the upper part of the spine the word 'kyphosis' is used. In the lower part of the spine the word 'lordosis' is used to describe this forward bend. Lordosis often causes the buttocks to appear unduly prominent.

#### **How common is Scoliosis in NF1?**

The general consensus is that between 5 – 10% of people with NF1 have a detectable scoliosis but this is often mild and does not progress. However sometimes it can be the opposite and so cause problems.

#### **What checks are done for scoliosis?**

It is important that a growing child with NF1 should have their back examined at least annually as part of their regular health check. Your GP or paediatrician can do this.

To check for spinal curvature, the doctor will look along the child's back when they are bending down towards their toes with their knees straight. If there is an obvious difference on the two sides or any sharp angulation of the spine the doctor may decide to do an X-ray. If, as a parent, you notice this sort of change in your child's back you should seek the advice of your doctor. It is a relatively easy check to do, say every six months or so, in between appointments. Once your child reaches the age of 16 or 17 years, if their back is straight then it will remain so. Early detection is preferable as it can result in easier and therefore better treatment if that should be necessary.

#### **When do spinal curvatures first appear?**

The most severe spinal curvatures will appear during the first few years of life,

typically before the age of 5. As most spinal curvatures are disorders of growth, they will tend to become more severe if there is a long period of growth ahead. The less severe types tend to appear around the age of puberty, at which time they might progress rapidly during the fast growth of adolescence. On the other hand, curvatures which are only just visible to the naked eye may not worsen.

If by the end of adolescent growth, at around the age of 17 or 18, the curvature is only just visible to the naked eye, it will probably not worsen later in life.

### **Will scoliosis become painful or disabling?**

Pain and disability are caused by mechanical failures of the spine. In scoliosis this will occur if the curvature increases over 40-60% of angulation (off the vertical) in adolescence or in later life when the normal stresses and strains produce increasing wear and tear over the curved part of the spine. Pain and disability are very variable in different individuals. For example, an individual might have a severe curve with only minor pain whilst someone with a relatively minor curve could experience more severe pain and disability.

### **How should spinal curvatures be treated?**

This is a large and complex subject and impossible to cover here in detail. The most important step is to consult your GP, Specialist or Nerve Tumours UK. If it is felt that there is a spinal curvature you will be referred to an orthopaedic surgeon with special interest and expertise in this field. Virtually all regions of the country now have such a surgeon but your own doctor will be able to give you more specific information.

Treatment of spinal curvatures usually consists of monitoring the curvature with X-rays every few months to assess its behaviour. If it is mild and static then nothing else needs to be done. If it is worsening, then your surgeon may prescribe a brace or recommend surgery to stabilise the deformity before it deteriorates further. There is no doubt that surgery is a major procedure but currently with much safer and more effective techniques, surgery can be advised earlier. It should be remembered that everyone with NF1 is different and the treatment and management of each individual will vary. The timing and type of treatment suggested is a matter of careful judgement and will only be considered after full discussion between the surgeon, yourself and your relatives. Fortunately, treatment is slowly improving all the time.

### **Is there a risk of paralysis?**

With early detection of deformities and timely skilled surgery this is exceedingly rare.

### **Summary**

The most important priority is to locate the relevant specialist in your area who will provide you with the best possible treatment. Your GP or Nerve Tumours UK will be able to help and refer appropriately. NF1 is a comparatively common condition but the orthopaedic problems are rare and the pooling of experience is crucial. Each region will differ in the services they provide e.g. rehabilitation, physiotherapy and occupational therapy. Other support organisations can also be extremely helpful (see details at the end of this information sheet).

## **CONGENITAL PSEUDARTHROSIS (now called dysplasia unless the bone is fractured at diagnosis)**

### **What is congenital pseudarthrosis?**

A pseudarthrosis is a 'false joint' and occurs when a bone fracture fails to unite and remains mobile at the fracture site. Congenital pseudarthrosis is comparatively rare and affects about 3-5% of people with NF1. (1)

### **When and where does it occur?**

Pseudarthrosis can be congenital (from birth), or more frequently, the bone involved is noted to bend and then fracture in the first few years of life. The weakness in the bone is caused by the presence of neurofibromatosis tissue around the bone, which causes it to deteriorate.

Pseudarthrosis occurs most frequently in the tibia (shin bone), radius or ulna (forearm) but can also occur in any of the long bones: femur (thigh), clavicle (collar bone), humerus (arm), first rib or fibula (leg).

### **Diagnosis**

Ideally pseudarthrosis is diagnosed whilst the bone is bent and before fracture has occurred. Early diagnosis means that precautions to prevent further bending and fracture are the best treatment.

### **Treatment**

Once a fracture has occurred treatment can be very difficult and it is a challenge to get the bones to unite. The best chance of uniting the bones is achieved by using techniques such as an external fixator to stabilise and compress the fractured bone, or the fibula is used as a vascularised bone graft. There is around an 80% success rate being achieved in the tibia. (2)

Surgical treatment usually begins when children are between 2 and 4 years old. Unfortunately, failures do occur and some children undergo many attempts at trying to achieve union. Should union of the bone not be achieved, then either long term orthotic stabilisation is required or some children may require amputation, particularly where there is pseudarthrosis of the tibia.

### **Summary**

The most important priority is to locate the relevant specialist in your area who will provide you with the best possible treatment. The National Complex NF1 services in London and Manchester or Nerve Tumours UK will be able to help. In the event of difficulty identifying a specialist in this highly specialist area then the NCG funded service for complex NF1 is available for advice (see details at the end of this information sheet).

Although NF1 is a comparatively common condition the orthopaedic manifestations are rare and so the pooling of experience is crucial to a positive outcome.

## **DISORDERS OF BONE GROWTH**

### **How to recognise disorders of bone growth**

NF1 is known to be a significant cause of disorders of bone growth. There might be mal-development of the bones in the skull or congenital (present at birth) sphenoid wing dysplasia; this can cause prominence of one or both eyes. Overgrowth of other bones may occur occasionally.

### **Chest wall changes**

A proportion of children with NF1 have a change in the shape of their chest wall. They may have a chest that has a hollowed out appearance (pectus excavatum) or a

prominent breastbone (pectus carinatum or "pigeon" chest).

This problem is caused when the tough connective tissue that holds the ribs to the breastbone develops unusually causing changes to the normal shape of the chest. If the cartilage flares out away from the chest wall it causes the chest to stick out; if the opposite occurs and the cartilage grows inwards, it causes the chest to grow inwards pushing the breastbone in. The problem can occur on both sides of the chest wall in which case the changes are symmetrical or on one side only giving rise to an asymmetrical shape.

These changes in the shape of the chest wall rarely need any surgical treatment. However they can be a source of cosmetic concern for patients.

### **What treatment is available?**

The bone problems we have outlined can often be helped or eliminated by timely surgery. Special shoes, braces and callipers might be necessary for any of the orthopaedic problems of NF1. These devices are being constantly improved and are often not nearly as bad as they sound.

Occasionally limb length inequality can result in asymmetrical overgrowth of a leg so that one leg is noticeably thicker and longer than the other. Leg length discrepancy can be treated with an operation called epiphysiodesis. This is a surgical procedure whereby the surgeon inserts an inhibitor across the growth plate of the affected bone to stop further bone growth, effectively allowing the shorter leg to catch up. The skill in this procedure is in knowing at what stage it should be carried out so that both legs grow to achieve equal length.

### **Vitamin D and NF1**

Research has shown that some people with a diagnosis of NF1 can suffer from vitamin D deficiency and have decreased bone density and reduced muscle strength. Vitamin D is a fat soluble vitamin that is needed to maintain normal blood levels of calcium and phosphorous in the body. These minerals are needed for the normal process of bone mineralisation (a process essential for the hardness and strength of the bone itself). Crystals of calcium phosphate are produced by bone forming cells and are laid down in specific amounts in the bone's fibrous structure. Problems can occur if the amounts are incorrectly regulated so the bone may become too soft (osteomalacia).

Vitamin D is naturally present in only a few foods including oily fish and dairy products. However it can be synthesised naturally by exposure to sunlight. People with dark skin pigmentation, those who cover their skin or people who rarely expose their skin outdoors can be at risk of vitamin D deficiency. It is important to protect the skin from excessive sun exposure with sun creams.

Research in this area is ongoing.

### **Osteoporosis**

Osteoporosis is a condition that affects the strength of the bone. The bone becomes less dense and therefore more likely to break. Osteoporosis mainly affects older people (both men and women) but research has shown that it occurs more frequently in people who have NF1. Your GP can advise you about the risk factors and preventative strategies that should be beneficial.

Finally there is a **nationally commissioned service for complex NF1** in England

(patients from elsewhere are seen but funding must be agreed beforehand) The two major Neurofibromatosis centres are located at Guy's Hospital, London and St Mary's Hospital, Manchester. In the event of difficulty identifying a regional specialist, both centres are able to offer advice and expertise. This is a nationally funded service that reviews, assesses and co-ordinates the care of patients in England whose needs are complex and require management by a team of clinicians with expertise in NF1.

### **Useful websites:**

[www.changingfaces.org.uk](http://www.changingfaces.org.uk), [www.sauk.uk](http://www.sauk.uk) [www.strongbones.org.uk](http://www.strongbones.org.uk)

### **References**

1. Huson et al 1988; Ferner 2007 Stevenson et al.1999
2. Journal of Paediatric Orthopaedic Surgery; 2009; 9, 67-74
3. Instructional review: Children's Orthopaedics August 2013. Controversies in the management of congenital pseudarthrosis of the tibia and fibula. T Khan, B Joseph. Bone joint J. August 2013 95-B: 1027-1034

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**Please seek further information from [www.nervetumours.org.uk](http://www.nervetumours.org.uk), or contact our helpline on the details above.**

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