



Introduction

The orthopaedic manifestations of Neurofibromatosis Type 1 (NF1) fall into three main categories:

- congenital pseudarthrosis,
- disorders of bone growth/soft tissue
- scoliosis

In this fact sheet we will try to answer the most frequently asked questions about congenital (which means present at birth) pseudarthrosis.

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% of people with Nf1. (1)

When and where does it occur?

Pseudarthrosis can be congenital, 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 (hip), 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 is usually commenced 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 and your GP will be able to help.

Nf1 is a comparatively common condition but the orthopaedic manifestations 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. Please speak to your Specialist Advisor for more information about your local area.

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References

- 1) Huson et al 1988; Riccardi, 1992; orth 1993
- 2) Journal of Paediatric Orthopaedic Surgery; 2000; 9, 69-74

Produced
with financial
support from



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Updated 07/09