



Introduction

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

- disorders of bone growth and soft tissue changes
- scoliosis
- congenital pseudarthrosis

In this fact sheet we will try to answer the most frequently asked questions about disorders of bone growth and soft tissue.

How to recognise disorders of bone growth and soft tissue changes

NF1 is known to be a significant cause of disorders of bone growth and soft tissue change and any bone or soft tissue can be affected. Overgrowth of facial bones is rare but can occur and also involve surrounding tissue. 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.

Soft tissue overgrowths can occur anywhere in the body, e.g. soles of the feet, over the buttocks or on the face; these can be unsightly but can often be easily removed. Tumours on the nerves and in critical places where they pass through bony tunnels may cause erosion and weakening of the surrounding bone. Occasionally this leads to vertebral (spinal) collapse and deformity. Pressure on the nerve tissue can itself be the most important effect. This may lead to nerve deficits or weakness anywhere in the body and depending on the nerve involved, can result in nerve deafness or localised numbness and weakness.

What treatment is available?

These problems can often be helped or eliminated by timely neurosurgery. 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.

Living with a face or body that is visibly different is not always easy; anyone who has a condition which affects their appearance can contact Changing Faces for further information and practical help, see details overleaf.

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.

Helpful Organisations

Changing Faces

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London W2 1PN

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For more information and a full list of publications please contact:

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