

## NEUROFIBROMATOSIS AND MULTIPLE SKULL DEFECTS: A CASE REPORT

Kader Naina Mohamed

### SYNOPSIS

**A young girl presented with plexiform von Recklinghausen's disease. Skeletal survey was done to detect osseous anomalies. As expected in this patient, multiple defects were discovered in the skull.**

### INTRODUCTION

Multiple neurofibromatosis was described as a distinct nosologic entity by von Recklinghausen in 1882. The cause of this disease is owing to the meso-ectodermal maldevelopment. However, the pathogenesis is not well understood. Cellular derivatives of neural crest undergo excessive multiplication producing various manifestations in the nervous system and skin. Endocrine organs and bones are other systems involved. (1) It is inherited as an autosomal dominant in 50% of the patients and the remaining half from genetic mutation. The prevalence is 1 in 3,000 of the population (2) and males are more affected than the females. Additional dermatological features of this disorder found in recent literature include blue-red pseudo arthropic macules (3) and melanotic macules of the palms. (4) Bony abnormalities of the skull with greater involvement of the orbital cavity have been recorded. (5) Defects in the skull are probably rare. However, in 1961, Hunt and Pugh have reported this type of skeletal involvement in neurofibromatosis. (6) The present paper describes a patient with three large defects in the occipital bone.

Department of Dermatology  
Sultanah Aminah General Hospital  
80100 Johor Bahru  
Malaysia

K N Mohamed, MBBS (Madras),  
Dip Derm (Lond),  
Dip Ven (Lond)

## CASEREPORT

A 17-year old Malay girl presented with plexiform von Recklinghausen's disease. The onset was insidious beginning at the age of 6 when the parents noticed soft tumours coursing as overgrowths along the right cervical region and spreading to the upper chest, shoulder and the face, causing disfigurement (Fig. 1 & 2). The father has multiple pedunculated



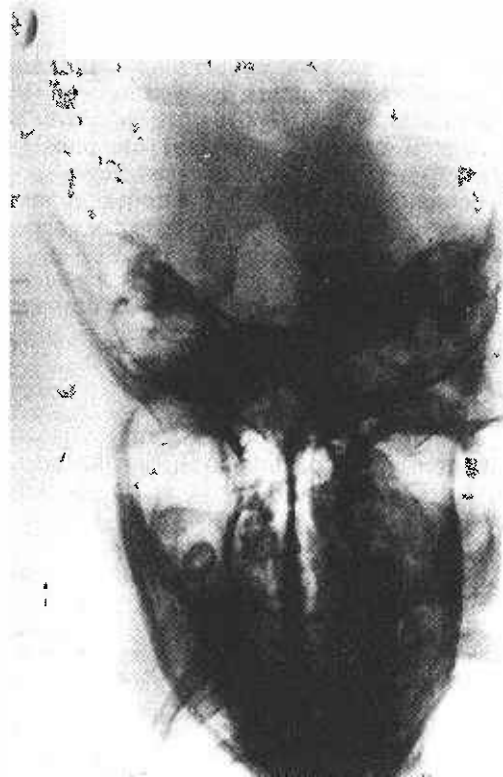
**Figure 1** Plexiform neurofibromatosis and asymmetry of the face.



**Figure 2** Posterior extension of the tumour and laxity of the skin over the occipital region.

neurofibromas. She exhibited normal intelligence and did not attain menarche. There were multiple cafe-au-lait spots predominantly scattered over the trunk and the axillae. There was asymmetry of the face noticed since childhood but no evidence of muscle weakness. Her blood pressure was 100/80 mm. Hearing was slightly diminished on the right ear since the growth was approaching the external auditory meatus. Acuity of vision was: RE 2/12, LE 6/6. Pupils were equal and reacting to light. Iris nodules were not seen and the fundus was normal. Cranial nerve functions were clinically nor-

mal. Palpation of the skull revealed soft depressions on either side of the occipital bone. Complete radiological survey was done. In the skull at least three irregular, large defects were present in the occipital bone (Fig. 3).



**Figure 3** Towne's view of the skull showing the three large defects.

The clinoid, processes, the sphenoid, the petrous and the orbits were normal. Cervical, thoracic and lumbar spines, chest, pelvis and the bones of the upper and lower limbs showed no abnormalities. There was also no calcification in skull x-ray. Computed tomographic (CT) scan of the brain was done and there was no abnormality. The orbits, sphenoid and the petrous were serially scanned but owing to the asymmetry of the skull, interpretation was very difficult. The parents did not respond to the call for consideration of plastic surgery of the neurofibroma or for the investigations of her amenorrhoea.

## DISCUSSION

To the best knowledge of the author, this is the first case report of skull defects in neurofibromatosis from this region. Among the osseous manifestations of this genodermatosis, scoliosis is the commonest feature. One is the idiopathic type mostly seen in girls and the other is sharp, short angular scoliosis frequently occurring in the thoracic region with rapid progression and carries a bad prognosis (7). Tumours which arise proximally at the intercostal nerve may progress to occupy partly in the spinal canal and partly in the intervertebral foramen-dumb-bell tumours, causing pressure symptoms. Small intercostal nerve tumours may cause notching of the inferior surface of the ribs. The other noteworthy changes in the bones are erosions, subperiosteal bone cysts, pseudoarthrosis of tibia which may be easily fractured in childhood and pose difficulty in treatment, increase or decrease of long bones, asymmetry of axial skeleton and spina bifida (8).

Among the skull bones, orbital changes have been highly prevalent. These include defective ossification of the posterior wall of the orbit leading to enlarged sella turcica, widened outline or empty appearance of the orbit, orbital haemangiomas presenting as pulsating exophthalmos, enlarged optic foramen and hypertelorism (5). Congenital absence of the sphenoid and dysplasia or hypoplasia of the maxilla, mandible and temporal bones have been recorded.

Non-interference is the safe guideline for cutaneous tumours. However, surgical intervention in this genodermatosis may be required to improve the cosmetic disfigurement in cases of plexiform neurofibroma appearing on the face where post-operative complications should be borne in mind, tumours showing tendency for malignancy, and to relieve pressure symptoms.

The subject of this report clinically presented with skin and occipital abnormalities and was unresponsive to investigations of her delayed puberty.

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