

**NEUROFIBROMATOSIS PRESENTING WITH A LARGE  
MASS OVERLYING THE SACRUM***A CASE FROM THE ARCHIVE OF A RETIRED ORTHOPEDIC SURGEON***Hassan K Mohammed**

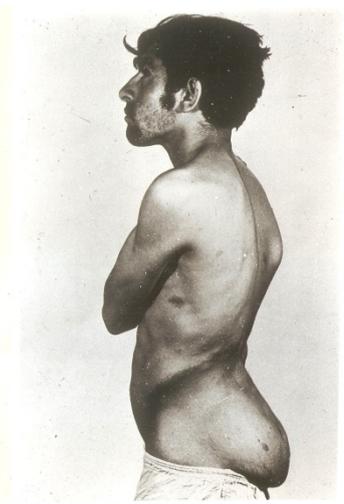
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**Introduction**

Neurofibromatosis is well recognized for its variable manifestations which include features such as café-au-lait pigmentation, cutaneous and deep fibromata, pseudarthrosis of long bones particularly the tibia, and scoliosis<sup>1</sup>. Local hypertrophy of soft tissues may also occur and may be associated with megalodactyly or gigantism of an entire limb<sup>2</sup>. Here is a report of a case of neurofibromatosis with local hypertrophy of soft tissues in an unusual site. This case was presented to and managed in the Department of Trauma and Orthopedics at Basrah General Hospital in 1977.

**Case history**

An 18 year old laborer presented with a large swelling overlying the sacrum that has been present since childhood. It gradually increased in size to the point where it became a major source of embarrassment due to its size and appearance. It also caused significant discomfort as it moved from side to side on walking and made sitting and lying flat difficult and uncomfortable. Figures (I a & b)

**Figure (Ia)****Figure (Ib)**

Seven years earlier a biopsy was taken but unfortunately no results were available. On examination, he appeared to be a fit young man with widespread areas of café au lait pigmentations and multiple skin nodules, the largest of these nodules measuring 3x2 cm in the perineum. The mass overlying the sacrum measured 20x25 cm, it felt firm, subcutaneous and was freely mobile over the underlying sacrum. In prone position, it spread over the lumbosacral area like a jellyfish. When erect, it hangs like a pedunculated mass. There were no neurological changes involving the lumbar or sacral nerve roots. Radiography of his pelvis showed spina bifida of the first sacral vertebra with acute forward angulation at L3-4 level, bone hypertrophy on the posterior aspect of the sacrum with an elongated coccyx was also noted. As CT and magnetic resonance imaging were not available in our unit in 1977, a lumbosacral myelography was done. The myelogram, demonstrated a saccular enlargement of the distal subarachnoid space with no obstruction to the flow of contrast. Figures (II a & b)

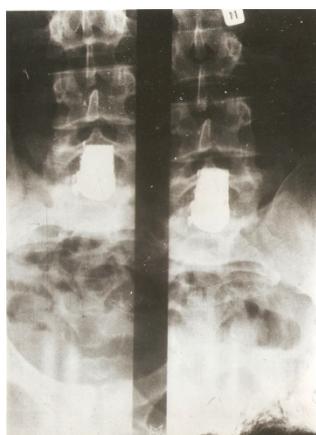


Figure (IIa)

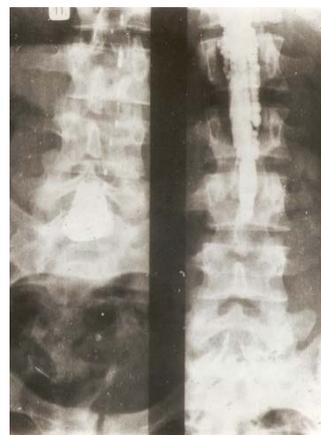


Figure (IIb)

### Operative procedure

The mass was removed via a transverse elliptical incision. It was very vascular and adherent to the underlying lumbar fascia from which it was separated by sharp and blunt dissection. No communication with bone or dura was found. The wound was primarily closed over a vacuum drain and subsequently healed with no complications. Figures (III a & b)

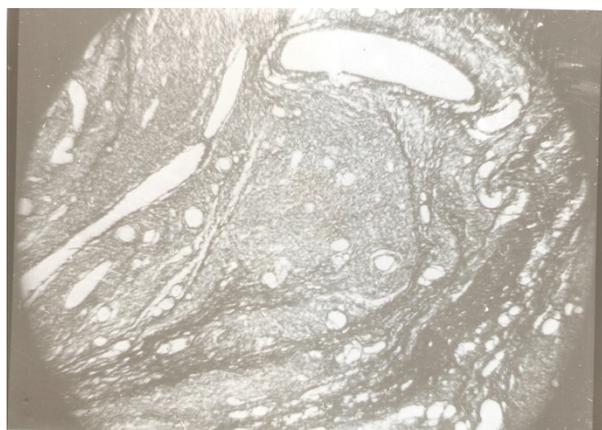


Figure (IIIa)



Figure (IIIb)

The mass weighed one kilogram. Histopathological assessment of the mass showed an abundance of blood vessels in a fibro-fatty matrix with numerous nerve fibers identified by special staining, an appearance consistent with the diagnosis of neurofibromatosis. Figure (IV)



**Figure (IV)**

Clinical examination of the patient's father revealed numerous café au lait pigmentations with multiple skin nodules.

## Discussion

Neurofibromatosis as a disease was described in 1982 by Fredrich Daniel Von Recklinghausen<sup>1</sup>. There are three major distinct forms of neurofibromatosis: type 1, 2 and schwannomatosis. Type 1 being the commonest and also known as Von Recklinghausen disease, is inherited as an autosomal dominant genetic disorder with an incidence of around 1:3000. It affects all races with males and females equally affected<sup>2</sup>. Half the cases present with a family history while the other half present as a de-novo mutation. The gene responsible for neurofibromatosis type 1 (NF1) is located on chromosome 17 and encodes the protein neurofibomin. Loss of production or reduced function of this protein leads to the wide spectrum of clinical findings associated with NF1.

The skin manifestations of NF1 are characterized by the presence of neurofibromas and café au lait pigmentations. These pigmented skin lesions often appear in the first year of life and usually increase in number through childhood. Neurofibromas can be broadly classified into four types: superficial cutaneous neurofibromas and subcutaneous neurofibromas, both benign with no increased risk of malignant transformation, nodular neurofibromas and diffuse plexiform neurofibromas, both of which can transform into malignant peripheral nerve sheath tumors. Plexiform neurofibromas can be a major cause of disfigurement secondary to overgrowth of skin and soft tissue leading to localized gigantism as seen in our case. The excised mass was highly vascular and was associated with enlargement of the sacrum and coccyx with expansion of the subarachnoid space.

## References

1. Antonio JR, Golloni-Bertollo, Tridico LA: Neurofibromatosis: chronological history and current issue. *An Bras Dermatol.* 2013; 88(3): 329-43.
2. Korf BR: Neurofibromatosis type 1 (NF1): Pathogenesis, clinical features and diagnosis. Update [http://www.uptodate.com/contents/neurofibromatosis-type-1-nf1-pathogenesis-clinical-features-and-diagnosis?search=von%20recklinghausen&source=search\\_result&selectedTitle=1~131&usage\\_type=default&display\\_rank=1#H20885999](http://www.uptodate.com/contents/neurofibromatosis-type-1-nf1-pathogenesis-clinical-features-and-diagnosis?search=von%20recklinghausen&source=search_result&selectedTitle=1~131&usage_type=default&display_rank=1#H20885999).