

UNCOMMON PRESENTATION OF NEUROFIBROMATOSIS FOCAL GIGANTISM IN THE UPPER EXTREMITY

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SUMMARY:

Neurofibromatosis with the uncommon presentation of focal gigantism of the left forearm in a 15 year old school boy is presented. There was no evidence of the disease elsewhere in the body.

The radiographic features of a huge lobulated soft tissue mass and the markedly thinned over tubulated and bowed bones of the forearm with cortical defects were suggestive of the disease and this was confirmed on histology. Angiographic features however raised suspicion of a sarcomatous transformation. Extensive surgical excision produced a good result with restoration of function.

The clinical and radiological features are discussed.

INTRODUCTION:

Neurofibromatosis (NF) is an inherited autosomal dominant disease^{1,2}. The incidence of the disease among Caucasians is 1:2000 3000 births with equal male to female ratio³. In our environment the incidence and sex ratio of the disease is not yet determined. Two types of the disease are known and they exist together. Type I (NF1) which has an incidence of 1:4000 live births is otherwise known as Von Recklinghausen's disease or peripheral neurofibromatosis. This type is characterized by multiple cutaneous and subcutaneous nodules (café au lait spots) which increase in number with age and are situated along dermal nerves and the neural plexus branches. DNA studies have shown the causative gene deviation to be mapped on chromosome 17⁴.

Type 2 (NF2) with the incidence of 1:50,000 is otherwise known as central neurofibromatosis with diagnostic criteria including acoustic neuroma, or any two of the following; neurofibroma, meningioma, glioma or schwannoma. Genetic studies revealed a gene deviation located on chromosome 22⁴.

Fredrick Von Recklinghausen⁵ was the first to describe in details, generalised neurofibromatosis. The literature on neurofibromatosis is extensive for the Caucasian population.

For the black population especially in Africa only a few cases have been studied with much emphasis on clinical presentation and management^{6,7,8}.

In this case a peculiar presentation of focal gigantism in the upper extremity which is an uncommon sight for the disease^{8,9} is discussed.

CASE REPORT:

EU, a 15 year old school boy, presented at the UNTH surgical unit with a soft tissue mass in the proximal two-thirds of the left forearm. The mass overlaid the elbow joint and slowly increased in size over a period of three years. Scarification marks inflicted by the herbalist were evident on the mass. There was no history of antecedent trauma. The mass was painless and later grew to become painful with difficulty in the use of the limb.

Past medical and family history were unremarkable. General physical examination revealed a soft lobulated mass, overlying the left elbow joint and extending posteriorly along the proximal two-thirds of the forearm, restricting full extension of the arm. The mass was

slightly relatively warmer to touch.. Auscultation findings were equivocal. The rest of the systems and laboratory findings were essentially normal.

A working impression of arterio-venous malformation (AVM) was made with plexiform neurofibromatosis and haemangiopericytoma as differentials in that order.

Plain radiographic findings were a huge lobulated soft tissue mass overlying the left elbow joint and the proximal two-thirds of the forearm. The ulna and the radius were markedly thinned, overtubulated and bowed laterally with the radius more thinned out than the ulna. Erosive cortical defects were present on the medial margins of the bones (Fig I). Angiography of the brachial artery demonstrated the ulna and radial arteries. There was increased vascularity of the lesion with large tortuous and pathologically looking vessels (Fig. 2 & 3). Ultrasonography and CT. were not done.

Histology report of the specimen taken at biopsy revealed numerous fibroblast cells with dense regions of collagen. Degenerative changes with vascular features and fat cells were seen. All these were consistent with neurofibromatosis.

Extensive surgical excision of the mass was done. The patient made good progress and was discharged home to be followed up at the surgical our patient (SOP). One year after there was no evidence of recurrence and there was good restoration of the use of the arm.

DISCUSSION:

Neurofibromatosis has a familial incidence with equal sex distribution. Sometimes the disease occurs without a family history³ as in the case reported and can arise following a spontaneous gene mutation. The characteristics of the disease include café-au-lait-spots (seen in 95% of cases³). The disease may sometimes be associated with sharp angle kyphoscoliosis, macrocephaly bare orbit, widening of the sella turcica and vessel naevi. The diagnostic criteria is that at least two of the following lesions must be present:-

- café-au-lait spots
- two neurofibromas of any type

discrete rounded soft tissue mass or plexiform neurofibroma which is a tortuous tangle of fusiform enlargement of peripheral nerves.

- Distinctive osseous lesions e.g long bone cortical thinning with or without pseudoarthrosis
- First degree relative (parent, sibling or child) with peripheral neurofibromatosis.

In our patient the distinctive feature of a plexiform lesion and the osseous changes of cortical thinning and overtubulation were present. A peculiar manifestation of localized enlargement of a part of or of one extremity focal gigantism with bone thinning was seen. This lesion is most often seen in the lower extremities but very uncommon in the upper limbs^{8,9}.

The radiographic features seen when extremities are involved include soft tissue masses which may produce focal gigantism, bone over growth, over tubulation, antero lateral bowing of the lower half of the tibia (most common) or the fibula (frequently involved) and the upper extremities (very uncommon)^{8,9}.

In our patient, there was no family history of neurofibromatosis or any other characteristic lesion elsewhere in the body except for the focal enlargement of the left forearm which also is an uncommon site for the lesion^{8,9}. In the patient however most of the radiological features of soft tissue mass and the osseous changes were present.

The angiographic findings in this case were very informative to the extent that certain pathological conditions like AV-malformation; A-V fistula and haemangiopericytoma have to be considered as differentials. In AV fistulae or AV malformation, at angiography the contrast material passing through the fistula reaches the communicating veins. These abnormal veins are filled early in the examination and are usually grossly dilated and tortuous. Soft tissue mass, cortical bone defects and increased bone density with increased bone growth are also seen.

Phleboliths are often present and the most frequent site of location is in the lower extremities. Majority of AV fistulae are usually sequelae of trauma. In the case of our patient, there was no

antecedent trauma. This coupled with upper extremity location of the lesion and the presence of overtubulation of the bones all made the differential diagnosis of AV fistula or malformation very unlikely.

Haemangiopericytoma of the bone could be a considered differential in view of the angiographic features of dense tumour staining, cockscrew vessels, and vascular displacements. The lesion is usually located in the lower extremities and the age incidence is the fourth and fifth decades¹⁰. Our patient was only 15 years old and the site of the lesion was at typically located in the upper extremity hence the differential of haemangiopericytoma was unlikely.

Fibrosarcomatous transformation has been reported in 10% of cases of neurofibromatosis⁹. In such cases angiography may show pathological circulation, vascular encasement and beading, arteriovenous shunts and tumour staining. Some of these features were present in our patient and this raised the possibility of a sarcomatous change. The histology of the specimen from the lesion revealed numerous proliferative fibroblasts, vascular changes and dense areas of collagen and degenerative changes. These seriously suggest malignant transformation which could be consistent with the complex angiographic

findings.

The treatment of neurofibromatosis depends on the type and the severity. When the condition either involves vital structures or constitutes a functional obstacle by rapid growth or excruciating pain like in our patient, extensive surgical excision is indicated. This approach would give a good result and make for a good functional restoration all of which were achieved in our patient.

In lucky cases if excision is complete recurrence is uncommon^{6,7}.

Fig. 1:

Plain radiograph of the left forearm. This shows the large lobulated soft tissue mass in the proximal 2/3 (an unusual site for neurofibromatosis). And radius are thinned out overtubulation (ulna > radius). Note also the marked bowing of the bones and the erosive cortical defects on the medial margins, giving them a wavy and undulation appearance.

Fig. 2:

Angiogram of the brachial artery. The early phase here demonstrates the ulna and radial arteries with early filling of abnormally looking vessels.

Fig. 3:

This phase still demonstrates the arteries and an increase in the vascularity of the lesion with large tortuous and pathologically looking vessels.



Fig 1



Fig 2



Fig 3

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