Case Report

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Surgery Section

Giant Plexiform Neurofibromatosis of the Thoracolumbar Region: A Case Report

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ABSTRACT

Neurofibromas are tumours arising from connective tissue of the nerve. Cutaneous manifestations are generally seen in Neurofibromatosis Type 1 (NF1). Whereas, Neurofibromatosis Type 2 (NF2) has CNS manifestations. Plexiform variety is notorious because of the extreme disfigurement that it produces. Plexiform neurofibromatosis cases should be treated with early excision before they become massive and before malignant transformation. This condition is common in the western countries, but in the Indian subcontinent it is rarely seen.

Here we are presenting a case of a 25 years old Indian male with a large plexiform neurofibroma involving the right chest and right lumbar regions with severe kyphoscoliosis. The patient was treated with surgical resection with primary closure. He had a good recovery.

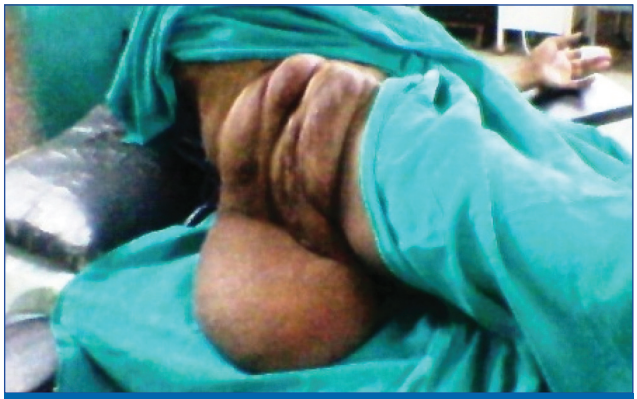
Keywords: Kyphoscoliosis, Pendulous mass, Von Recklinghausen's disease

CASE REPORT

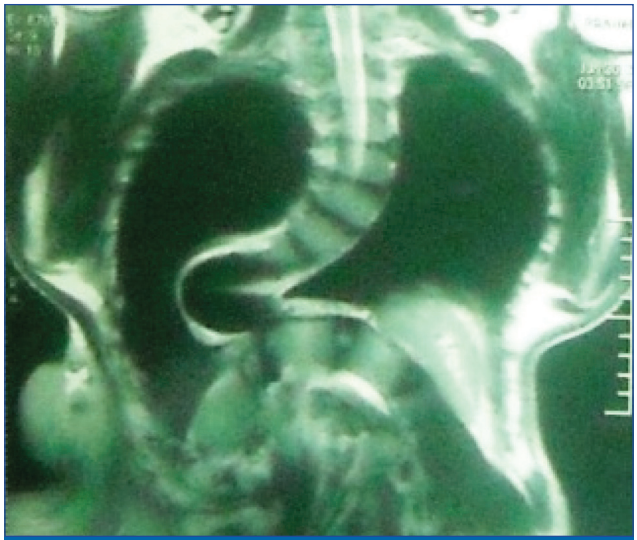
A 25-year-old male reported to us with presenting complain of a large pendulous mass arising from the right chest and lumbar regions. The patient gave consent to undergo surgery and also to use his image for academic purpose [Table/Fig-1]. The patient first noticed this swelling when he was a small child of five years age. Initially, it was small but enlarged progressively and reached the present size. Apart from disfigurement and embarrassment associated with swelling, there was no complaining of pain. There was no lump in any other part of the body. There was no family history of any similar complain.

On examination, a pendulous mass over right side of chest and lumbar region, extending from lower rib cage to iliac crest was noted. The mass had hyper pigmented patches and multiple nodules were palpated in the mass. The size of the mass was about 25X15 cm. It was not reducible and there was no cough impulse. Marked kyphoscoliosis of the thoracolumbar spine was also noted. Chest X-ray confirmed kyphoscoliosis. Magnetic Resonance Imaging (MRI) of the chest and abdomen showed the mass lying above the muscle plain with marked vascularity within the lesion [Table/Fig-2]. All other investigations were within normal limits.

Surgery was planned, and the mass above the muscle plain was resected. Primary closure of the wound was done with

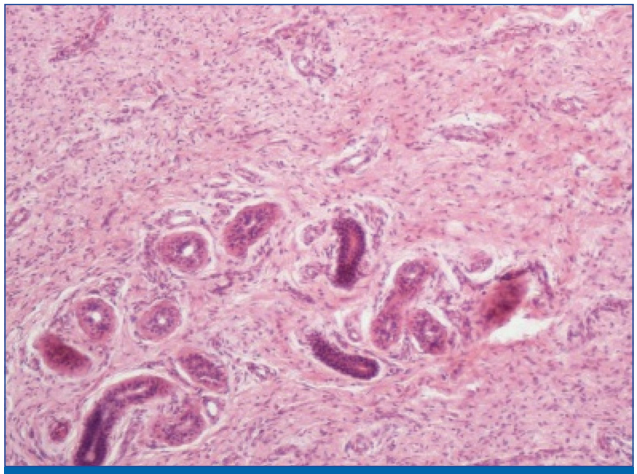


[Table/Fig-1]: Giant lumbar plexiform neurofibroma.

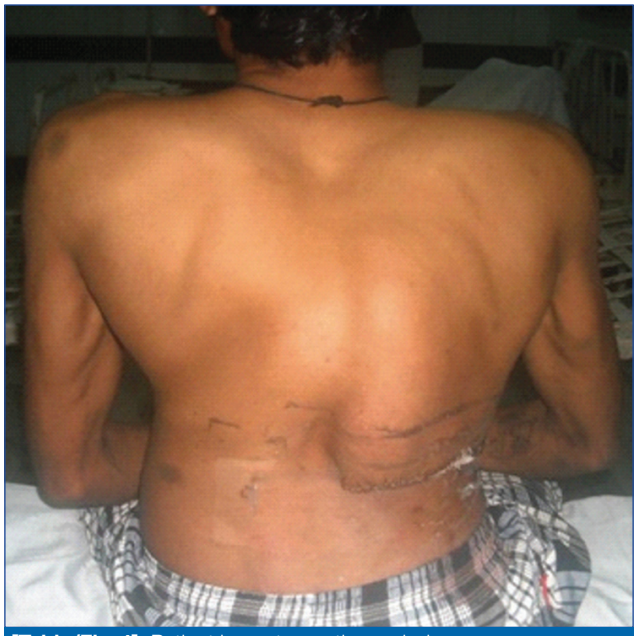


[Table/Fig-2]: MRI showing mass and kyphoscoliosis of spine.

suction drain. The excised tissue weighed about 4.8 Kg. Histopathological examination confirmed the diagnosis of neurofibromatosis [Table/Fig-3]. Patient complained of weakness in lower limbs initially, physiotherapy was started and he showed good recovery. Patient is now on regular follow-up [Table/Fig-4].



[Table/Fig-3]: Histopathological examination of excised mass.



[Table/Fig-4]: Patient in postoperative period.

DISCUSSION

Neurofibroma is a cutaneous manifestation seen in NF 1 [1]. These are tumours arising from connective tissue of the nerve. NF1 is also known as Von Recklinghausen's disease. It is autosomal dominant with an incidence of 1 in 4000. Male to female ratio is equal [1]. NF1 is caused by mutation of a gene on the long arm of chromosome 17 which encodes a protein called neurofibromin.

Specific criteria for the diagnosis of Neurofibromatosis type 1 NF1 has been given by National Institute of Health (NIH). To make a positive diagnosis, any two of these cardinal clinical features are required [2].

1. In prepubertal individuals, six or more Café-au-lait spots over 5 mm in greatest diameter and in post pubertal individuals over 15 mm in greatest diameter. Cafe-au-lait spots can be caused by a number of other conditions. Multiple Café-au-lait spots alone are not a definitive diagnosis of NF1.

2. Two or more neurofibromas of any type or one plexiform neurofibroma.

3. Axillary or inguinal region freckles.

4. Optic glioma.

5. Lisch nodules (two or more).

6. A distinct lesion of bones, such as kyphoscoliosis [3], sphenoid dysplasia, or thinning of the cortex of long bones with or without pseudoarthrosis.

7. If NF1 is diagnosed in a first degree relative (parent, sibling or offspring) by the above criteria.

Our patient had one plexiform neurofibromatosis and a distinctive osseous lesion, kyphoscoliosis, so he was diagnosed as a case of NF1 according to these criteria. The NF1 gene mutations manifest a phenomenon called variable expressivity. It means the disorder is expressed differently even amongst people of the same family.

In most of the cases, plexiform neurofibromas are congenital [4]. A typical lesion of plexiform neurofibromatosis is composed of sheets of neurofibromatous tissue. These may infiltrate and encase nerves, blood vessels and other important structures. The primary treatment option for plexiform neurofibromatosis is surgery [5]. Plexiform neurofibromas can be large and cross tissue boundaries hence, their removal is difficult [6]. As these lesions encase and infiltrate structures so it is difficult and sometimes impossible to routinely resect them. This may cause significant damage to surrounding nerves and tissue. About 10% of plexiform neurofibromas undergo transformation into a Malignant Peripheral Nerve Sheath Tumour (MPNST) [7]. There is a possibility of malignant transformation and it may be the cause of removal in some cases besides pain [6]. In the present case histopathology of the excised mass showed no malignant transformation. Radiation and chemotherapy can be used as treatment modalities if a plexiform neurofibroma has undergone such changes. After excision, secondary closure or skin grafting or skin flap is also indicated due to the involvement of large area [8]. In this case primary closure was done as the skin edges approximated properly and there was no need of skin grafting or secondary closure.

CONCLUSION

As the size of plexiform neurofibromatosis increases excision

becomes more difficult. It can encase vital structures and excision may result in injury. Also the risk of malignant transformation increases with the delay. Thus, these cases should be treated with early excision before they become massive and before malignant transformation.

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