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A case report of Plexiform Neurofibroma: A 28 kg baggage on an 18 year old

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ABSTRACT

This is a case report on how an 18 year old had to live with a growth on her back since last twelve years due to lack of medical facilities and taboos associated with these growths in developing nations like India. The girl had stunted growth due to the weight she was carrying and associated deformities in her spine. She was evaluated and finally underwent surgery for the same. Plexiform neurofibromas are tumors of the peripheral nerve sheaths. They occur in people born with genetic disease called neurofibromatosis. They are slow growing tumors and keep on growing with age. They are soft to begin with. There are instances when plexiform neurofibromas have shown rapid growth. Neurofibromas are generally considered benign and not removed until they cause any complication or for cosmetic reasons. We hereby present a case of plexiform neurofibroma which grew to reach a weight of 28 kgs and ended up causing spinal deformities in a young girl.

Background: Plexiform neurofibromas are tumors of the peripheral nerve sheaths. They occur in people born with genetic disease called neurofibromatosis. They are slow growing tumorsand keep on growing with age. They are soft to begin with. There are instances when

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plexiform neurofibromas have shown rapid growth. Neurofibromas are generally considered benign and not removed until they cause any complication or for cosmetic reasons.

Methods:. We hereby present a case of plexiform neurofibroma which grew to reach a weight of 28 kgs and ended up causing spinal deformities in a young girl.

Results: This is a case report on how an 18 year old had to live with a growth on her back since last twelve years due to lack of medical facilities and taboos associated with these growths in developing nations like India. The girl had stunted growth due to the weight she was carrying and associated deformities in her spine. She was evaluated and finally underwent surgery for the same

Conclusions:in India, the lack of medical facilities and the negligence still met to the girl child is responsible for such humungous growths presenting at the tertiary centres even in the 21st century. It is the need of the hour to work on both these challenges**.**

Keywords: plexiform neurofibroma, neurofibromatosis, scoliosis

INTRODUCTION

Neurofibromatosis 1 is an autosomal dominant genetic condition caused by mutation in the Neurofibromatosis(NF1) gene chromosome 17 which encodes neurofibromin, a tumor suppressor protein. The skin manifestations include café au lait macules, axillary freckles and neurofibromas along the nerves. All the autosomal conditions express incomplete penetrance and variable expressivity.

Plexiform neurofibromas are an uncommon variant of this disorder where there are multiple bulging masses which typically give the impression of ‘bag of worms’on palpation. They develop early in life and might transform to malignant nerve sheath tumors [1] .We report a case of plexiform neurofibroma which grew to a massive size and led to spinal deformities and stunting in a young girl. It was not associated with any defaecation or micturition difficulties but the girl had difficulty in standing up and walking. We hereby discus how the lesion was evaluated and the girl was operated and discharged without any complications. Individuals with this disorder have a predisposition to benign and malignant tumor formation and the hallmark lesion is the neurofibroma, a benign peripheral nerve sheath tumor [2]. Malignant transformation is usually marked by rapid increase in size and associated pain. Neurofibromatosis 1 (NF1) is an inherited, fully penetrant ,tumor predisposition syndrome with a birth incidence as high as 1in 2000 live births[3].

It belongs to a group of disorders which are referred to as phakomatoses or neurocutaneous syndromes causing typical skin manifestations..NF1 is a common disease that mainly affects

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the skin and peripheral nervous system and causes characteristic bony dysplasia. By contrast, NF2 where neurological symptoms are the predominant problem and the cardinal sign is bilateral vestibular schwannomas occurring along the course of the eighth cranial nerve [4] . WHEN THE EIGHTH NERVE IS AFFECTED THERE ARE SYMPTOMS RELATED TO HEARING AND BALANCE.

Case report:

An 18 year old female was referred for evaluation of the mass which has been growing on her back since the last 12 years. There was no similar history in her relatives. The girl was comparatively shorter in height as compared to her other siblings which could be due to the weight of the mass. She also had an associated scoliosis though there was no infiltration of the bones by the mass. She had difficulty in walking. We hereby display the image of the mass trying to give an idea about the size in figure *1*. figure *2* shows the associated deformity of the spine

Figure 1: Tends to give an idea about the size of the growth

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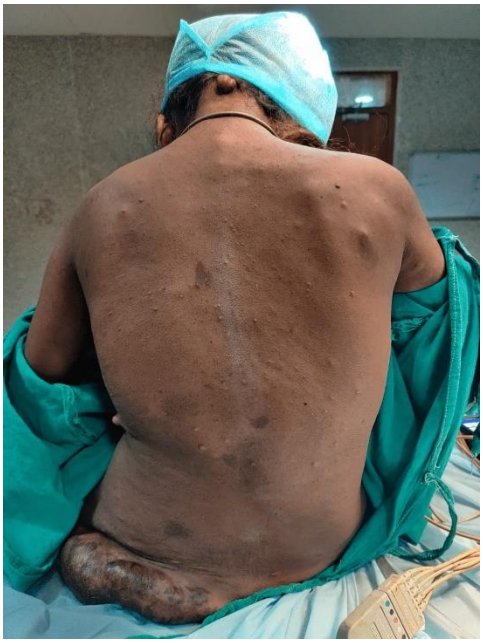

Figure 2: Spinal deformity, associated scoliosis

On examination, her spine was deviated to the right. The patient did not complain of any difficulty in evacuation of bowel or bladder although she complained of difficulty while sqautting.There were multiple neurofibromas on other parts of her body. The surgery team decided to remove the most massive neurofibroma on her back which restricted the movement of the patient and the patient was counselled to defer the surgery for the other smaller neurofibromas.

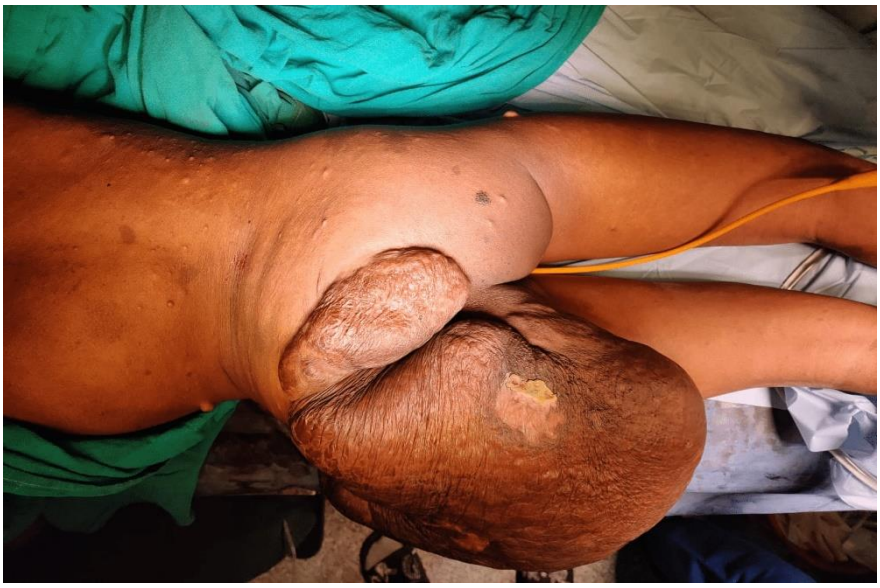

Figure *3* Multiple neurofibromas on different parts of the body of the patient.

Her routine investigations were normal apart from her hemoglobin for which she received blood transfusion. Her histopathological reports suggested spindle cell neoplasm which

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favoured plexiform neurofibroma. Her MRI reports showed loss of cervical lordosis, straightening of dorsal spine with mild right lateral wedging of D11 vertebra. Severe scoliosis of lumbar spine with possible features of plexiform neurofibromatosis involving dorsolumbar spine and diffuse cutaneous neurofibromas. figure *4* shows the MRI imaging with the sagittal and transverse section views wherein the spinal cord and the mass have been labelled.

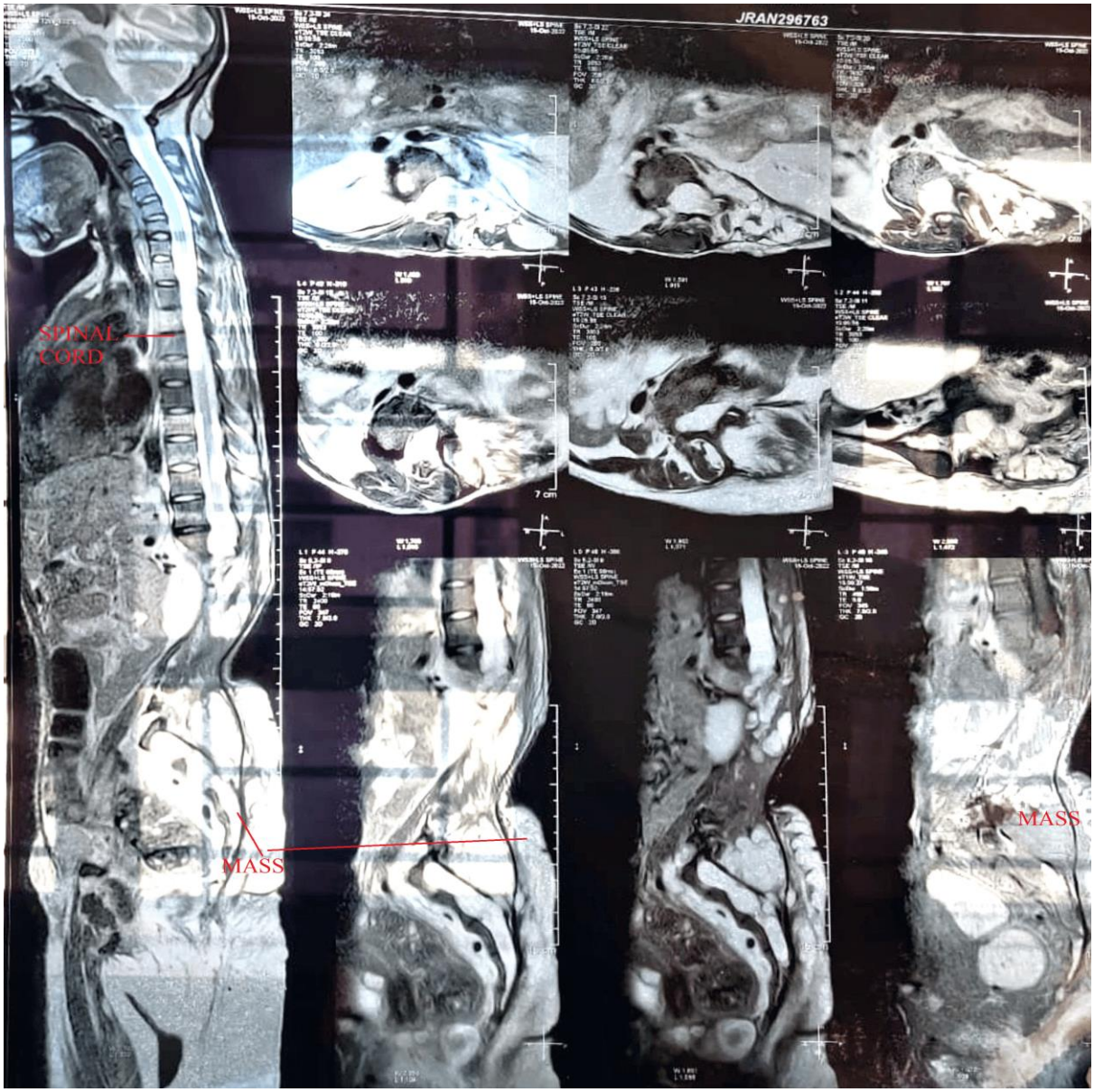

Figure 4- MRI imagine of the lesion showing it's relation with the spinal cord

After pre anesthetic checkup the patient was posted for surgery. The patient was operated under spinal anesthesia in prone position. The patient was kept under observation for 24 hours. There were no post operative complications. figure *5* shows the post operative image of the patient after removal of the mass. The wound was closed with a negative suction drain

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in situ due to the cavity formed post removal of the mass to prevent accumulation of any collection.

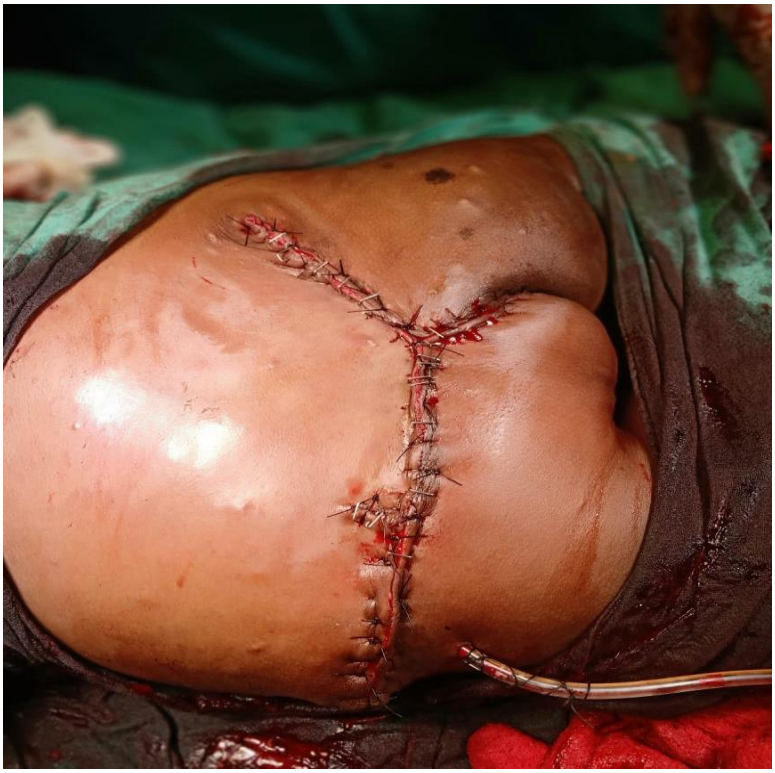

Figure 5- Image after excision of the mass with romo vac drain in situ

The patient was orally allowed liquids after 24 hours and semisolid diet was introduced after 48 hours. Urinary catheter was removed on post operative day 3.The drain output was 50 ml for two consecutive days and became minimal on post operative day 3 following which the suction drain was removed on post operative day 4. There were no associated neural deficits. The patient was able to pass stool and micturate. She was discharged on post operative day 5.

Discussion

Plexiform Neurofibromas are a rare variant of neurofibromas which can occur anywhere in the body. There have been case reports of massive growths in various parts of the body. The lesions can be diagnosed clinically based on the feel of the growth described as bag of worms as mentioned by Tchernev et al [1].

The role of histopathology is to rule out the malignant transformation. Ferner et al states neurofibromatosis is related to the mutation in NF1 gene located on chromosome 17q11.2 [2].

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Management is surgical in order to remove the deforming masses and areas of suspected malignant transformations. Cases with malignant transformation are associated with rapid growth and pain.The diagnosis of NF1 is made on clinical grounds when two or more of the clinical criteria like irish nodules axillary freckling etc are present.

These disorders are associated with increased chances of malignancy. although the chance of malignant conversion in plexiform neurofibroma ranges from 2-5% [3].

Conclusions

We hereby present a case of plexiform neurofibroma in an 18 year old female which was massive enough to cause growth stunting and scoliosis in the patient. The delay in treatment was assumed to be due to the lack of facilities in resource poor countries and also the taboo associated with such growths and lack of care for the girl child.

The female underwent all investigations and finally was operated on. She was kept nil per oral for twenty four hours following which liquids and semisolids were introduced. The drain and urinary catheter were removed subsequently and the patient was discharged on post op day 5 without any complications.

DECLARATIONS

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Conflict of interest: none

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