Spindle cell rhabdomyosarcoma in neurofibromatosis patient: A rare entity

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Abstract
Problem with rare tumour is that there is no much literature available to know how the tumour behave to different modalities of treatment. Here we report a case of spindle cell rhabdomyosarcoma in a patient of neurofibromatosis. This might be first case report of such condition.

Keywords: Spindle cell rhabdomyosarcona, Neurofibromatosis.

Introduction
Problem with rare tumour is that there is no much literature available to know how the tumour behave to different modalities of treatment. We have to mostly rely on our clinical acumen and past experiences to deal with such tricky situations. Here we report a case of spindle cell rhabdomyosarcoma in middle aged female patient who is also a known case of neurofibromatosis.

Neurofibromatosis type 1 is an autosomal dominant, multi-system disorder affecting approximately 1 in 3500 people.[1] It is characterised by the presence of multi-system tumors throughout the skin and central nervous system, which carries a risk of malignant transformation.[2]

Spindle cell rhabdomyosarcoma is a rare variant of embryonal rhabdomyosarcoma. It is a rare tumour that mostly occurs in childhood and it is more rare in adulthood. It most commonly involves the paratesticular region followed by head and neck.[3]

In adults only isolated cases has been reported in the literature. Nascimento et al[4] in their study reported 16 cases, mainly involving head and neck region, followed by retroperitoneum, thigh, leg, sub-scapular area, hand, vulva, and paratesticular region (1 case each).

Though there are various case reports of Spindle cell rhabdomyosarcoma in the literature now available but this tumour in patient with neurofibromatosis has been not reported. This might be first case report of such condition.

Case Report and Technique
A 47 year old female patient referred to us for evaluation from orthopaedic department with tumour over volar aspect of right forearm. She was suffering from tumour over her right forearm since 5 months. She already underwent excision once one month back from other hospital but she had recurrence 2 weeks after surgery. Then she was referred to our hospital.

Her previous histopathological examination report suggested malignant mesenchymal tumour. At our institution we got re-examination of the slides and subsequently immunohistochemistry was done. It was confirmed to be spindle cell rhabdomyosarcoma. Patient was then evaluated with MRI to plan proper management accordingly.

Clinically she had a large ulcerative fungating tumour of size 15x8 cm over volar aspect of right forearm. She had restriction of movement at wrist as well as decreased power of the right hand.

We then plan to completely removed the tumour and do the reconstruction accordingly. Intra op tumour was found to involve Flexor digitorum superficialis muscle, Palmaris longus tendon, Flexor carpi radialis muscle and superficial radial nerve.

Median was displaced and carefully separated and safe guarded. Tumour along with involved structures removed en bloc. Reconstruction was planned with free ALT flap for skin cover with neurotised vastus lateralis muscle for FDS muscle. Recipient vessels were radial artery and cephalic vein.

Post op recovery was uneventful. She was put on active physiotherapy protocol after two weeks. Her hand functions improved significantly. Now she is undergoing adjuvant radiotherapy.

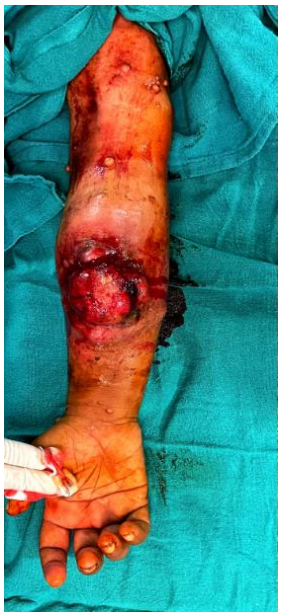

Fig. 1: Pre-operative picture showing tumour over right forearm volar aspect.

Fig. 2: intra operative picture showing tumour and carefully separated median nerve.

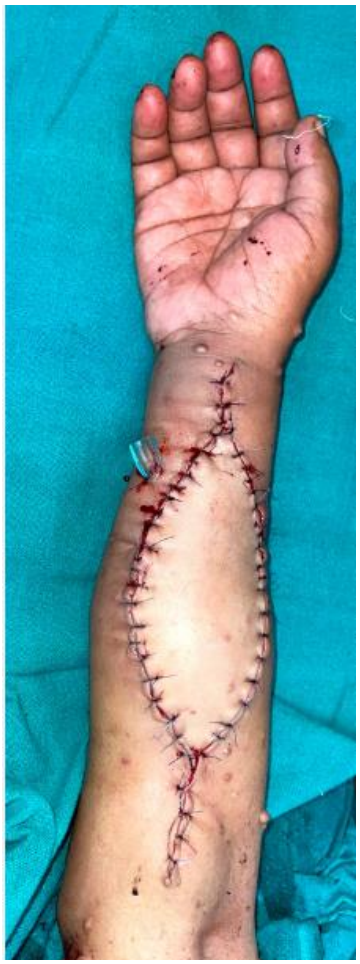

Fig. 3: Immediate post-operative picture.

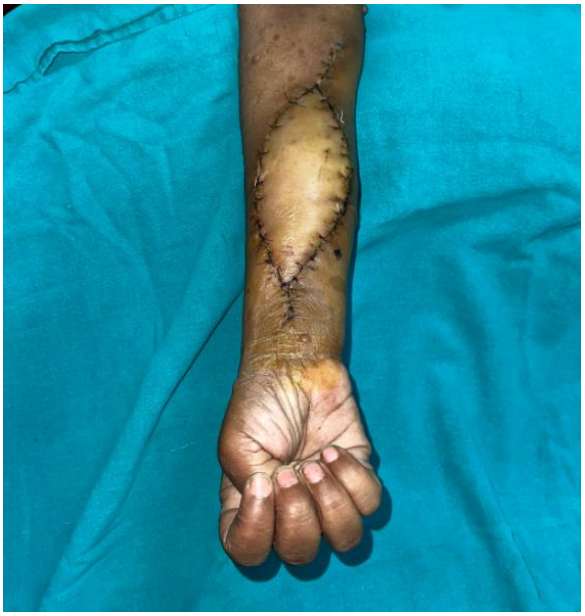

Fig. 4: POD 7 showing settled flap and patient is able to make a fist.

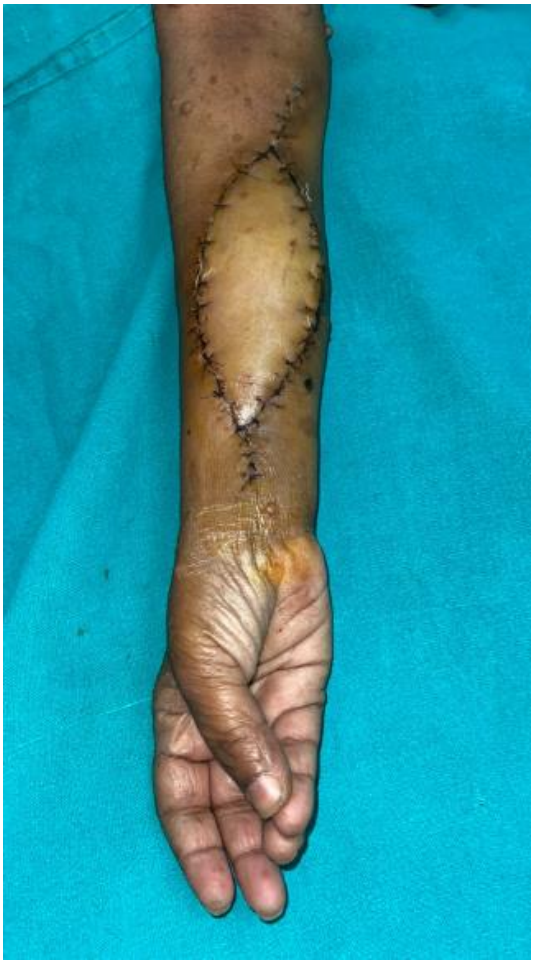

Fig. 5: POD 7 showing patient showing opposition.

Fig. 6: Late post-operative showing well healed and settled flap.

Discussion

Rhabdomyosarcoma is a discrete entity characterised by morphologic, immunohistochemical, and ultra- structural evidence of sarcomeric differentiation.[5] It is a well-recognised variant of embryonal RMS that was first described in 1992 in the paediatric population by the German-Italian Cooperative Sarcoma Study.[6]

In 1998, Rubin et al described the first examples of spindle cell RMS affecting adults. Based on their initial observations, this tumour appeared to have a more aggressive clinical course with both patients dying of disease shortly after diagnosis.[7]

Treatment for this type of tumour is multi-disciplinary. In children chemotherapy, radiotherapy and surgery all have role in disease control. But in adult due to paucity of data, they are mainly treated with protocol devised for children. Different studies shows varied response to different modalities of treatment.[8]

As our case presented with tumour over the periphery that is right forearm and also ulcero-fungating in nature as well as compromising hand functions, we decided first to do the surgical clearance and then go for other modalities of the treatment. Once patient healed she was then referred to radiotherapy department for further management.

Conclusion

Here we reported a rare (first) case of spindle cell rhabdomyosarcoma in a middle aged female with neurofibromatosis. We hope this piece of information might help others who face this that of scenario where there is paucity of data available.

Conflict of Interest

The authors declare that there are no conflicts of interest in this paper.

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