Duchanne muscular dystrophy and rhabdomyosarcoma; association or coincidence?

[1]Narhari Prashant, [2]Singh Sarbhjit
[1]Orthopaedic unit, Loh Guan Lye Hospital, Pulau Pinang.
[2] Orthopaedic department, Hospital Raja Permaisuri Bainun, Ipoh.

INTRODUCTION:

Duchanne muscular dystrophy (DMD) and rhabdomyosarcoma (RMS) are both rare diseases that occurs predominantly in children. It has been reported only 3 times in the literature.[1] We report a rare case of DMD with embryonal RMS in early adult patient.

REPORT:

A 22 year old man with DMD and cardiac failure presented with a painless growing mass at the back of his right thigh for 6 months. He has no lower limb functional movement however does have satisfactory bilateral hand function allowing him to operate a powered wheel chair. On examination, the swelling measuring 10 x 8 cm is deep and fixed to the muscle. MRI and CTTAP suggested an isolated tumour in the hamstring compartment of the right thigh. Biopsy confirmed embryonal RMS. Considering poor cardiac function, meticulous discussion with a MDT, it was deemed that upfront surgical resection will be the treatment of choice rather than neoadjuvant chemotherapy. Chemotherapy would carry the risk of cardiotoxicity and neurotoxicity which will potentially have significant morbidity. Wide resection of the entire hamstring compartment preserving the sciatic nerve was done. There were no wound complication. Resection margin was excellent at 5mm. Despite the good margin, patient was offered radiotherapy however the family did not opt of it in view of potential complications. At nine months follow up, he has no local recurrence nor distance metastasis. His functional status remains same as pre operatively. DMD is a rare x-linked rescesive disease caused by mutation in the DMD gene, and RMS is a rare soft tissue sarcoma. In view of RMS arising from the skeletal muscle, these 2 diseases has been linked but the evidence is scarce.

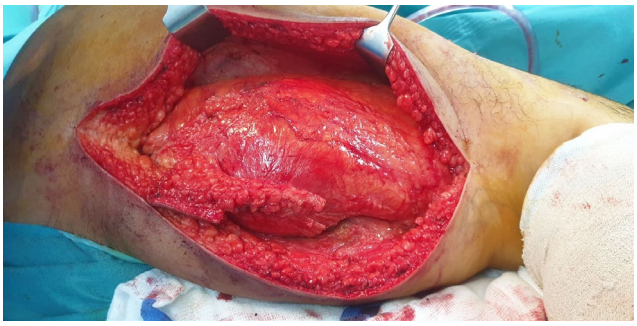

Figure 1: wide excision of posterior thigh rhabdomyosarcoma

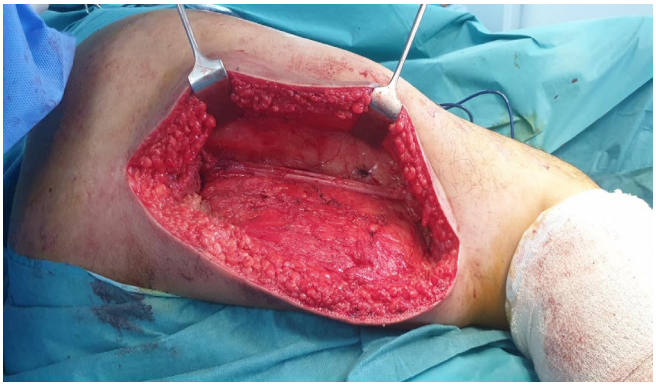

Figure 2: visualised sciatic nerve post tumour resection

CONCLUSION:

Gene abnormality in DMD patients probably predispose them to developing other malignancies especially RMS. Due to its rarity, this patient impose a treatment dilemma. A MDT discussion can be helpful to map out the best care.

REFERENCES:

1. Chandler, E., Rawson, L., Debski, R., McGowan, K. and Lakhotia, A., 2021. Rhabdomyosarcoma in a Patient With Duchenne Muscular Dystrophy: A Possible Association. *Child neurology open*, *8*, p.2329048X211041471.