Challenges in Managing Extra-abdominal Desmoid Fibromatosis in Children

Safie K[1], Zaman A[1], Osman M[1,] Lim FSK[2], Mohd Zaki F[3], Abd Shukor N[4],

1. Paediatric Surgery Unit, Department of Surgery, Hospital Canselor Tuanku Muhriz, University Kebangsaan Malaysia Medical Center, Kuala Lumpur, Malaysia.
2. Anaesthesiology Department, Hospital Canselor Tuanku Muhriz, University Kebangsaan Malaysia Medical Center, Kuala Lumpur, Malaysia
3. Radiology Department, Hospital Canselor Tuanku Muhriz, University Kebangsaan Malaysia Medical Center, Kuala Lumpur, Malaysia
4. Pathology Department Hospital Canselor Tuanku Muhriz, University Kebangsaan Malaysia Medical Center, Kuala Lumpur, Malaysia

Introduction

Desmoid Fibromatosis (DF) is a benign soft tissue tumour, which is locally invasive, does not metastasize but has a high potential of recurrence. We report a rare case of extra-abdominal desmoid fibromatosis presenting as a slow growing left gluteal mass.

Case Summary

An 8-year-old boy presented with a slow growing painless left gluteal swelling for 6 months. He has no associated bowel, bladder, or lower neurological deficit symptoms. Examination of the gluteal region showed a mobile mass 1x2 cm on the medial side of the left gluteal fold, firm in consistency, non-tender with no overlying skin changes. Pelvic MRI, showed a subcutaneous nonspecific soft tissue tumour with no malignant changes. In view of the diagnosis issue, we proceeded with excision of the gluteal mass. Intraoperatively, a well-circumscribed lump measuring 2x 2 cm was removed. Histopathological examination revealed a well-circumscribed nodular lesion with a focal infiltration. The lesion is composed of fascicles of neoplastic spindle cells with collagenized background and foci of myxoid change. The tissue stain positive for SMA and Beta-catenin immuno-stain and diagnostic for desmoid fibromatosis tumour. Tumour margin was involved. The case was discussed during multidisciplinary team meeting and was agreed for active monitoring and surveillance.

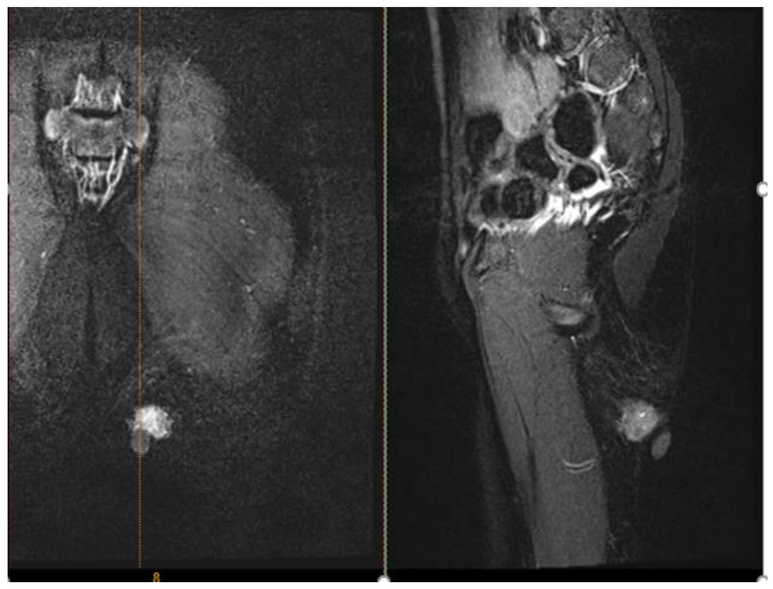

Figure 1: MRI of left gluteal mass

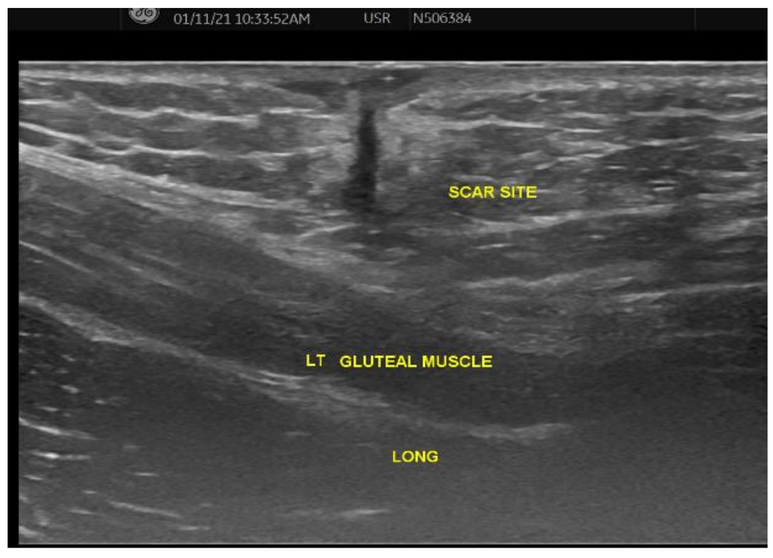

Figure 2: USG of left gluteal post removal of lesion

Discussion

Diagnosis of desmoid fibromatosis (DF) requires a pathologic review of a tissue specimen. Peak incidence among paediatric patients occurs between 5 to 8 years of age. These tumours can occur throughout childhood, and most commonly extra-abdominal. Extra-abdominal desmoid tumours (DF) are often difficult to remove because of the difficulty in differentiating an intraoperative demarcation between the diffusely infiltrating tumour and the surrounding healthy tissue. The recurrent rate after surgery reported range between 22% and 76%

In view of this, multimodal therapy is advocated in adult patients. Radiotherapy though a treatment for adult is not recommended for children due to its effect on bone growth. Results for chemotherapy is encouraging but again has its own side effect. The POG 9650 study trial is looking at the usage of NSAIDs as chemotherapy. Although the result is encouraging, the study is still in phase II and complication and side effect has yet to be determine. We have decided on wait and watch approach as patient is asymptomatic and this approach proven to be safe as 50% showed no disease progression in 5 years. Furthermore, about 20% to 28% of DF tumour showed spontaneous regression if managed non-surgically.

Conclusion

This case highlights the challenge in managing desmoid fibromatosis as usually tissue is required for diagnosis as imaging is often non-diagnostic. However with surgical intervention there is a higher risk of recurrence, especially in extra-abdominal desmoid fibromatosis. So far no consensus on the management of residual tumour after excision, wait and watch is one of the options.

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