Polish Annals of Medicine

Journal homepage: https://www.paom.pl

Case report

The dilemma in management of desmoid tumour: A case report

Fei Yee Lee[1], Arvind Vashdev Jagwani[1], Mohamad Fairuz Mohamad Sharin[1], Andee Dzulkarnain Zakaria[2], Firdaus Hayati[3], Ismail Sagap[4]

[1] *Department of Urology, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, Selangor, Malaysia*
[2] *Department of Surgery, School of Medical Sciences, Hospital Universiti Sains Malaysia, Kubang Kerian, Kelantan, Malaysia*
[3] *Department of Surgery, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia*
[4] *Colorectal Unit, Department of Surgery, Universiti Kebangsaan Malaysia, Cheras, Kuala Lumpur, Malaysia*

Article info

Article history
Received: March 20, 2022
Accepted: May 30, 2022
Available online: September 20, 2022

Keywords
Familial adenomatous polyposis
Papillary thyroid carcinoma
Desmoid tumour

Doi
https://doi.org/10.29089/paom/150516

Abstract

Introduction: Desmoid tumours are rare benign tumours. The tumours may develop sporadically or they may be linked to familial adenomatous polyposis (FAP) in Gardner's syndrome.

Aim: This article highlights a young lady with an intra-abdominal desmoid tumour that manifested as an acute abdomen and we discuss our management strategy.

Case study: A 24-year-old lady with a known FAP who had undergone a pan-proctocolectomy with an ileal pouch-anal anastomosis and total thyroidectomy, later was complicated with acute abdomen for symptomatic desmoid tumour. Computed tomography of the abdomen showed a large intraperitoneal heterogenous mass with small bowel displacement and was confirmed by magnetic resonance imaging. An exploratory laparotomy and en-bloc resection of the tumour with an end ileostomy were performed.

Results and discussion: Intra-abdominal desmoid tumours usually present as a painless slow-growing mass, however, in severe form, it can cause bowel ischemia, intestinal obstruction, or deterioration of function in the ileoanal anastomosis, among post total colectomy. Surgery is indicated upfront in specific clinical scenarios namely complications (occlusion, perforation, or bleeding) or major cosmetic issues.

Conclusions: Desmoid tumour, despite being benign, is challenging to manage due to its compressing nature. As a key point, the diagnosis of a desmoid tumour should be suspected and followed up closely in patients with a previous history of FAP in combination with extracolonic manifestation.

Corresponding author: Firdaus Hayati, Department of Surgery, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, 88450, Kota Kinabalu, Sabah, Malaysia.
E-mail address: m_firdaus@ums.edu.my

1. INTRODUCTION

Desmoid tumours originate from fascial or musculoaponeurotic structures, with a tendency for fibroblastic proliferation. Being such, they tend to be locally aggressive with a higher recurrence rate, but depict no metastatic potential. Due to the inability to metastasize, desmoids are classified as benign. They account for a lesser 3% of all soft tissue tumours and about 0.03% of all neoplasms.[1] Desmoid tumours may develop sporadically or be linked with familial adenomatous polyposis (FAP), known as Gardner's syndrome. They are rare in the general population but in FAP, it affects approximately 15% of patients.[2] Both FAP and Gardner's syndrome arise from similar APC gene mutations. We present a 24-year-old lady with an intra-abdominal desmoid tumour that manifested as an acute abdomen and we discuss our management strategy.

2. AIM

We describe a case of a young lady with an intra-abdominal desmoid tumour that manifested as an acute abdomen and we discuss our management strategy.

3. CASE STUDY

A 24-year-old lady underwent a panproctocolectomy with an ileal pouch-anal anastomosis and total thyroidectomy 4 years ago for FAP and papillary thyroid cancer (PTC) respectively. She presented to the emergency department with progressive abdominal distension over 3 months. The distention was associated with intermittent colicky abdominal pain, diarrhoea, poor oral intake, and painless left lower limb swelling. There was no history of vomiting. On admission, she was found to be febrile, malnourished, and cachexic with a body mass index of 17. There was no peritonism.

Blood investigations showed leukocytosis, anaemia, electrolyte imbalances, and hypoalbuminemia. The chest radiography was unremarkable. The patient was treated for pouchitis with intravenous antibiotics and started on total parenteral nutrition because of her poor nutritional status. Computed tomography (CT) of the abdomen showed an intraperitoneal heterogenous mass measuring 130 × 175 × 295 mm. The mass was located centrally displacing the small bowel loops anteriorly and laterally. She was subjected to magnetic resonance imaging (MRI) of the abdomen to determine the tumour origin and to assess the resectability of its relationship with the surrounding structures. The MRI revealed a large intraperitoneal soft tissue mass causing displacement of bowel loops and anterior compression of the abdominal aorta as well as the lower part of the inferior vena cava (Figure 1). There was no obvious infiltration into surrounding structures. Bilateral hydroureteronephrosis was also observed for which a retrograde pyelogram and bilateral ureteric stenting were performed.

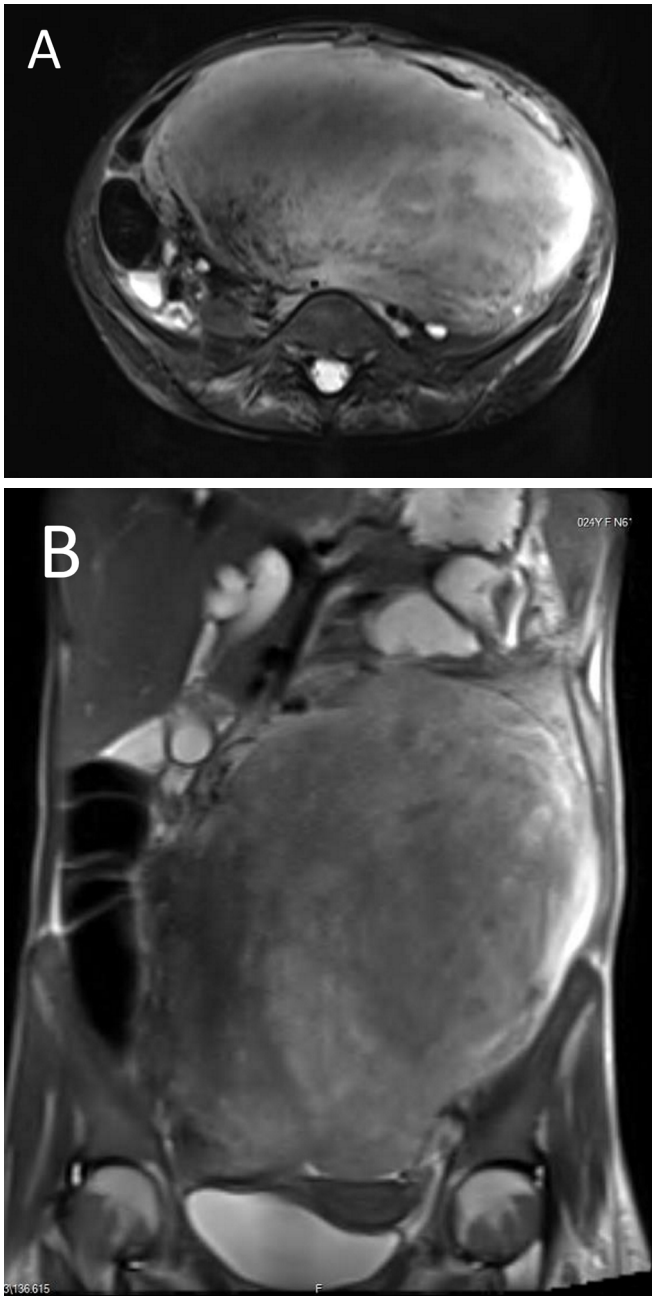

Figure 1. The MRI in axial (A) and coronal (B) view revealed a large abdominal mass pushing the bowel loops to the sides and above, posteriorly compressing the abdominal aorta, the lower part of inferior vena cava, and both ureters causing bilateral hydroureteronephrosis.

Her general condition gradually improved over 2 weeks. An exploratory laparotomy and tumour excision were then carried out due to the compressive nature of the large tumour. The tumour measured 20 × 20 cm and was noted to arise from the small bowel mesentery close to the ileoanal pouch. En-bloc resection of the tumour with an end ileostomy was performed (Figure 2a). The patient was nursed and closely monitored in the intensive care unit. She had an uneventful postoperative recovery and was discharged well 10 days later. The histopathology examination revealed a macroscopically fairly circumscribed tumour measuring 200 ×

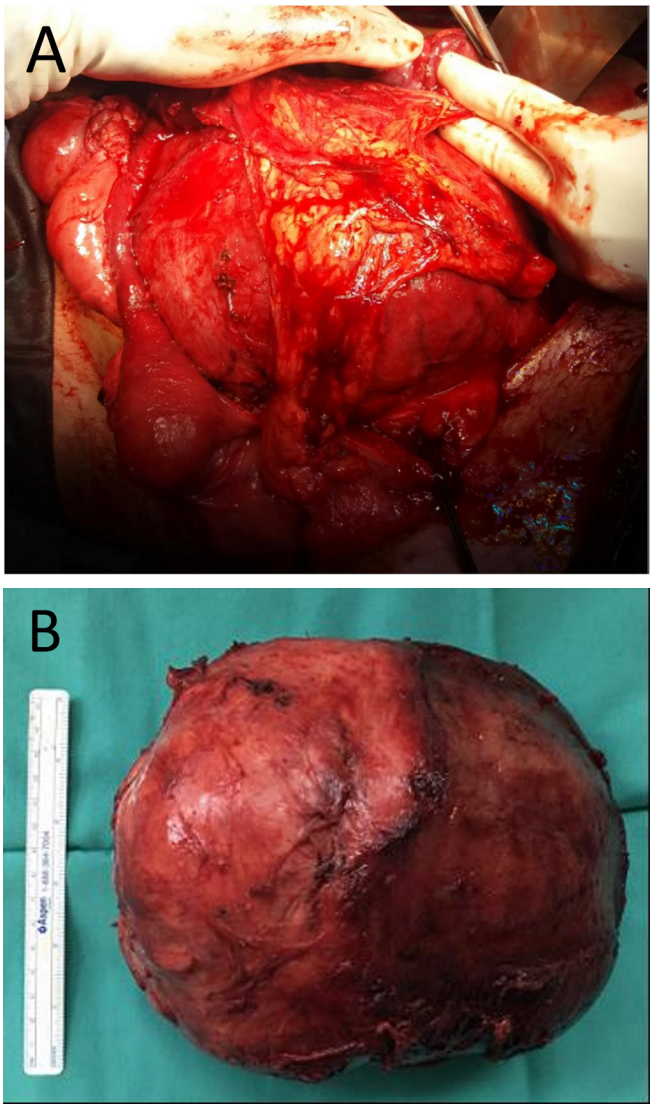

Figure 2. En-bloc resection of the tumour was performed (A); the gross specimen of the complete resection of a fairly circumscribed tumour with a smooth surface measuring 20 × 15 × 12 mm and weighing 2 kg (B).

150 × 120 mm, weighing 2 kg with a homogenous greyish surface on the cut section (Figure 2b). Microscopic examination of the resected specimen showed interlacing fascicles of uniform spindle cells set in the background of the stroma. The circumferential margin is free from the tumour cells. No nuclear atypia and mitosis are seen. Immunohistochemically, the neoplastic spindle cells are negative toward smooth muscle actin (SMA), Desmin, CD34, and S100.

4. RESULTS AND DISCUSSION

Even though desmoid tumours are microscopically benign with negligible metastatic potential, the behaviour of these tumours can cause substantial morbidity and mortality. Bowel obstruction, hydroureter, hydronephrosis, and ischemic lesions are among the complications, but in severe forms, they can complicate with abscess, fistulas, gastrointestinal bleeding, and perforation.[3] In our case, she presented with an acute abdomen without apparent severe complications. Desmoid tumours are recognized to be the main reason for mortality in FAP patients who underwent a colectomy. Gardner's syndrome is also associated with an increased risk of several extracolonic tumours including gastric and duodenal polyps, PTC as well as dermoid tumour.[4,5] This is clearly exhibited in our patient who also had PTC and patients younger than age 35 are at greater risk of getting well-differentiated thyroid cancer namely PTC. Therefore all patients with FAP should undergo regular thyroid examination, screening as well as surveillance, as in our case.[6] Besides, they also need to undergo upper endoscopy based on the Spigelman scoring system.[4]

CT or MRI of the affected area is used to determine the association of the tumour to the surrounding structures and for assessment of resectability as well as any anticipated complications.[7] Commonly, intra-abdominal desmoid tumours can be adequately evaluated by CT but in the case of truncal or extremity tumours, MRI is the preferred imaging use.[8] There are no radiographic characteristics that can reliably distinguish desmoid tumours from malignant soft tissue tumours. Histologically, desmoid tumours are characterized by a fairly-poor circumscribed pattern and are composed of interlacing fascicles of uniform spindle cells set in the background of stroma or whorling patterns with bland nuclear features and dense keloid-like collagen in areas. The cells usually exhibit no nuclear atypia or hyperchromasia. Tumour cells often invade the surrounding structures including the soft tissues and skeletal muscles. Vimentin will be strongly positive but SMA will show variable reactions upon immunohistochemical stain. Histologically, there is no difference between sporadic and FAP-related desmoid tumours found.[8]

The current treatment strategy for managing desmoid tumours includes active surveillance for a period of 1–2 years followed by medical treatment, surgery, and/or radiation in cases of prolonged disease progression.[9] Surgery is indicated upfront in specific clinical scenarios namely complications (occlusion, perforation, or bleeding) or major cosmetic issues, as in our case.[9] Complete surgical excision with negative microscopic margins will be the cornerstone of treatment in the active management of desmoid tumours.[10] When initial resection offers a risk of intestinal ischemia, adhesions, blockage, or fistula, positive microscopic margins might be acceptable.[10] The objective is to decrease morbidity, as resection appears to have no effect on survival. Patients with intra-abdominal desmoid tumour may find surgery difficult or impossible due to a higher risk of local recurrence.[9] In our case, en-bloc resection of the tumour with an end ileostomy was performed, but final histology demonstrated a negative tumour margin. Most of these patients have malnutrition and this has an important implication for patients undergoing surgery. Adequate preoperative nutrition can lower in-hospital mortality when compared to inadequate nutritional support.[11] These patients also have increased susceptibility to infection, poor wound healing, and bacterial overgrowth in the gastrointes-

tinal tract.[12] Therefore thorough assessment, early intervention, and involvement of a multidisciplinary team may be appropriate in the management of patients who are severely malnourished at presentation.

If surgery is not an option and the existence of a residual tumour necessitates active management, moderate dosage of radiotherapy may be an effective therapeutic option if the adverse effects, such as radiation enteritis and late radiation malignancy, are managed appropriately.[13] It can be used in both adjuvant and definitive therapy for desmoid tumours. Based on the recommendation, the dose given is 50–56 Gy in 2 Gy fractions.[8] The choice of systemic therapy is given to patients who are inoperable or not suitable for radiotherapy and those with initial treatment of a large intra-abdominal desmoid. Hormonal therapy, non-steroidal anti-inflammatory drugs, chemotherapy, and targeted therapy are among the systemic therapy treatments of choice.[10] Close monitoring with CT or MRI should be done monthly for the first 2 months, then three times a year for the 1st year, then six times a year until the 5th year, and then once a year after that.[8] A robust and intensive monitoring regimen is required, particularly during the 1st year, to allow early detection of the rapid progression of diseases.

6. CONCLUSIONS

The rarity of desmoid tumours, besides the diversity in their clinical courses, makes them a challenging entity to diagnose and demonstrate the efficacy of any single intervention. Despite active surveillance, for any patient with symptoms or complications of the desmoid tumour, surgery is warranted with intention of negative microscopic margin. The diagnosis of a desmoid tumour should be suspected and followed up closely in patients with a previous history of FAP in combination with extra colonic manifestation such as thyroid cancer as seen in our patient.

Conflict of interest

The authors have declared no conflict of interest.

Funding

None declared.

Acknowledgments

We would like to thank everybody who has involved directly and indirectly in managing this case.

References

1 Fletcher CDM, Bridge JA, Hogendoorn PCW, Mertens F. *Desmoid-type fibromatoses. WHO Classification of Tumours of Soft Tissue and Bone (IARC WHO Classification of Tumours)*. 4th ed. Lyon, France: IARC; 2013.

2 Fallen T, Wilson M, Morlan B, Lindor NM. Desmoid tumors – a characterization of patients seen at Mayo Clinic 1976–1999. *Fam Cancer.* 2006;5(2):191–194. https://doi.org/10.1007/s10689-005-5959-5.

3 Sanchez-Mete L, Ferraresi V, Caterino M, et al. Desmoid tumors characteristics, clinical management, active surveillance, and description of our FAP case series. *J Clin Med.* 2020;9(12):4012. https://doi.org/10.3390%2Fjcm9124012.

4 Sahid NA, Hayati F, Sani I, Zakaria Z. Extracolonic manifestation of familial adenomatous polyposis in an asymptomatic patient; literature review on current recommendation. *GMJ.* 2020;31:114–116. http://dx.doi.org/10.12996/gmj.2020.32.

5 Dinarvand P, Davaro EP, Doan JV, et al. Familial adenomatous polyposis syndrome: An update and review of extraintestinal manifestations. *Arch Pathol Lab Med.* 2019;143(11):1382–1398. https://doi.org/10.5858/arpa.2018-0570-ra.

6 Feng X, Milas M, O'Malley M, et al. Characteristics of benign and malignant thyroid disease in familial adenomatous polyposis patients and recommendations for disease surveillance. *Thyroid.* 2015;25(3):325–332. https://doi.org/10.1089/thy.2014.0107.

7 Chan KH, Sellappan H, Hayati F, Azman A. Uterine rupture mimicking Wunderlich's syndrome in pregnancy: An unfortunate case. *Pol Ann Med.* 2022;29(1):74–77. https://doi.org/10.29089/2021.21.00201.

8 Garcia-Ortega DY, Martín-Tellez KS, Cuellar-Hubbe M, et al. Desmoid-type fibromatosis. *Cancers (Basel).* 2020;12(7):1851. https://doi.org/10.3390%2Fcancers12071851.

9 Kasper B, Baumgarten C, Garcia J, Bonvalot S, Haas R, Haller F; Desmoid Working Group. An update on the management of sporadic desmoid-type fibromatosis: a European Consensus Initiative between Sarcoma PAtients EuroNet (SPAEN) and European Organization for Research and Treatment of Cancer (EORTC)/Soft Tissue and Bone Sarcoma Group (STBSG). *Ann Oncol.* 2017;28(10):2399–2408. https://doi.org/10.1093/annonc/mdx323.

10 Desmoid Tumor Working Group. The management of desmoid tumours: A joint global consensus-based guideline approach for adult and paediatric patients. *Eur J Cancer.* 2020;127:96–107. https://doi.org/10.1016/j.ejca.2019.11.013.

11 Kaegi-Braun N, Mueller M, Schuetz P, Mueller B, Kutz A. Evaluation of nutritional support and in-hospital mortality in patients with malnutrition. *JAMA Netw Open.* 2021;4(1):2033433. https://doi.org/10.1001/jamanetworkopen.2020.33433.

12 Gomes F, Baumgartner A, Bounoure L, et al. Association of nutritional support with clinical outcomes among medical inpatients who are malnourished or at nutritional risk: An updated systematic review and meta-analysis. *JAMA Netw Open.* 2019;2(11):e1915138. https://doi.org/10.1001/jamanetworkopen.2019.15138.

13 Howard JH, Pollock RE. Intra-abdominal and abdominal wall desmoid fibromatosis. *Oncol Ther.* 2016;4(1):57–72. https://doi.org/10.1007/s40487-016-0017-z.