

Case Study

Bizarre Retroperitoneal Fibromatosis: Challenges in Management

Abstract

Desmoid tumour is a type of benign neoplasm arising from proliferation of fibroblast cells. Although histologically benign, retroperitoneal desmoid tumours resection can be challenging to surgeons due to its infiltrative nature. Surgery can be mutilating and pose significant morbidity and mortality. We present a case of a 27-year-old lady with retroperitoneal mass presented with chronic abdominal pain.

Case Report

A 27-year-old lady presented with right iliac fossa pain for 1 year duration. There were no history of trauma, fever or significant loss of weight of note. Physical examination revealed non tender abdomen with no palpable mass. Blood investigation revealed no abnormalities. Computed tomography (CT) scan of the abdomen/pelvis with urography was arranged. CT scan showed right retroperitoneal mass measuring 4.4 x 6.4 x 8.1 cm (AP x W x CC) encasing the right ureter, causing right unilateral obstructive uropathy.

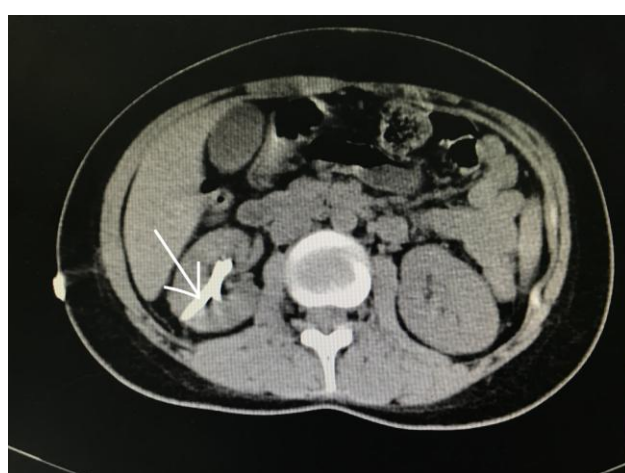


Figure 1: CT scan (Axial view) White arrow showing right hydronephrosis with percutaneous nephrostomy tube.



Figure 2 CT Scan (Axial view)

The white arrow indicating a right heterogeneous, cystic retroperitoneal mass.



Figure 3 CT scan (Coronal view)

White arrow – The right retroperitoneal mass seen to lie close to the right common iliac artery with poor fat plane demarcation.

White arrow head – The right external iliac artery traversing through the retroperitoneal mass.

Emergency right percutaneous nephrostomy has been performed to relieve obstruction. Exploratory laparotomy and tumour resection done soon after the diagnosis of this mass. The right retroperitoneal tumour measured 14 x 8 cm adhering closely to surrounding structures; namely lower pole of right kidney with ureter and major vessels such as right internal and external arteries and veins. Tumour was excised and shaved off from surrounding structures. Iatrogenic tear of vessel and bladder were encountered during surgery and were repaired primarily with sutures and vein patch. Urology team was consulted during the surgery. In order to maximise tumour excision, right gonadal vein was sacrificed. The internal iliac artery was completely encased by the tumour and therefore was also ligated. A small proportion of tumour with the size of 2 x 1 cm was unable to be excised in view of adherence to iliac arteries. The surgery took 9 hours with minimal blood loss. Postoperatively,

patient was put on TED stocking with adequate analgesia. Physiotherapy was commenced as soon as day 1 after surgery to reduce the incidence of deep vein thrombosis. Prolonged catheterisation was planned by the Urology team to allow bladder healing. Repeated blood investigation after the operation were normal. Removal of Foley's catheter done after 2 weeks time after surgery to allow bladder healing after primary repair. Patient was discharged well with no significant postoperative issue and has been under our follow up till now. The histopathological analysis revealed desmoid-type fibromatosis tumour. Microscopically the tumour is poorly circumscribed composed of moderate proliferation of elongated and slender spindle shaped cells arranged in fascicle pattern in a collagenous stroma containing blood vessels. The tumour cells are seen infiltrating the adjacent smooth muscle and adipose tissue. Immunohistochemical studies show the cells are positive towards Beta Catenin. There are no consensus guidelines on medical imaging in follow up. However, as our patient has had an incomplete excision we have chosen to do so with CT scan. CT scan follow up so far revealed no significant interval change of residual tumour and patient remains symptom free.

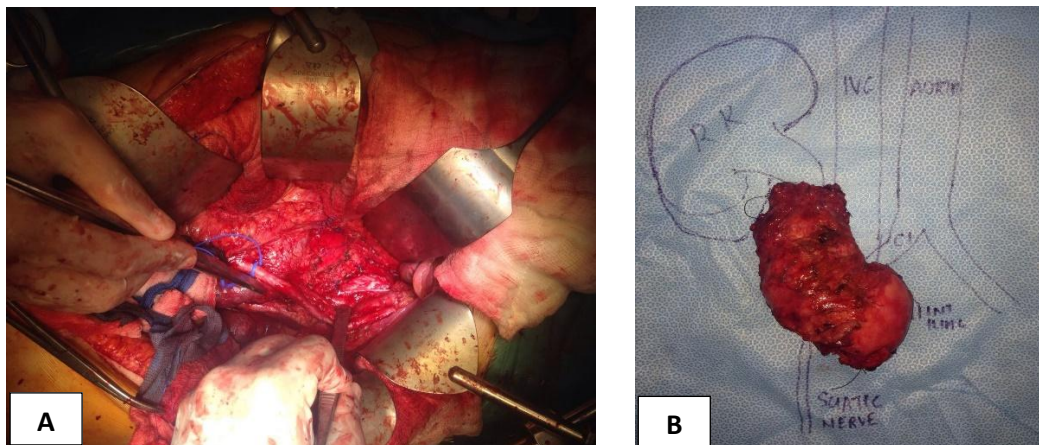


Figure A Intra-operative view of the tumor

Figure B Resected tumor in relation to its surrounding structures

Discussion

The word desmoid comes from the Greek word *desmos*, which means 'tendon-like'. Histologically, desmoid tumours are well-differentiated, overgrowths of fibroplastic tissue. The more aggressive form of desmoid tumour are called desmoid-type fibromatosis (DTF). It has a marked cellularity and aggressive local behaviour. It arises from fascial and musculoaponeurotic tissues. All desmoid tumour lacks capsule and may infiltrate along fascial planes and invade neurovascular structures. It may also cause potential obstruction of vital structures and organs. In this case, patient has obstructive uropathy that requires urgent decompression of the pelvicalyceal system with percutaneous nephrostomy. Macroscopically and microscopically, all desmoid tumours share common traits. The size varies- from as small as less than 5 cm to more than 20cm. The gross appearance is white to tan in colour, with fibrous trabeculations and infiltrative border¹. Desmoid tumour consist of fibroblastic cells with pseudocapsule, hence it tend to infiltrate to surrounding structures².

Desmoid tumour arising from retroperitoneal region is extremely rare with the incidence of 0.2-0.4 / 100000 only throughout the world. It can affect any age group but commonly between 25 to 35 years of age³. Risk factors include female sex, previous surgery or trauma, Gardner's syndrome and familial poliposis coli⁶. Desmoid tumours are twice as common in females than males, and the female-to-male ratio was 1.2:1. In children however, sex incidence is equal. Although desmoid tumours are more commonly diagnosed between 25 to 35 years old age group, they can still occur in children or elderly. Lee et al described that the average age of diagnosis is 41.3 years¹⁰. A person is at risk of developing desmoid tumours if there is a germline mutation of distal codon 1399; any family history of gastrointestinal disease and a previous family history with desmoid tumours. Sporadic mutation of the gene encoding for Beta-catenin d (CTNNB1) has been implicated in the pathogenesis of the desmoid tumour¹¹. Mutation of the Adenomatous polyposis Coli gene or APC gene has also being blame for tumour development. Accumulation of Beta-catenin d results in cellular proliferation and diffferentiation^{3,5}.

Desmoid tumours can be classified according to its sites of occurrence. It can be classified into:

a) Abdominal (commonly associated with presence of Pfannenstiel scar);

b) Extraabdominal (muscles of the extremities and trunks) ;

c) Intraabdominal type (retroperitoneal and pelvic region)

Extra-abdominal desmoid tumours may present as a firm, smooth and mobile mass. They can also adhere to surrounding structures while the overlying skin is spared. Extra-abdominal desmoid tumour may also arise from urological system structure such as scrotum and the bladder^{12,13}

Retroperitoneal desmoid tumour commonly presents with non-specific symptoms such as vague abdominal pain. Retroperitoneal desmoid tumour is rare but is found to be more common in familial polyposis coli and Gardner syndrome after abdominal surgery¹⁴.

CT (Computed Tomography) scan as well as Magnetic Resonance Imaging (MRI) remains the imaging modality of choice in diagnosis and follow up. CT scan is sensitive to evaluate the extent of desmoid tumour and its relation to surrounding structures. This is valuable in planning for surgery. MRI however, is more superior in determining extent and to detect recurrence post surgical excision. CT scan evaluation was invaluable in this case as we managed to anticipate challenges ahead of surgical resection. Preoperative CT scan of the abdomen showed that the right retroperitoneal mass was encasing the right ureter, and therefore Urology unit was alerted prior to the operation day.

Management protocols are not standardised and may range from watchful waiting to wide local excision (WLE), radiotherapy, systemic therapy, hormonal treatment and anti-inflammatory drugs. This is due to its variable clinical behaviour as some may spontaneously regress while others may remain stable or progressively enlarge over time. Watchful waiting is generally advocated for individuals with asymptomatic stable tumors as intervention may result in increased morbidity and mortality.⁴⁻⁵ Moreover, the rate of progression is low (22%) and a higher rate of spontaneous regression is observed (66%).⁸⁻⁹ Literature however does not conclude on factors contributing to the identification of these patients.

Patients experiencing pain, presenting with a palpable mass or obstructive symptoms are generally indicated for intervention. Where medically and technically feasible, complete resection of the tumor with negative margin is performed. This is frequently challenging due to the complex anatomical location of the tumor and its infiltrative nature. Therefore, it is not uncommon for residual tumor to be left inside. Although many sources have claimed that a positive resection margin contributes to the recurrence of DTF, in literature the relationship

between the two is rather unclear . Many case series reported an independence between the risk of recurrence and resection margin. Moreover, this uncertainty has led to the controversy with regards to the utilisation of post-operative radiotherapy or chemotherapy among patients with residual tumors. Primary radiotherapy however can be used in those who are not suitable for surgery, those who decline surgery and those with potentially high surgical morbidity since DTF is relatively radiosensitive. Neoadjuvant radiotherapy and systemic therapy are relatively new, hence data is limited on the utility of this approach.³⁻⁵

As for our patient, surgical excision was performed as she was symptomatic. Post-intervention, despite having residual tumor, radiotherapy was not administered as she remained asymptomatic. Although there are no evidence-based post- treatment surveillance protocols, follow up is done every six months for the first three years, following which every 12 months up to year 6, then every other year. Imaging should be done whenever indicated, every three to six months for the first three years and then, annually³.

Conclusion

Retroperitoneal Desmoid-type fibromatosis (DTF) is a rare disease but surgically challenging to resect due to its infiltrative nature to surrounding organs and vessels. Surgery can be mutilating and has a significant degree of morbidity. Adjunct treatment such as radiotherapy remains controversial as there is a need for further studies.

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