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CASE REPORT

Intra-abdominal Fibroid Lesion of Colonic Involvement

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SUMMARY

Fibroid is a benign lump of growth, occurring within or outside the uterus. It is a rarity for such growths to occur intra-abdominally. Such cases are of aggressive fibromatosis which are marked by the presence of desmoid tumours. It is most commonly seen in patients with a history of familial adenomatous polyposis (FAP) or past surgical procedures. As FAP has a wide fibroepithelial growth spectrum, the symptomatology and therefore the prognosis varies. Unpredictable clinical behaviour, varied location and non-specific presentation are the factors accounting to diagnostic difficulty. Therefore, misdiagnosing the disease is not uncommon. Here we report a case ofa26-year-old pregnant female presenting with lower abdominal pain and mass. Preoperative diagnosis was of uterine fibroids as per radiological evidence, but on exploration it turned out to be a large right colonic mass. The patient underwent right hemicolectomy. Histopathological reports confirmed morphological and immuno-histochemical features indicating fibromatosis. This fibromatous mass though intra-abdominal was not growing from the mesentery/mesocolon or the pelvis. It appeared to grow from the wall of the colon. This is a confirmatory presentation of true colonic wall fibromatosis which is deep and isolated form of intra-abdominal desmoid tumours. As this lesion was isolated in nature, the treatment of choice was surgical resection. It is essential to have a multi-disciplinary team approach in the management of such a patient. This improves the treatment and the prognostic outcomes.

Keywords: Fibroid, Colon, Fibromatosis, Hemicolectomy, Desmoid Tumour

INTRODUCTION

Intra-abdominal desmoid tumour or fibromatosis is a rare locally aggressive, proliferative disease originating from musculoaponeurotic structures.[1]They are benign in nature as they do not have predisposed metastatic characteristics. However, if the tumour is aggressive, non-encapsulated and infiltrating with irregular margins, it should be considered as a malignant variant.[1,2]These benign tumours account for 0.03% of all neoplasms and 3% of soft tissue tumours.[2] However, when it presents in association with familial adenomatosis polyposis (FAP), especially the Gardner syndrome variant of FAP, the prevalence is reported to be 13%.[2] Such growths can occur in any age group, but gender preponderance is noted in females of child-bearing age (during or post-pregnancy), especially in the age group between 25 to 35 years old.[3]McFarlane in 1832 reported the first case of this type of tumour, which was a case of a young post-partum woman, having a mass on the abdominal wall. It was after six years of that case that in 1838, Muller coined the term desmoid.[3] He derived this term from a Greek word “desmos” meaning bands of tendons, from a similar tumour which was also of fibroepithelial origin.[4]

Desmoid tumours have different clinical presentations and hence, subjective prognosis. This owes to the variable biological behaviour they exhibit. Desmoid tumours can be classified according to their presenting site. They can be abdominal, intra-abdominal or extra-abdominal. Intra-abdominal ones, can be further subdivided as mesenteric fibromatosis and pelvic fibromatosis.[4]Accurate diagnosis is crucial for appropriate therapeutic decision and to prevent complications leading to poor prognosis. This depends on meticulous clinical examination supported by radiological imaging but still there are high chances of misdiagnosis, as happened in our case.[5]Initial treatment is always gross total resection with safe margins avoiding any significant disfigurement. Recurrent cases can be well managed by radiotherapy, especially for local control of disease[6].

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CASE PRESENTATION

A 26 -year-old female presented to the Emergency Department at a Private Hospital with complaint of severe abdominal pain and fever for the last five days. She had a one-month history of vague abdominal pain, which was more concentrated in the hypogastrium region. Pain was not associated with vomiting or altered bowl habits. Patient remained afebrile in her initial course of disease. She was 16th week primary gravida. No obstetric complain was reported by the patient. Routine investigations were done and the patient was diagnosed with uterine fibroid as supported by ultrasonography (USG) reports. Surgical intervention was planned by the team of gynaecologists for the patient’s management, treating it as uterine fibroid. On exploration, a large right colonic mass was found rather than uterine fibroid and general surgeons were called to proceed the case.

Our team of general surgeons took over the care of the patient. Intra-operative findings revealed the mass to be firm and rounded but it was ruptured (Figure 1-a). It was originating from the colonic wall and was not attached to the mesocolon. There was 100ml pus in the peritoneal cavity. An open right hemi-colectomy with ileo-colic

anastomosis was done. The right hemicolectomy specimen was sent for pathological testing. Upon gross examination it measured 22.0x17.0x12.0cm in size. Separately present fatty tissue measured 12.0x6.0x3.5cm. The serosal surface of bowel was intact and unremarkable (Fig. 1-b). The mass was measuring 15x15x11cm, which was 19.0cm away from one resection margin and 6.0cm from the other resection margin (Fig. 1-c). Serial slicing of mass revealed firm homogenous tan with white gelatinous cut surface. On opening, the lumen was filled with faecal material. Mucosa was unremarkable. No ulcerations or areas of flattening were seen. Microscopic histological examination of the sections *(*Fig. 2*)***,** revealed the lesion to be composed of elongated fascicles of spindle cells with pale cytoplasm present in collagenous stroma. No nuclear hyperchromasia, pleomorphism or atypical mitosis was seen. Dense inflammation in the intestinal mucosa was also identified. All resections' margins were free of tumour. Five reactive lymph nodes were identified. The immuno-histochemical characteristics (with immuno-stains) showed that they were positive for Beta-catenin and negative for CD-117, DOG-1, SMA, Caldesmon, CD-34 and S-100.

The pathology report confirmed intra-abdominal fibromatosis. It was a well-capsulated form of fibromatous mass and showed no evidence of local invasion into other surrounding organs. However, the colonic location is very rare in this type of tumour. Hence, a second opinion was also requested to confirm the diagnosis which also showed the same conclusion. This patient had an uncomplicated recovery post-operatively and was discharged from the health-care facility on the 6th post-operative day. As advised by the multidisciplinary team, the patient has to get regular follow-ups and will be a part of life-long surgical surveillance as chances of recurrence are still there. Further, if facilities can be availed, genetic testing for FAP is recommended.

DISCUSSION

Fibroids arise from fibroblasts, which are located throughout the body. These cells have small and regular nuclei with rare atypical mitosis. They have a role in providing structural support, protection to the vital organs, and are critical for wound healing. The pathogenesis is unknown but various etiological factors may contribute in the genesis and growth of desmoid tumours, for instance pregnancy-induced hormonal changes, chromosomal abnormalities (trisomy 8 or 20), history of trauma or previous surgery[5,6]. FAP diseases are determined by genotype and phenotype factors.[6]Hence, having a family history or being a known case of FAP is at a higher risk of developing such conditions.

Clinical manifestation of these tumours is very vague and less decisive of the presence of disease. There is diagnostic difficulty due to unpredictable clinical behaviour, varied location and non-specific presentation. Patient may remain asymptomatic in the initial course of disease but later on, may present with dull abdominal pain with a feeling of mass. This maybe accompanied with pyrexia of unknown origin (PUO) and any bowl symptoms; where there is an invasion of the surrounding bowl as the infiltration of muscularis propria and submucosa is seen in some patients[7].

Our patient was having dull aching pain at lower abdomen with a feeling of lump in hypogastrium and right iliac fossa region, without having any bowl disturbances. Keeping in mind that this was a non-specific presentation, misdiagnosing the disease could happen and it may be taken as inflammatory fibroid polyps, fibrosarcoma or gastric lymphoma[4]. That is what happened, our patient was diagnosed as a case of uterine fibroid. Some clinicians have reported their cases of misdiagnosing desmoid tumour, mimicking other intra-abdominal tumours[7]. Due to this non-specific behaviour of disease, clinical findings are less supportive in making the diagnosis and therefore, histological analysis is necessary for disease confirmation. Immunohistochemistry is an instrumental investigation in remarkably differentiating the tumour's nature, especially where mixed-type lesions are suspected. In this patient, the histopathological marker Beta-catenin was positive. This could be because there is an absence of the APC gene in FAP which allows the beta-catenin to enter the nucleus and increases the cellular component. Hence, it functions as a reliable distinguishing marker from other similar types of lesions[8]. Another marker, RHAMM (Receptor for Hyaluronan Mediated Motility)protein which has recently been designated for cluster of differentiation 168(CD168), has also been found in higher expression in desmoid tumours, responsible for neoplastic transformation[9]. However, it was unremarkable in this case.

Nonetheless, apreoperative working diagnosisis mandatory to determine the most appropriate treatment for the individual patient; to monitor and modify ongoing treatment and to establish a correct diagnosis, imaging remains the mainstay of all preoperative investigations. The radiological scans and endoscopic biopsies; are pertinent for the surgical workup of the patient with baselines as well as hormonal profiling of oestrogen receptor, HER2 and progesterone receptor .Ultrasound guided biopsy could have been done for further assessment of the lesion, if the radiological and endoscopic biopsy results were contrastingly inconclusive.[10] Since the mass was a soft tissue lesion, an MRI could have been requested on urgent basis especially if there was a suspicion of extra-abdominal involvement.

Multi-disciplinary approach is required for successful treatment of mesenteric fibromatosis. Surgical excision with adequate margins is the mainstay of treatment[11].Margin clearance is of utmost importance in reducing the rate of local recurrence[4,11]. The report of one meta-analysis shows 2X increased recurrence rate, in patients treated with surgical resection alone and having positive margins[12]. In such patients, when tumour margins remain positive after excision, radiotherapy has a proven role in improved recurrence rate as the rate reduces to 20-40% when given any adjuvant radiotherapy,while as high as 40-70% with resection only. However, there is no proven benefit of radiotherapy once the margin clearance has been achieved[13]. Other than resection and radiotherapy, some clinical trials are given on use of cytotoxic chemotherapy and systemic pharmacological agents such as tamoxifen, doxorubicin and anti-inflammatory drugs which can be employed in cases where surgery and radiotherapy are not

rewarded by the desired success[14]. Despite all these treatment modalities, surgical gross total resection with a 3 cm safe margin (if it can be performed without significant disfigurement),remainsthe gold standard treatment for mesenteric fibromatosis[15].

CONCLUSION

Amongst the types of intra-abdominal aggressive fibromatosis, it is very rare for colonic involvement to occur. There is variable clinical presentation of the patient. Hence, confirmatory diagnosis can only be made with radiological imaging and histopathological biopsy reports. Just like other desmoid tumours, colonic fibromatosis remains a benign condition. However, due to the lesion's growth in size, it causes pressure symptoms but has no metastatic risk. The best intervention is surgical resection but that does not eliminate the significant risk of local recurrence. Where the growths have to be incompletely resected or the lesion's resection is contraindicated, targeted Radiotherapy is recommended. It can play a definitive role in controlling the disease extent. Unfortunately, in Pakistan there are not enough health centres with such facilities for patient management. The hospitals that have the set-ups, should work on forming a national networking registry and keep records, to study results in order to improve the patient prognosis in this region. Moreover, the government should invest in advancing awareness regarding such rare health conditions, so that patients consult health providers in early stages of their disease. This can only happen efficiently, if screening programs and a new parable of genetic testing is introduced in the country.

Conflicts of interest: None

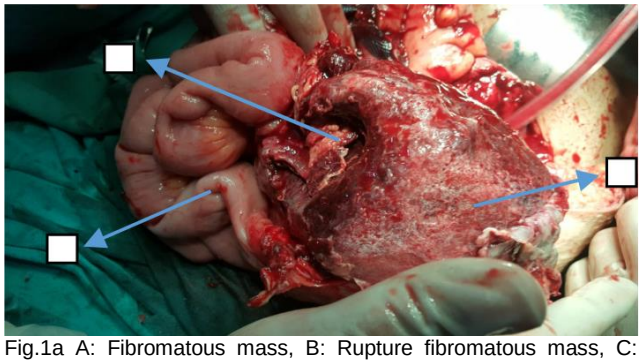

Fig.1a A: Fibromatous mass, B: Rupture fibromatous mass, C: ileum

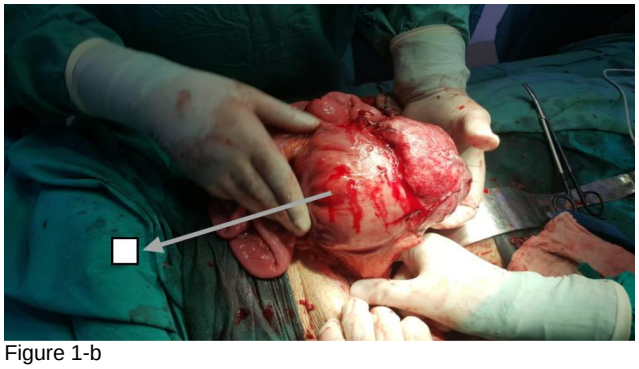

Figure 1-b

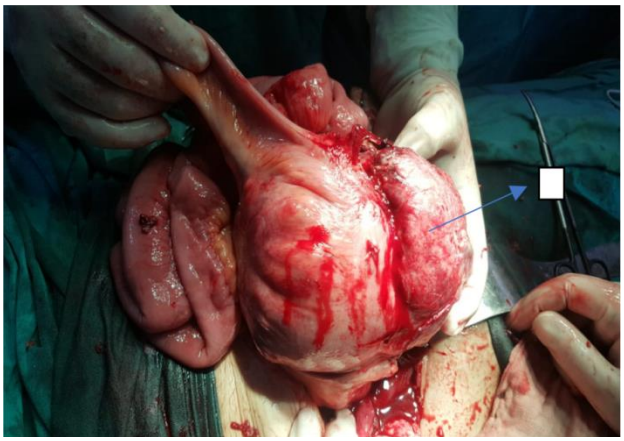

Figure 1-c

Fig. 2: Right Hemicolectomy's Specimen Size: 22.0x17.0x12.0cm , showing Spindle Cell LesionAll resections margins are free of tumor- Five Reactive lymph nodes identified

Authors contributions: MA:- intellectual genesis and case report writing, **SJ:** - literature review and case report writing, **MM:** - literature review and case report writing, **MKZ:** - case report editing, **UR:** case report editing, **HW** – Figures formatting **SM:** Figures formatting

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