VOLUME - 14, ISSUE - 05, MAY - 2025 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Obstetrics & Gynaecology

AN UNUSUAL PRESENTATION OF ANTERIOR ABDOMINAL WALL DESMOID FIBROMATOSIS IN A SCARRED UTERUS OF PREGNANT WOMAN-A RARE CASE REPORT AND REVIEW OF LITERATURE.

Dr Kalaivani Thirupathi — MSOG, Assistant Professor, Department of Obstetrics and Gynaecology, Sri Venkateshwara Medical college and Hospital and Research centre, Ariyur, Puducherry.

Dr Jayalakshmi Mega Nathan — MS OG,Assistant Professor, Department of Obstetrics and Gynaecology, Sri Venkateshwara Medical college and Hospital and Research centre, Ariyur, Puducherry.

ABSTRACT Desmoid tumor or Desmoid Fibromatosis(DF) is a rare benign soft tissue neoplasm arising from muscle aponeurosis characterised by monoclonal fibroblastic proliferation with variable presentation and unpredictable outcome may occur, when it is associated with trauma or pregnancy. It is a locally aggressive, infiltrative tumor with high risk for recurrence after resection but it lacks malignant potential and metastasis making treatment decisions more challenging. If Desmoid Fibromatosis occurs in pregnancy, optimal management should be considered for safe delivery, based on past history tumor size, location and behavioural pattern of tumor.We hereby report a case of 24 year old pregnant woman, presented with abdominal wall lump (4cm × 3cm) at 21 weeks of gestation with history of previous caesarean section. Observational treatment approach was feasible for this small tumor, for preserving function and anatomical integrity. Emergency Caesarean section was done at 34 weeks of gestation in view of impending scar dehiscence, concurrently radical resection of tumor also performed and uncomplicated follow up was reported without any recurrences. Also, literature review was discussed here focussing on marginal or radical resection of tumor, molecular genetics and available systemic therapeutic approaches.

KEYWORDS : Desmoid Fibromatosis(DF) , Abdominal wall tumor, Benign neoplasm, Desmoid tumor, Soft tissue tumor.

INTRODUCTION

Desmoid Fibromatosis is a rare soft tissue neoplasm, locally aggressive with high risk of recurrence after resection. It lacks malignant potential and does not metastasise. [1]As per WHO definition(2020), DF is characterised by clonal proliferation of fibroblasts in deep soft tissues, with infiltrative growth and tendency to have local recurrence without an ability to metastasise and it was classified under ICD D48.1.[1] The reported incidence of DF is 5-6 cases per 1 million population per year accounting for 0.03% of all neoplasms and <3% of all soft tissue tumors, with peak age at incidence is 30-40 years of age. Most common location of DF was abdominal wall(40%) as superficial and deep whereas extra abdominal constitutes 49% and intra abdominal constitutes 8% including retroperitoneal space. [1] The association of DF is 5% with Familial Adenomatous polyposis (FAP- Gardner syndrome). Most commonly it occurs in patients with history of abdominal trauma or surgery(open / laparoscopic/robotic).Extra abdominally, it can occur in neck, shoulder, chest wall, back, breast, Arm, thigh, Buttock and leg. Mac farland in 1832, first described these tumors, whereas Johannes Muller(1801-1858) etymological described the word "desmoid" which means band or tendon like, in his monograph on cancer. DF was commonly associated with FAP as described by Gardner, 1951.[1,2] Due to its unpredictable course of clinical features, Multidisciplinary management approach should be considered in all cases, which includes active surveillance, medical therapy , surgery and radiation. Surgical approach includes marginal resection or radical resection of tumor which aims to prevent recurrence rate in large tumors and for small tumors it should preserves function and maintain anatomical integrity.[1]

We hereby report a case of 24 year old pregnant woman, G2P1L1 with previous history of caesarean section ,who presented with abdominal lump of size 4cm× 3cm at 21 weeks of gestation. Moreover, an overview of literature was discussed here to outline the diagnosis and treatment challenges of Desmoid fibromatosis in pregnancy.

Case Report

A case of 24 year old pregnant woman with obstetric score of Gravida 2, Parity 1 and Live 1(G2P1L1), who had delivered by Caesarean section 24 months previously, came to our Antenatal OPD of our college at 21 weeks of gestation. She had a abdominal swelling of size 4cm ×3cm present over the left hypochondriac region, with complaints of vague pain over the swelling site. Initially, she noted a small swelling at 12 weeks of gestation, which gradually increased in size to attain the present size. Her past medical and surgical history was insignificant in relation to FAP associated polyposis, except short interpregnancy intervals. Her Antenatal Ultrasound at 21 weeks of gestation revealed a swelling of size 2cm ×2cm in the subcutaneous plane, with possible diagnosis of Abdominal wall endometriosis. She was advised to perform MRI abdomen and core needle biopsy for confirmation of diagnosis. Due to its affordability, she lost follow up. She came with complaints of scar tenderness at 34 weeks of gestation to our Obstetric casualty in good general health, oriented with Fundal height corresponds to 34weeks of gestation, fetal heart rate of 138 bpm, with 2 active contractions lasting for 40 sec and 2 cm dilatation. She was taken up for Emergency Caesarean section in view of impending scar dehiscence and Pfannensteil incision was preferred. Abdomen was opened by transverse Pfannensteil incision through skin, subcutaneous tissue and aponeurosis layers. In the deep subcutaneous plane, a firm mass of size 4cm ×3cm in diameter was found between the anterior and posterior peritoneal layers of rectus abdominis and the same was completely resected. LSCS was proceeded in usual way to deliver an alive preterm girl baby of birth weight 2.1 kg with Apgar score of 7/10, 9,10. Abdomen closed in layers after achieving haemostasis. Postoperative period was uneventful and patient was discharged on 5th Postoperative day with advise for regular follow-up. The Histopathological findings were consistent with intra operative findings, a firm mass of size 4cm ×3cm showing the presence of fibroblasts proliferation with sparse areas of spindle cells immersed in collagenous matrix. Immuno-histochemistry shows positive results for B- catenin, vimentin, Estrogen receptor beta and negative for desmin , S-100 and CD34 . The final diagnosis of Deep Desmoid fibromatosis was made. She was asymptomatic during her follow-up visit at 3 and 6 months postpartum. Colonoscopy was performed at 6 month of postpartum period to detect FAP associated

polyposis and results were negative. During her follow-up visit, clinical examination revealed a flaccid abdomen, no mass was found on superficial deep palpation. Also, Follow-up ultrasound was performed at the same time, negative findings were detected. Patient was followed up for 24 months after delivery, no recurrence found.

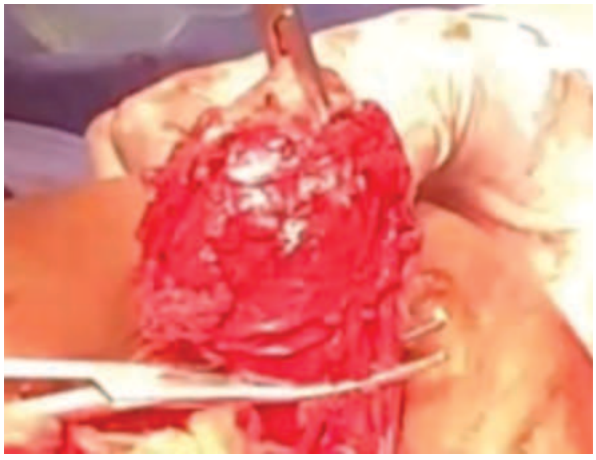

Figure 1. Intraoperative Findings of Desmoid Tumor During Caesarean Section.

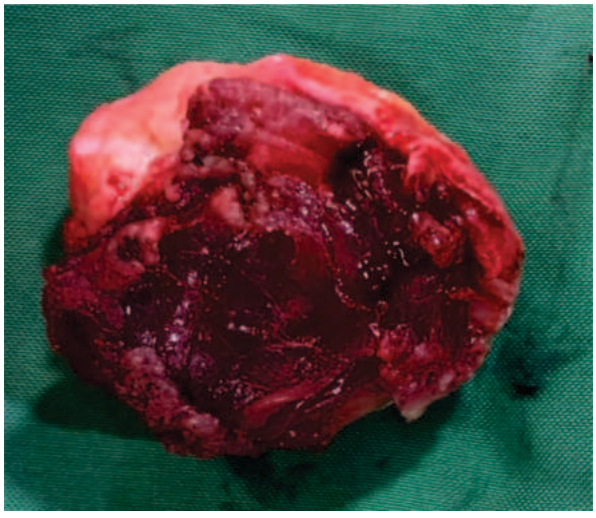

Figure 2. Macroscopic Appearance of Deep Desmoid Fibromatosis After Resection.

DISCUSSION

The abdominal wall Desmoid Fibromatosis occurrence in association with pregnancy is mainly due to trauma, excessive strain on rectus abdominis muscle and increased levels of growth factors and hormonal factors such as estrogen and progesterone.[1] The pathogenesis of DF involves monoclonal proliferation of myofibroblasts or fascia or aponeurosis and impairment of signalling pathways like Wnt/b- catenin or mutations in APC gene/ CTNNB1 genes which act as a key trigger desmoid development.[1,2] Most of the desmoid tumors are sporadic in nature (90%) and 5-10 % of tumors are associated with FAP(Gardner syndrome). Sporadic cases occurs most commonly in extra-abdominal sites, whereas FAP associated DF occurs in small bowel mesentery, abdominal wall predominantly in soft tissues. FAP associated DF often develops intra abdominally after abdominal surgery or trauma due to mutated APC genes.The reported complications of FAP associated DF are Bowel obstruction or Bowel ulceration and ureteral stenosis.[3]

During pregnancy, DF has high progression rate but carries good prognosis.[4] As per Speranzini et al, study report approximately 30-59% of cases with Desmoid tumor appeared to increase in size during pregnancy. In our study also, the Size of tumor was increased during consecutive weeks of pregnancy which was consistent with Vianna TSC et al. Study which reported 158% chance of increase in size of tumor during pregnancy.[5] The growth of the tumor during pregnancy was mainly influenced by distension of abdominal wall and aponeurosis, as well as by female sex hormones such as Estrogen and trauma caused by previous caesarean section. In recent times also, diagnosis is still challenging due to its rare occurrence, non specific symptoms and imaging modality such as Ultrasound can be considered for initial evaluation. Varying treatment modalities should be considered to achieve best possible outcomes including NSAIDS, Radiotherapy, Hormone therapy and Cytotoxic chemotherapy.[5] The standard treatment of choice was radical resection of tumor with a wide surgical margins, but in recent times, conservative approach should be opted for initial evaluation of asymptomatic cases, due to self limiting nature of desmoid growth. In asymptomatic cases, spontaneous regression of the tumor occurs in 25% of cases after childbirth.[5,6]

Microscopically, DF appearance resembles a firm,scar tissue with whitish grey in colour. Histologically it was characterised by proliferated spindle cells in a collagenous stroma, acapsular in vascular network without cellular atypia/ necrosis/ increased mitotic activity. Only the molecular profile was different between Sporadic and FAP associated DF. On Immunohistochemistry (IHC) , positive markers for DF are B-catenin, vimentin, smooth muscle actin, COX 2, PDGFRb, Estrogen receptor-b and negative for desmin, S-100, caldesmon, CD34. Recently, the expression of PD1/ PD-L1(programmed death-1/ programmed death ligand-1) immune checkpoints in correlation with CD4 and B catenin involved in development of DF and also it was related to prognosis.[7,8] The gold standard for diagnosis of Desmoid Fibromatosis is Histopathological examination whereas imaging modalities like Ultrasonography, MRI/ CT abdomen act as the basic tool for diagnosis as well as for minimal invasive interventions and monitoring tool for its effectiveness. [7]

The clinical manifestations of DF may vary from asymptomatic to impairing tumors with unpredictable growth and regression, based on its location, size and growth rate. Due to its biological behaviour of local infiltration and rapid progression it often mimics malignant tumor, which leads to impairment and death. Large Intraabdominal tumor of desmoid fibromatosis often leads to complications like obstruction of intestine, urinary tract, vessel obstruction/ perforation / bleeding or ischaemic tissue damage. Meanwhile, small DF may exhibit spontaneous regression, very rarely complete remission is possible.[8,9]

The outcome of DF depends on its anatomical site, proximity to vital organs, FAP association and its biological behavioural pattern, the treatment approach ought to be individualized to reduce the incidence of treatment failures, recurrence rate(25-60%) and to achieve possible quality of life.[10] Wide local excision is the ideal treatment of choice in small tumors. For large tumors, the multimodal treatment approaches like surgical resection, adjuvant radiation, systemic therapy and close monitoring should be considered, since it is associated with significant organ impairment, chronic non specific symptoms of disabling nature.[1,10]

As per Fiore et al.,multicentric study (2014) , 92 pregnant women with DF were analyzed for disease presentation during pregnancy. According to that, DF in association with pregnancy was treated by resection (52%), expectant management (43%), tumor progression (63%), medical treatment was given to only 4% of cases and 13% of cases relapsed after surgery. [12]After baby delivery, only 46% of DF cases undergone treatment, 54% of cases were expectantly managed and 17% of cases reported to be progressed after

treatment and 14% of cases spontaneously progressed. In subsequent pregnancies, 27% of cases progressed spontaneously. The reported Adverse Obstetric event was increased rate of Caesarean section. As per Hanna et al., study, rapid progression of tumor occurs in postpartum period which was consistent with reports of Debaudringhien et al study. [13]

Cates et al.,study suggested that surgical resection of DF during pregnancy does not increase the local recurrence risk. Though pregnancy is a major risk factor for growth and progression of DF due to elevated levels of Estrogen in pregnancy. But it has low impact on DF recurrence after surgical resection.[13] Although the occurrence of DF was raised in fertile age group women in recent years, pregnancy should not be discouraged. The systemic treatment options including NSAIDS, anti hormonal agents, Tyrosine kinase inhibitors, conventional chemotherapy and low dose chemotherapy regimens should be considered for large tumors. Moreover the management of DF in pregnancy should consider core needle biopsy to exclude malignancy for optimal individualised management.[14] Although, DF is a hormone dependent tumor as per immunohistochemical positivity for estrogen receptors, occurrence is more predominant in females of fertile age group, relapse of DF after pregnancy, shrinkage of tumor size after anti estrogen use in postpartum period.[15]

The standard treatment of choice is complete resection of tumor with negative margins, if this compromises functions, marginal resection of tumor with positive margins may be preferred. But similar recurrence rate may occur in both approaches, but higher rate was reported in large tumors than small tumors. During pregnancy, the recurrence rate after resection was reported as 13%. As per Drabbe et al., study reported that there was no statistical difference in 5 year progression free survival rate between microscopically positive and negative margins. [13] In our case , no recurrence was found even after 24 month follow up of the case.

CONCLUSION

Desmoid Fibromatosis is a rare disease, that usually occurs in women of reproductive age, especially during pregnancy. A high level of suspicion should be considered during pregnancy to avoid unnecessary surgical interventions. Adopting expectant management during pregnancy, improves the best possible quality of life. However , recurrence rate should also be considered during follow up.

REFERENCES

1. Zubor, P.; Henriksen, C.M.; Økstad, M.E.; Cerskuviene, E.; Visnovsky, J.; Kajo, K.; Valkov, A.; Lind, K.O. Desmoid Fibromatosis of the Anterior Abdominal Wall in Pregnancy: A Case Report and Review of the Literature. Diseases 2024, 12, 27.
2. R. Watanabe et al. Case Reports in Women's Health 46 (2025) e00701.
3. Bernd Kasper. The Desmoid Tumor Working Group / European Journal of Cancer 127 (2020) 96e107
4. Fletcher CDM, Bridge JA, Hogendoorn P, Mertens F. WHO classification of tumours of soft tissue and Bone (IARC WHO classification of tumours). 4th ed. 2013.
5. Viana TSC, et al. This article is distributed under the terms of the Creative Commons Attribution 4.0 International License.
6. Penel N, Coindre JM, Bonvalot S, et al. Management of desmoid tumours: a nationwide survey of labelled reference centre networks in France. Eur J Cancer 2016;58:90e6.
7. V. Ravi, Desmoid Tumors: Epidemiology, Molecular Pathogenesis, Clinical Presentation, and Diagnosis, Accessed November 15, 2023.
8. Sakorafas, G.H.; Nissotakis, C.; Peros, G. Abdominal desmoid tumors. Surg. Oncol. 2007, 16, 131–142.
9. Reitamo, J.J.; Häyry, P.; Nykyri, E.; Saxen, E. The desmoid tumor. I.: Incidence, sex-, age- and anatomical distribution in the Finnish population. Am. J. Clin. Pathol. 1982, 77, 665–673.
10. Müller, J. Handbuch der Physiologie des Menschen; Biodiversity Heritage Library: Washington, DC, USA, 1835; pp. 1833–1838.
11. Wanjeri, J.K.; Opeya, C.J. A massive abdominal wall desmoid tumor occurring in a laparotomy scar: A case report. World J. Surg. Oncol. 2011, 9, 35.
12. Fiore M, Coppola S, Cannell AJ, et al. Desmoid-type fibromatosis and pregnancy a multi-institutional analysis of recurrence and obstetric risk. Ann Surg. 2014; 259: 973-978.
13. C. D Drabbe, W.T.A. Van der Graaf, O. Husson, J.J. Bonenkamp, C. Verhoef, W. J. Van Houdt, Pregnancy-associated desmoid fibromatosis: a Dutch multi-centre retrospective study, Eur. J. Surg. Oncol. 49 (5) (2023) 921–927.
14. Skubitz KM. Biology and treatment of aggressive fibromatosis or desmoid tumor. Mayo Clin Proc. 2017; 92: 947-964.
15. Priolli DG, Martinez CAR, Mazzini DLS, et al. Desmoid tumor of the abdominal wall during pregnancy a case report. Rev Bras Ginecol Obstet. 2005; 27: 283-288.