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Case Report
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Fibrosarcomatous Transformation of Dermatofibrosarcoma Protuberans (DFSP) - An Aggressive Variant Presenting as Abdominal Mass

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Abstract

Dermatofibrosarcoma Protuberans (DFSP) is a slow growing mesenchymal tumour arising from dermal fibroblasts. It usually occurs in middle-aged population and starts as a firm plaque-like lesion and is most commonly seen in the trunk. When there is a fibromatous transformation within the DFSP, the lesion can rapidly increase in size and develop an increased metastatic potential. Treatment of choice for DFSP is surgical excision with negative margins. Adjuvant therapy is indicated based on the margin status and aggressiveness of the tumor. Here we present a patient with a large DFSP with fibromatous transformation which presented as a large abdominal wall mass with gross extension into the peritoneal cavity, which was successfully excised.

Keywords: Dermatofibrosarcoma protuberans; Abdominal mass; Fibromatous transformation; Excision

Introduction

Dermatofibrosarcoma Protuberans (DFSP) is a rare mesenchymal tumour. DFSPs are low grade tumors with low metastatic potential. In 5% to 20% of cases, DFSP have high grade fibrosarcomatous component and this transformation is responsible for high incidence of local relapse and distant metastases. Here we present a case of a giant DFSP of the abdominal wall which underwent rapid increase in size due to fibrosarcomatous transformation and was successfully excised.

Case Presentation

A 40 year old gentleman, otherwise fit and well, presented with a large swelling in the upper abdomen for about 5 years. The swelling was initially small and was slow growing up until a year ago when it rapidly increased in size to attain the present size. On examination, there was a large lobulated swelling in the midline measuring about 12 cm × 15 cm, extending from xiphisternum to umbilicus craniocaudally and up to the midclavicular lines on either sides laterally (Figure 1). The skin on the surface of the mass was ulcerated at a few areas.

Ultrasonography of the abdomen was suggestive of heterogeneously hypoechoic mass arising from the anterior abdominal wall with moderate vascularity and cystic spaces within it. In view of giant size of the mass, contrast enhanced CT scan of abdomen was performed to look for infiltration into deeper structures. CT suggested heterogeneously enhancing lobulated protuberant lesion of 12 cm × 15 cm × 13 cm, involving the skin, subcutaneous fat and musculature. The lesion was posteriorly protruding into the omentum, abutting the transverse colon and displacing the stomach posteriorly (Figure 2, 3). Multiple prominent internal mammary and epigastric vessels were seen traversing into the lesion. Biopsy of the lesion was suggestive of dermatofibrosarcoma.

He underwent wide local excision of the giant lesion with clear margins. In view of infiltration into the abdominal wall musculature, tumor was excised along with muscular wall resulting in entry into peritoneal cavity. There was no obvious infiltration of the lesion to the intra-abdominal organs. Primary closure of the abdominal wall was done. Small defect of skin at the epigastric region was left open for healing by secondary intention. Intraoperative period was uneventful. There was no significant blood loss. Postoperatively he made a smooth recovery and he was discharged on the 2nd postoperative day. Histopathological examination of the mass on cut section showed grey white, fleshy and nodular lesion with margin clearance of 1.1 cm (Figure 4). Microscopic examination showed neoplastic cells arranged in a storiform and herringbone pattern with hypocellular

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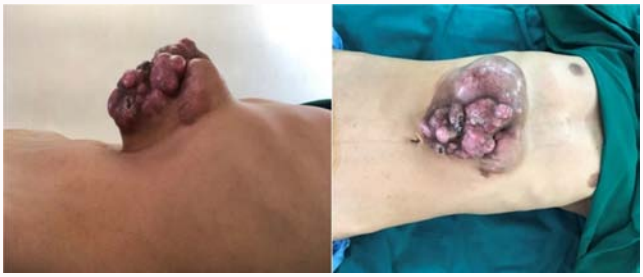

Figure 1: Large multi-lobulated swelling in the upper abdomen extending from xiphisternum to umbilicus.

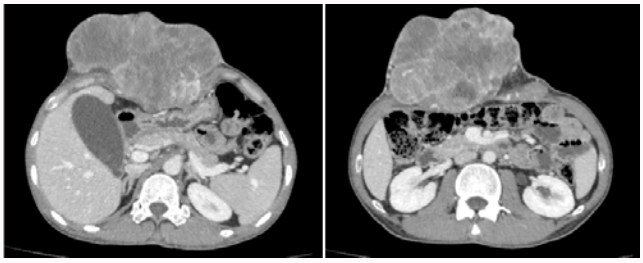

Figure 2: Axial section of CECT abdomen showing giant tumour involving the anterior abdominal wall musculature with abutment of stomach and transverse colon.

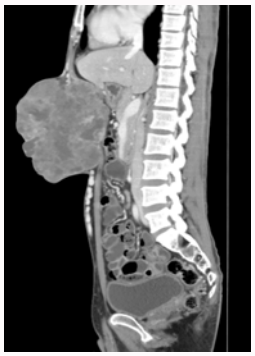

Figure 3: Sagittal section of CECT abdomen showing extension of mass from xiphisternum to the umbilicus and its heterogenous appearance.

area representing dermatofibrosarcoma and hypercellular area representing fibrosarcomatous transformation along with 5% to 10% necrotic areas. Spindle cells showing hyperchromatic nuclei were present and mitotic count was more than 30 per 10 high power fields. These features were suggestive of Grade II fibrosarcomatous transformation of DFSP. Medical oncology opinion was obtained and he was advised regular follow-up.

Discussion

Dermatofibrosarcoma Protuberans (DFSP) is a slow growing, low to intermediate grade soft tissue sarcoma. It was first described by Darier and Ferrandas "progressive and recurring dermatofibroma" in 1924 [1]. DFSP arises from the dermal fibroblasts and infiltrates into the deeper subcutaneous tissues. It constitutes 1% to 6% of all soft tissue sarcomas. It is more common in the middle age adults between 20 and 50 years of age with an almost equal incidence in both sexes. It occurs commonly in the trunk followed by proximal extremity and head and neck region [2].

DFSP develops as a firm plaque like lesion over the skin and

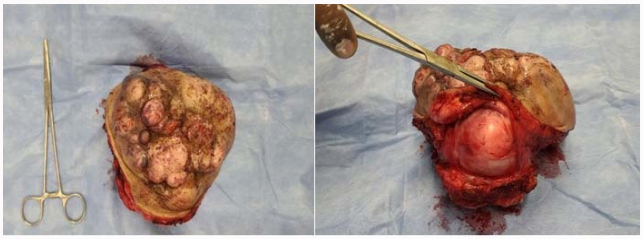

Figure 4: Specimen excised with clear margins.

characteristically grows slowly for several years and becomes protuberant with multiple nodules. Preoperatively, ultrasound scan, MRI or CT scan can be used to determine the nature of the lesion and its infiltration into the deeper tissues and surrounding vital structures. On ultrasound, DFSP appears as a well-defined hypoechoic mass. In some cases it can be mixed hypoechoic with high cellularity and hyperechoic with fibrous tissue along with tumour cells. DFSP is a moderately vascular lesion, mainly in the periphery which can be seen on color Doppler. MRI is more useful due to its high soft tissue contrast resolution. On T2 weighted images DFSP shows high signal intensity and on T1 weighted images it has similar intensity as skeletal muscle. On CT, DFSP appears isodense to the skeletal muscle and can be homogenous or heterogenous with non-enhancing areas of necrosis and cystic degeneration. CT is also useful in demonstrating involvement of underlying bones [3]. Histopathological examination of DFSP consists of uniform spindle shaped cells with elongated nuclei arranged in a storiform pattern. These cells have low mitotic index and infiltrate in to the subcutaneous tissue in a honeycomb appearance. DFSP has different histological variants, which include myxoid, pigmented (Bednar tumor), giant cell fibroblastoma, granular cell, sclerotic and Fibrosarcomatous (FS) components. Fibrosarcomatous transformation in a classical DFSP occurs in 5% to 20% of cases. It is an aggressive variant with high rate of local recurrence and high distant metastatic potential. Features suggesting fibrosarcomatous transformation within DFSP are at least 5% area of spindle cells showing mitotic index >7/10 high power field, high pleomorphism and herringbone growth pattern [4].

Genetically dermatofibrosarcoma is a result of translocation of chromosome t(17;22)(q22;q13) in 90% of cases, which leads to fusion of the genes COL1A1 (collagen type 1 alpha 1) on chromosome 17q21-22 and PDGFβ (Platelet Derived Growth Factor Beta) on chromosome 22q12. On immunohistochemistry classical DFSP is diffusely positive for CD34 staining in about 90% to 100% of cases, whereas in fibrosarcomatous DFSP the tumor cells lack CD34 positivity. CD34 staining is not a specific marker for dermatofibrosarcoma as it can be positive in benign neoplasms like solitary fibrous tumors and fibrohistiocytic tumors. DFSP can also express CD99 and SMA (smooth muscle antigen) but are usually negative for S100 [5].

The standard treatment for DFSP is wide local excision with 2 cm to 4 cm margin. In tumors where cosmetically and functionally sensitive areas, Mohs micrographic surgery is preferred to preserve the vital tissue and prevent the local recurrence. Fibrosarcomatous variant of DFSP can also have a comparable local recurrence rate as classical DFSP, if surgical excision is done with adequate margins. Adjuvant radiotherapy improves the Disease Free Survival (DFS), mainly in patients with close or positive margin conditions. Du et al. [8] in their study, on role of adjuvant radiotherapy found that 5-year

DFS was 88.1% for surgery and radiotherapy group compared to 56.2% for surgery alone group [6-8]. Systemic therapy with Imatinib mesylate, a tyrosine kinase inhibitor is beneficial in locally inoperable cases and metastatic DFSP. Imatinib acts by competitively binding to platelet-derived growth factor receptors on tumor cells and block their tyrosine kinase activity. Detection of chimeric gene COL1A1-PDGFβ presence in tumour cells prior to starting systemic therapy with Imatinib is advisable to know the benefit. The usual dose of Imatinib is 400 mg/day and in cases with poor response, the dose can be increased to 600 mg/day to 800 mg/day. Novel VEGFR (Vascular Endothelial Growth Factor Receptor) inhibitors such as Pazopanib, Sorafenib and Apatinib are on evaluation in controlling the Imatinib resistant cases [7].

The prognosis of DFSP is affected by the age, resection margin, lesion number, and histological subtype. Patients with age >50 years, resection margin <2 cm and fibrosarcomatous variant have poor prognosis. Ki-67, tumor proliferation activity indicator is another prognostic factor. Tumors with high Ki-67 shows poor prognosis [8]. Our patient presented with a slow growing anterior abdominal wall mass for 5 years with rapid growth in the last 1 year. Final histopathological examination after wide excision showed fibrosarcomatous variant of dermatofibrosarcoma with high mitotic count (>30 per 10 high power field). We believe that the rapid increase in size is due to the fibrosarcomatous transformation within the DFSP.

Conclusion

Dermatofibrosarcoma protuberance is a rare soft tissue sarcoma with low distant metastatic potential. Fibrosarcomatous transformation which occurs in 5% to 20% of DFSP is an aggressive variant with high recurrence rates and high metastatic potential. Surgical resection with wide margins is the treatment of choice. Adjuvant radiotherapy in cases of close margins will prevent local recurrence. Imatinib mesylate, a tyrosine kinase inhibitor has a role in inoperable and metastatic DFSP.

Authors' Contribution

JKAJ performed the surgery, reviewed and finalized the manuscript. NS and GL wrote the initial manuscript. PKR reviewed the manuscript and offered guidance. All authors contributed to the discussion. All authors have read and approved the final manuscript.

References

1. Darier J. Dermatofibromas progress if set recurrent or fibrosarcomas of the skin. Ann Dermatol Venereol. 1924;5:545-62.
2. Li Y, Liang J, Xu X, Jiang X, Wang C, Chen S, et al. Clinicopathological features of fibrosarcomatous dermatofibrosarcoma protuberans and the construction of a back-propagation neural network recognition model. Orphanet J Rare Dis. 2021;16(1):1-9.
3. Mujtaba B, Wang F, Taher A, Aslam R, Madewell JE, Spear R, et al. Dermatofibrosarcoma protuberans: Pathological and imaging review. Curr Probl Diagn Radiol. 2021;50(2):236-40.
4. Stacchiotti S, Astolfi A, Gronchi A, Fontana A, Pantaleo MA, Negri T, et al. Evolution of dermatofibrosarcoma protuberans to dfsp-derived fibrosarcoma: An event marked by epithelial–mesenchymal transition–like process and 22q loss. Mol Cancer Res. 2016;14(9):820-9.
5. Thway K, Noujaim J, Jones RL, Fisher C. Dermatofibrosarcoma protuberans: Pathology, genetics, and potential therapeutic strategies. Ann Diagn Pathol. 2016;25:64-71.
6. Abbott JJ, Oliveira AM, Nascimento AG. The prognostic significance of fibrosarcomatous transformation in dermatofibrosarcoma protuberans. Am J Surg Pathol. 2006;30(4):436-43.
7. Llombart B, Serra C, Requena C, Alsina M, Morgado-Carrasco D, Través V, et al. Guidelines for diagnosis and treatment of cutaneous sarcomas: Dermatofibrosarcoma protuberans. Actas Dermosifiliogr (Engl Ed). 2018;109(10):868-77.
8. Du K, Li J, Tang L, Lin X, Kong X, Liao X, et al. Role of postoperative radiotherapy in dermatofibrosarcoma protuberans: A propensity score-matched analysis. Radiat Oncol. 2019;14(1):1-8.