

CASE REPORT

Myxoid dermatofibrosarcoma protuberans of the vulva in a postnatal woman: a case report

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(Archana Nagendiram)**Abstract**

Background: Vulval Dermatofibrosarcoma protuberans (DFSP) is a rare cutaneous sarcoma that develops from the dermis, with less than 70 reported cases. This case discusses a holistic approach to vulval DFSP in a postpartum woman—with the balance of surgical excision against functional, psychosexual and cosmetic outcomes. **Case:** A 24-year-old woman noticed a rapidly enlarging vulval mass during pregnancy, which was an 8 cm left sided vulval mass which stretched from the periclitoral area to the inguinal crease. A core biopsy showed an indeterminate low grade myxoid lesion. Magnetic Resonance Imaging (MRI) reported no local metastasis or fascial involvement and Positron Emission Tomography (PET) and Computed Tomography (CT) scan showed metabolically active soft tissue mass without distant metastasis. Postpartum, she was referred to a Gynaecological Oncologist at a tertiary centre where it was discussed with Sarcoma and Plastic Surgeons. A radical local excision of vulvar mass, mesh reconstruction of rectus sheath and left inguinal ligaments and superficial circumflex iliac perforator flap of the left thigh was performed. Initial histopathology exhibited a myxoid lesion with negative staining including CD34, which is characteristic for DFSP. Due to indeterminate histopathology, RNA-sequencing was used and showed disease-defining chromosomal fusion (*COL1A1::PDGFB* fusion), which assisted the diagnosis, as this is present in over 90% of tumours. The tumour was excised over 10 mm laterally and 5 mm deep. **Conclusions:** Over 70% of DFSP is positive for CD34 and this shows the importance of RNA-sequencing in cases of indeterminate histology. Due to high recurrence, guidelines have suggested lateral margins from 10 mm up to 30 mm. Though, in a young woman this was considered against long-term sexual and functional outcomes. She will undergo close surveillance both clinically and with MRI.

Keywords

Dermatofibrosarcoma protuberans; DFSP; Postnatal; Sarcoma; Vulva; Myxoid

1. Introduction

Dermatofibrosarcoma protuberans (DFSP) is a rare cutaneous sarcoma arising from the dermis that has a rapidly progressive growth, high risk of recurrence but low risk of metastasis [1]. DFSP is an uncommon malignancy that most commonly occurs on the trunk, extremities, head and neck [2]. Female genital DFSP is infrequent with an incidence of 1.7% of all DFSP cases and overall, there are fewer than seventy reported cases of vulval DFSP [2–4]. This report describes a rare case of myxoid DFSP excised postnatally that grew rapidly throughout this patient's pregnancy.

This was an unusual case given the rarity of vulval DFSP, the distinctive subtype and histomorphology of the lesion which made the diagnosis challenging, especially with its presentation in pregnancy in a young patient. Given the atypical clinical and histopathological features in this case, the diagnosis was reliant on identification of a disease-defining chromosomal

fusion (*COL1A1::PDGFB* fusion). This case demonstrates the essential role of next generation sequencing and molecular diagnostics in routine surgical practice, particularly for rare lesions or those with atypical presentations. It also highlights the importance of considered investigation and management of groin lesions in a young woman where the risks of recurrence and tumour-related morbidity must be balanced against functional, psychosexual and cosmetic outcomes. Thus, demonstrating the importance of the multidisciplinary team in providing patient centred care.

2. Case presentation

A 24-year-old woman presented with a rapidly enlarging left groin lump causing increasing discomfort. This mass was first noticed at the beginning of her pregnancy, and it had rapidly grown to 8 cm in size at four weeks postpartum. There was no preceding trauma prior to the formation of the

mass. On clinical examination, there was a mobile and large left sided vulval mass which stretching from the periclitoral area to the inguinal crease. The patient was now gravida 2 para 2 following two vaginal deliveries and had no other significant gynaecological history. She had a body mass index of 30 and no other co-morbidities. Ultrasound guided core biopsy was performed at 39 weeks gestation. It demonstrated a heterogenous, vascular, subcutaneous lesion measuring $40 \times 17 \times 31$ mm that was relatively well defined with no communication with adjacent veins.

MRI Pelvis showed a well encapsulated mass without fascial involvement or any metastatic lesions. Histopathology of core biopsies from the lesion showed low grade myxoid lesion with weak non-specific staining for CD10 only, as further molecular investigations were not attempted on these biopsies. Further imaging with PET/CT scan showed Fludeoxyglucose (FDG) uptake in keeping with a moderately metabolically active soft tissue mass, without convincing evidence of nodal or distant spread (Fig. 1). The case was reviewed at the Bone and Soft Tissue Multidisciplinary Meeting at a tertiary hospital, which recommended resection and referral to Gynaecological Oncology. The decision was made to proceed to surgery after consultation between the Gynaecological Oncology team with the Sarcoma and Plastic Surgery teams.

At eight weeks postpartum, the patient underwent a radical local excision of the vulvar mass with a 10 mm clinical radial margin, mesh reconstruction of rectus sheath and left inguinal ligaments and local flap reconstruction with a superficial circumflex iliac perforator (SCIP) flap in a joint procedure with the Plastic Surgery team. Intraoperatively she was found to have an 8 cm left vulval mass which was adjacent to the clitoris and pubic symphysis superiorly, 1 cm adjacent to the border of the introitus and 1 cm medial to medial border of the adductor longus laterally. Surgical resection was completed to the superficial perineal fascia, and dissection proceeded to macroscopically uninvolved tissue. The SCIP flap was raised and rotated to reconstruct the defect, and she proceeded to have an uncomplicated post-operative recovery (Fig. 2).

Grossly, the lesion consisted of a firm, myxoid lesion centred within the subcutis, and was well circumscribed and well defined (Fig. 3). Microscopy there was a moderately cellular, spindle cell lesion with very even cellularity and a prominent myxoid stroma, with a rich, uniformly thin-walled vascular network and scattered intralesional polymorphs. The cells lacked significant atypia and there was some mitotic activity (up to 20 mitoses per 5 mm^2). Despite extensive sampling, the lesion showed no necrosis or frankly sarcomatous morphology. It was well defined, with focal areas of limited infiltrative growth into fat. It was clear of margins by approximately 5 mm. A broad immunohistochemical panel was performed, however the lesion was positive only for CD10, p53 wild-type and negative for CD34, estrogen receptor (ER), progesterone receptor (PR), SOX-10, S100, smooth muscle actin (SMA), desmin, myogenin, cytok Cytokeratin AE1/AE3 (CKAE1/AE3), beta-catenin, MUC4, STAT6, anaplastic lymphoma kinase D5F3 (ALKD5F3), tropomyosin receptor kinase (TRK), ROS1, CKIT, DOG1, EBERISH, PRKAR1A (retained/normal staining), calponin, myoD1, caldesmon, human herpes virus 8 (HHV8), epithelial membrane antigen (EMA),

CAM5.2, PAX-8, brachyury, glial fibrillary acidic protein (GFAP), MDM2 RNA-ish (Fig. 4).

The presence of a highly vascular, myxoid spindle cell lesion in the vulva initially prompted consideration of differentials including deep aggressive angiomyxoma, angiofibroma and myofibroblastoma, however the lesion lacked the thicker-walled blood vessels, variable cellularity, myoid elements or ER/PR/SMA/desmin or CD34 positivity. Given the undifferentiated nature of the lesion, and its presence in a sensitive site in a young patient, the tumour was referred for next generation sequencing using the 507-gene RNA fusion panel (Illumina, Inc, CA, USA) which revealed a *COL1A1::PDGFRB* fusion between exon 25 of *COL1A1* and exon 2 of *PDGFRB*, retaining the entire platelet-derived growth factor domain and leading to its upregulation (Fig. 5). This fusion is diagnostic of dermatofibrosarcoma protuberans per National Comprehensive Cancer Network (NCCN) guidelines for soft tissue sarcoma [5].

3. Discussion

3.1 Clinical presentation

In a review of sixty-nine cases of vulval DFSP most presented on the labia majora (52.2%) and then the mons pubis (11.6%) [4]. Most patients are asymptomatic but 7.2% may experience itch, pain, bleeding and dyspareunia [4]. Unlike other forms of DFSP, vulval DFSP often does not present as a plaque-like lesion but rather as a firm and subcutaneous mass [4, 6]. It is a lateral spreading mass that is mobile but fixed to the skin, that may have peripheral telangiectasias or small satellite nodules. The peripheral nodules fuse to form a bulkier mass which gives the eponymous “protuberant” appearance. Tumours may infiltrate vertically and become fixed to deep subcutaneous layer and fascia [6]. The rate of metastatic disease of DFSP is 1–5.7% which occurs through lymphatic spread, with most common sites including the lungs, regional lymph nodes and bones [1, 7]. Initially DFSP may be misidentified as a Bartholin’s or sebaceous cyst, soft tissue neoplasm or giant cell fibroblastoma. Other differentials to be considered should include nerve sheath tumours, myxoid liposarcoma, desmoplastic melanoma, leiomyoma, leiomyosarcoma or solitary fibrous tumour [1, 8].

3.2 Histopathology

Conventional DFSP is characterized by the cellular proliferation of intermediate sized, bland spindle cells which lack significant atypia or significant mitotic activity. These cells are arranged in a characteristic “storiform” or “cartwheel” pattern. Immunohistochemically, the defining characteristic is strong, diffuse CD34 positivity. It shows infiltrative growth through dermis and subcutis, resulting in a “honeycomb” appearance where it entraps fat lobules. Due to its infiltrative growth, it is typically poorly defined and can show deep, extensive infiltration well beyond the macroscopically or clinically appreciable bulk of the lesion [4, 9, 10]. Because of the challenges in delineating tumour margins, local recurrences are frequent after excision with “clear” margins [9, 10].

Myxoid DFSP is a recognised rare DFSP variant. It is commonly seen as a component within conventional DFSP,

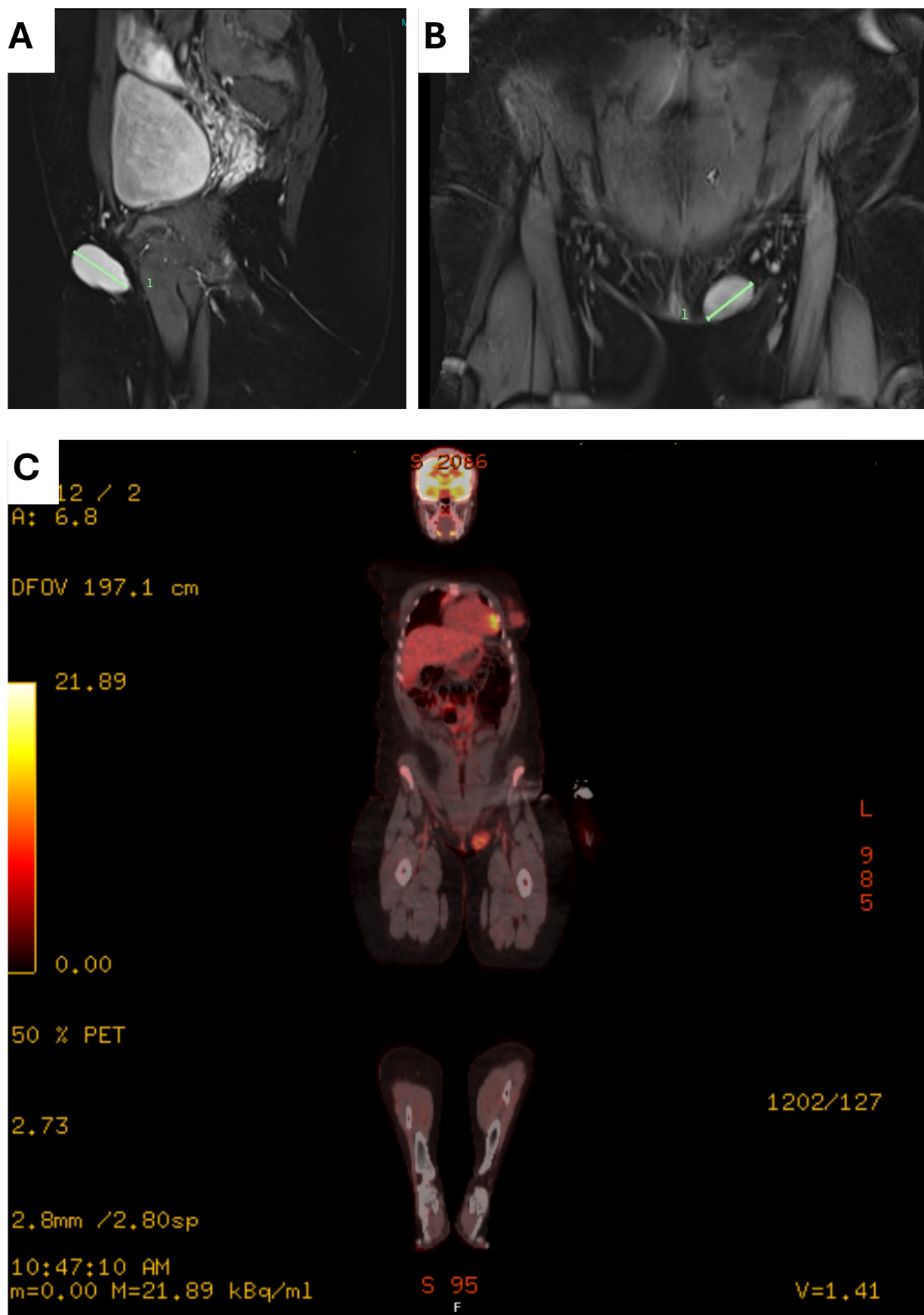


FIGURE 1. Imaging. (A) Sagittal MRI images that display that the tumour (marked in green) does not have fascial involvement. (B) Coronal images that display maximal tumour size (marked in green). (C) PET and CT scan showed metabolically active soft-tissue mass without distant metastasis. DFOV: Display field of view; PET: Position Emission Tomography.

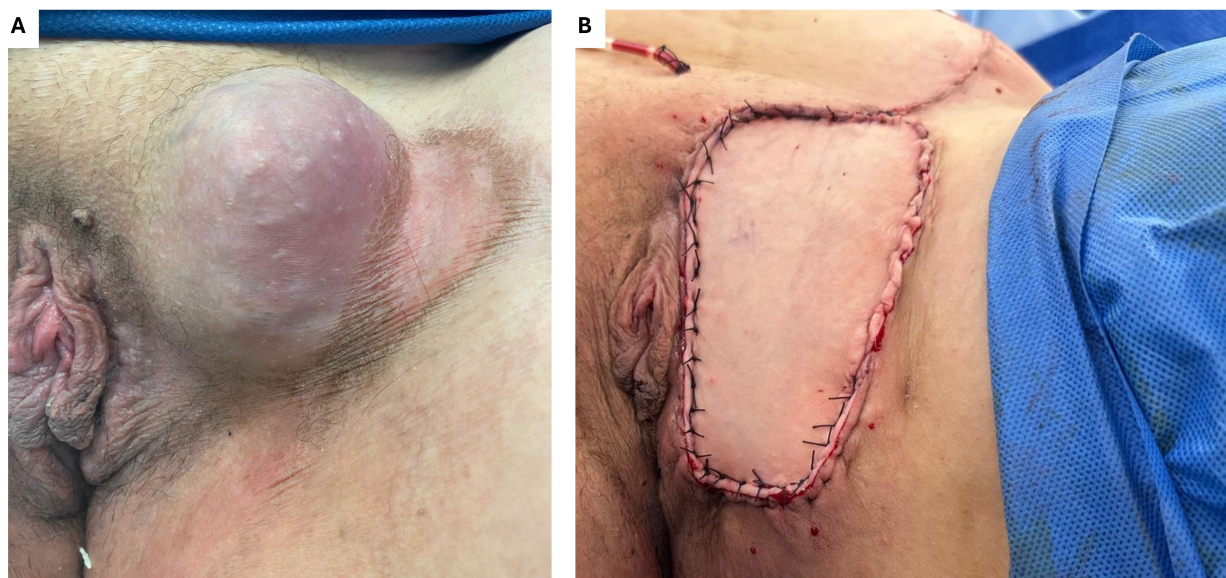


FIGURE 2. Intra-operative images. (A) Pre-operative image displaying the 8 cm left vulval mass adjacent to the clitoris and pubic symphysis superiorly. It was 1 cm adjacent to the border of the introitus and 1 cm medial to medial border of the adductor longus laterally. (B) Post-operative image after completion of radical local excision, mesh reconstruction of rectus sheath and left inguinal ligaments and local flap reconstruction with a superficial circumflex iliac perforator (SCIP) flap in a joint procedure with the Plastic Surgery team.

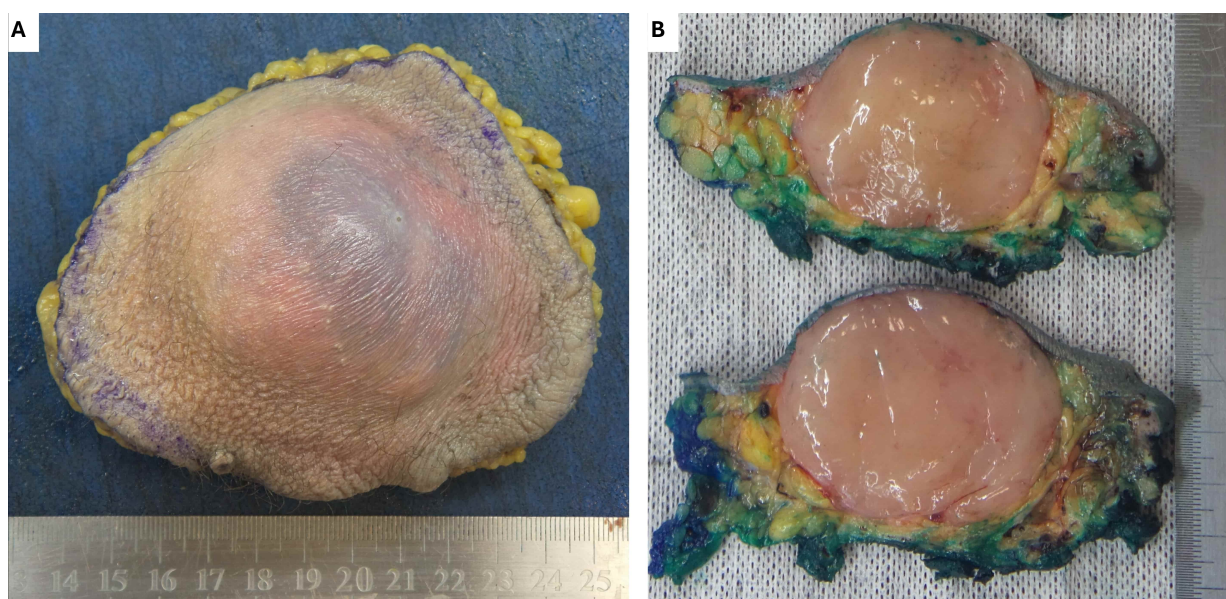


FIGURE 3. Macroscopic features. (A) Macroscopically the tumour consisted of an expansile, exophytic mass. (B) Its cut surface showed a well defined, glistening, “myxoid” lesion which lacked macroscopic evidence of infiltrative growth.

however no conventional component was seen in this case despite extensive tumour sampling. The presence of myxoid stroma was only described in 3 of the 69 vulval DFSP cases (4.3%) reported in literature, as per the MITO Rare Cancer Group [4]. Immunohistochemically, CD34 positive staining within the spindle cells is the most common identifier for DFSP [4]. However, the myxoid subtype is more likely to have patchy or absent CD34 expression as seen in this case, which makes the diagnosis particularly challenging on the basis of conventional histopathology [9, 10]. All histologic terms conform to WHO Classification (5th edn.).

3.3 Genetics

Over 90% of DFSP have a chromosomal translocation (17;22) (q22;q13), which causes subsequent fusion of the *collagen type 1 α 1* (*COL1A1*) and *platelet-derived growth factor β -chain* (*PDGFB*) genes [11]. The *COL1A1::PDGFB* fusion protein binds to the PDGF receptor which promotes continual activation of PDGF receptor β protein tyrosine kinase, resulting in dysregulated cell proliferation [12]. Thus, genetic testing via fluorescent in situ hybridisation (FISH) or RNA-Sequencing panel is a key diagnostic tool for DFSP in a tumour with suspicious histopathological features or immunohistochemical features. This unusual tumour evaded characterisation based

[illegible]

FIGURE 5. RNA Sequencing. 507-gene RNA fusion panel (Illumina, Inc, CA, USA) displaying *COL1A1::PDGFB* fusion between exon 25 of *COL1A1* and exon 2 of *PDGFRB* which was key in the diagnosis of DFSP.

on clinical, radiological and conventional histopathological findings, and because of its highly unusual morphology and immunoprofile, DFSP initially did not enter the differential diagnosis. This case demonstrates the value of next generation sequencing, a modality which canvases a wide range of diagnostically and clinically relevant gene regions in the diagnosis of undifferentiated lesion and tumours with unusual presentations.

3.4 Management

Complete surgical excision of tumour with clear margins is the gold standard for treatment of vulval DFSP. Lymphadenectomy has never been reported and is not recommended [4]. However, recommendations for surgical techniques and margins may vary. In this case, a wide local excision was performed to obtain clear surgical margins as the initial core biopsy was not diagnostic. The definition of suitable margins for DFSP remains controversial due to its high recurrence rate and the associated difficulty in histologically determining the peripheral boundaries of the tumour. The European consensus-based interdisciplinary guideline suggests that a 1–1.3 cm lateral margin is appropriate with micrographic techniques, but a lateral safety margin of 3 cm is generally advisable [13]. Whilst these margins may be appropriate in other anatomical sites, it is difficult to apply these requirements to vulval DFSP given the presence of sensitive adjacent structures and the limited soft tissue present in the vulva. For this patient, DFSP was retrospectively diagnosed after tumour excision and thus standard surgical margins were performed at the time of operation and dissection occurred to the rectus sheath. Additionally, to complete such broad surgical margins for a paraclitoral lesion in a 24-year-old woman who is sexually active could lead to negative outcomes for cosmetic and sexual function. In a tumour where DFSP is suspected, surgical planning could incorporate Mohs micrographic surgery (MMS) or complete circumferential and peripheral deep margin assessment (CCPDMA) which both utilise frozen section to provide confidence in complete excision [12]. However, these techniques rely on the involvement of a pathologist with experience with skin sarcoma to correctly identify clear surgical margins in these procedures, and the lack of established diagnosis meant that this was not possible in this case.

The plastic surgical team completed a mesh reconstruction of rectus sheath and left inguinal ligaments and SCIP flap from the left groin after the lesion was excised. A SCIP flap was chosen due to the proximity of the perforator adjacent to the defect, to allow the flap to be rotated or propellered. The donor tissue from the groin that provides adequate flap size, like with like reconstruction in terms of skin colour and quality with minimal scarring at the donor site. It encompasses the skin above the inguinal ligament [14]. Her post-operative recovery for the vulva and donor site was unremarkable.

There is no universal staging system for DFSP, though the NCCN states that stage I is local disease, stage II is regional disease, and stage III is distant disease [5]. After complete excision of vulval DFSP it is recommended that the patient undergoes close clinical surveillance, as mean recurrence time is 68 months [13]. Adjuvant chemotherapy or radiation therapy

is not recommended after complete excision [4]. Neoadjuvant radiation therapy may be recommended if histopathology has returned positive margins and repeat excision is not feasible [15]. In the MITO Rare Cancer Group analysis, rate of local recurrence was 42% with positive margins and 10.8% with negative margins [4]. The patient should be educated on the importance on self-examination and any concerns of recurrence require biopsy [16]. They should have clinical review every 6 months for 5 years and then annually until 10 years after excision. Imaging is not recommended routinely for surveillance unless previous recurrence or fibrosarcomatous changes on histopathology. This patient is to have 3 monthly surveillance indefinitely with MRI as per the Bone and Soft Tissue Multidisciplinary Meeting given its atypical presentation.

4. Conclusions

This case demonstrates the importance of timely investigation of vulval masses that appear in pregnancy and postpartum. It provides insight into myxoid DFSP which may not present with classical clinical, radiological or histopathological features of DFSP and shows the critical role of RNA-Sequencing in tumours with diagnostic molecular abnormalities, particularly those with atypical presentations. Finally, it reinforces the need of the multidisciplinary team including general Obstetrics and Gynaecology, Pathologists, Plastic Surgeons and Gynaecological Oncologists in the effective management of DFSP to ensure a positive operative outcome in balance with functional, psychosexual and cosmetic outcomes.

5. Limitations

The rare nature of vulval DFSP especially in postnatal women has resulted in a small amount of available published literature. This has led to limitations in data analysis and generalizability.

AVAILABILITY OF DATA AND MATERIALS

The data are contained within this article.

AUTHOR CONTRIBUTIONS

AN—completed the case report and literature review. JS—supervised data collection, analysis and was a major contributor in writing the manuscript. FW—provided clinical expertise in the review of DFSP pathology, contributed to manuscript preparation and developed figures including microscopic and macroscopic images. PD—provided clinical expertise in the review of DFSP pathology, contributed to manuscript preparation and developed figures for RNA fusion images. SL—contributed to the discussion of the plastic surgical reconstruction through surgical expertise and manuscript prescription. SP—supervised data collection, analysis and was a major contributor in writing the manuscript. All authors read and approved the final manuscript.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

The patient provided consent for the case report to be written and published. Formal ethics approval was sought through the Chris O'Brien Lifehouse Ethics Committee and was not deemed necessary for a case report, as patient permission had been obtained.

ACKNOWLEDGMENT

Not applicable.

FUNDING

This research received no external funding.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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How to cite this article: Archana Nagendiram, Jonathan Sandeford, Francesca Watts, Pranav Dorwal, Sarah Lonie, Selvan Pather. Myxoid dermatofibrosarcoma protuberans of the vulva in a postnatal woman: a case report. *European Journal of Gynaecological Oncology*. 2025; 46(9): 77-83. doi: 10.22514/ejgo.2025.123.