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Case report

An unusual case of dermatofibrosarcoma protuberans misdiagnosed as vascular malformation for over 30 years: A case report

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ABSTRACT

Introduction: Dermatofibrosarcoma protuberans (DFSP) is a rare and slowly growing soft tissue tumor and it is frequently misdiagnosed and mismanaged like more common masses. Therefore diagnostic delays are common and may result in challenging reconstructions.
Case presentation: We report the peculiar case of a 36-year-old patient with dermatofibrosarcoma protuberans of the right iliac fossa misdiagnosed as vascular malformation for over 30 years. Due to the delayed diagnosis resulting in a large tumor to be resected, surgical reconstruction was performed with a miniabdominoplasty approach with an excellent cosmetic and functional result.
Discussion: The review of the literature showed that mismanagements and delayed diagnosis of this sarcoma are frequent. Large skin and soft-tissue defects are frequently encountered in the surgical treatment of this tumor, and adequate knowledge of the reconstructive options is mandatory to provide the best possible outcome.
Conclusions: Superficial skin masses could be easily misdiagnosed. These diagnostic delays may lead to increased patient morbidity and more challenging reconstructive procedures. In this scenario, preoperative biopsies of suspicious lesions may be useful to avoid mismanagement of rare malignant neoplasms such as DFSP. In some challenging cases, the use of a surgical approach typical of cosmetic procedures may be useful to obtain satisfactory aesthetic and functional results.

1. Introduction

Dermatofibrosarcoma protuberans (DFSP) is an uncommon and slowly growing soft tissue tumor. Due to the rarity of this tumor and the lack of awareness among physicians, DFSP is frequently misdiagnosed and mismanaged like more common parietal wall swellings, such as lipoma, cysts, keloids, and fibromas [1]. Therefore, diagnostic delays are common with a median time from tumor onset to diagnosis of 3 to 5 years [2]. The proportion of pediatric DFSP cases reportedly ranges between 6 % and 20 %, even though DFSP cases in childhood are frequently missed, resulting in its discovery later in adulthood. The trunk is the most common anatomical site affected by DFSP (up to 70 %) [3].

We report this peculiar case of a 36-year-old patient with a misdiagnosed DFSP who had been present for over 30 years. This report would serve as an alert for those physicians who approach such slowly growing superficial lesions to avoid mismanagement of this rare malignant neoplasm. The present work has been reported in line with the SCARE criteria [4].

2. Case presentation

A 36-year-old female patient was referred to our outpatient clinic for a large skin mass on the right iliac fossa measuring 7 × 4 cm (Fig. 1). The lesion had been present for over 30 years, and it was presumed to be congenital, even though the medical records were missing and the patient was unable to precisely date the onset of the mass. In this period, the patient reported an extremely low growth rate of the lesion and she underwent several dermatologic consultations with a clinical diagnosis of tuberous angioma. The lesion was asymptomatic and the patient requested a plastic surgery consultation for cosmetic purposes only.

Physical examination revealed multiple brown-coloured, upraised, and hard nodules fixed to the dermis. The lump was mobile on the subcutaneous planes, without attachments to deeper structures. At first, the tumor was excised with a 0.5 cm margin as a hemangioma; a contralateral skin symmetrization like a miniabdominoplasty was

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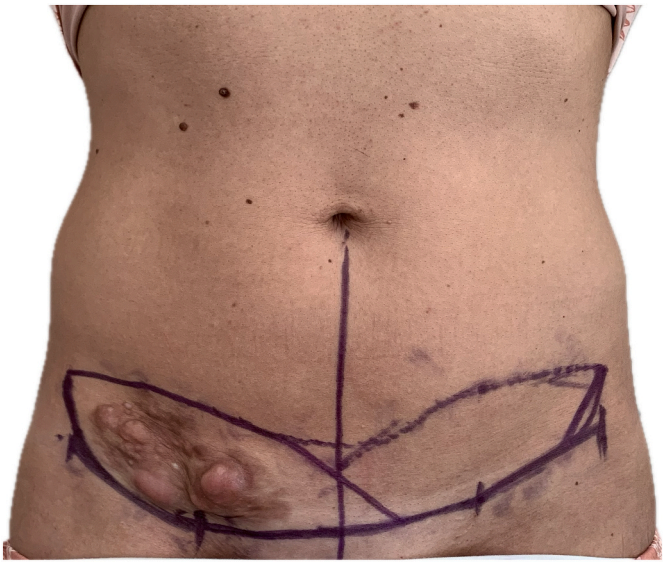

Fig. 1. Preoperative picture showing a large lobulated lesion in the right iliac fossa measuring 7 × 4 cm with preoperative markings of the miniabdominoplasty.

performed (Fig. 1).

Histological examination revealed an ill-defined and diffusely infiltrative tumor of the dermis and subcutis The tumor was composed of a uniform population of slender spindle cells arranged in a monotonous storiform pattern. The tumor showed a characteristic pattern of invasion of subcutaneous tissue leaving behind intact adipocytes thereby creating a honeycomb appearance. The mitotic activity was low (<5 mitosis/10 HPF) (Fig. 2A-B).

Immunohistochemical analyses confirmed the diagnosis by revealing a strong CD-34 positive staining with negative S100, Caldesmon, Desmin, EMA, and muscle-specific actin stains, which were consistent with the diagnosis of DFSP. After a multidisciplinary tumor board consultation, a second extended resection with adequate surgical margins (3 cm) was performed (Fig. 3), and wound closure was obtained by advancement of the cutaneous flaps from the abdomen and superior thigh region. In order to prevent scar diastasis or migration due to the excessive tension resulting from the abundant skin removal in a skinny patient, the surgical margins were strictly anchored with PDS 2/0 to the underlying muscular fascia at the level of the inguinal crease.

Staging whole-body computed tomography (CT) scan and bone scintigraphy showed no evidence of distant metastases. The one-year follow-up visit showed a fully satisfactory cosmetic and functional result. The scars were fully concealed in the inguinal crease and good symmetry with the contralateral side was reached (Fig. 4). The patient continues to be in follow-up without any recurrence.

3. Discussion

The rarity and heterogeneous clinical presentation of DFSP can confound its identification leading to mismanagement and delayed diagnosis. Despite the atrophic DFSP is one of the most rare and neglected histopathological variants of DFSP [5], even the classic nodular subtype could be misdiagnosed with a median total time from the tumor onset to diagnosis of 4 years. Interestingly, David et al. reported that potential clinical misdiagnoses are more frequently rendered by primary care clinicians (75 %) and dermatologists (33 %), even though misdiagnoses could be reported from more than one type of physician [2].

DFSP in children is reported very rarely and is commonly misdiagnosed as vascular malformation or other benign lesions such as cysts, lipomas, keloids, or fibromas [3,6]. Furthermore, most patients

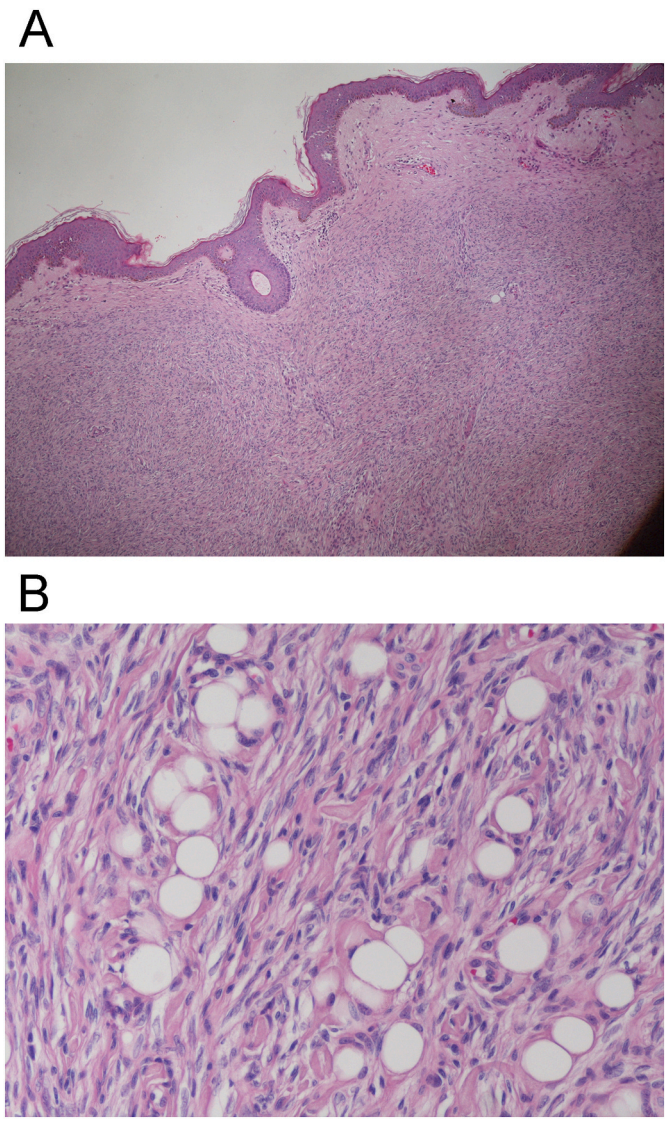

Fig. 2. (A) Histological findings showing a diffused dermal infiltration characterized by a population of monotonous spindle cells, Hematoxylin-eosin (HE) × 10; (B) The tumor diffusely infiltrates subcutaneous adipose tissue giving rise to the characteristic honeycomb pattern, HE × 10.

who have DFSP seek medical advice in the later stages of the disease as it is asymptomatic in the early stages and is a slow-growing tumor [1].

We report this almost unique case of a DFSP which had been present for over 30 years. In our opinion, this case faces all the challenging aspects of the diagnosis of DFSP. First, the onset of the lesion probably dates back to childhood or was even congenital. At this time, the patient referred several dermatologist consultations with a clinical diagnosis of tuberous angioma. Second, as it was an extremely slow-growing lesion, the patient neglected the presence of the tumor and did not seek medical advice for a long period, thus resulting in a large mass to be resected. Third, the surgical treatment of DFSP could be challenging from a reconstructive perspective. Complete surgical resection is the primary treatment method for DFSP; at the same time, functional and cosmetic preservation is mandatory, especially in young patients. As wide local excision with surgical margins ranging from 2 cm to 3 cm is recommended due to the high risk of recurrence, large skin and soft-tissue defects are frequently encountered thus requiring skin grafting or local/free flaps in some cases [3]. Where possible, the primary closure is the first choice. We faced this peculiar case by using a cosmetic procedure to achieve a post-oncologic reconstruction with excellent

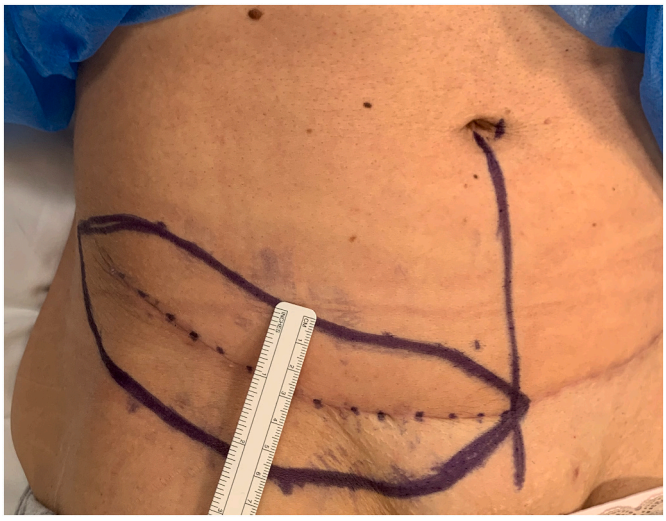

Fig. 3. Planning of the second extended resection with adequate surgical margins (3 cm).

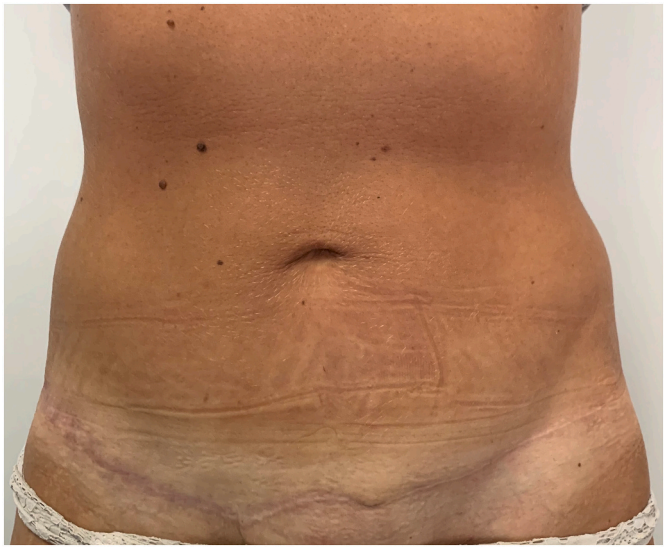

Fig. 4. One-year follow-up visit showing a fully satisfactory cosmetic and functional result.

cosmetic and functional outcomes.

Tumor excision through the miniabdominoplasty approach represents a useful option for the management of the lower abdominal wall masses and it offers a reliable alternative to achieve satisfactory cosmetic results [7]. The risk of complications associated with full-abdominoplasty is reduced as the dissection is limited to below the umbilicus. Furthermore, several complications related to the primary closure of large skin defects (i.e. wound dehiscence, hematoma, seroma, pathological scarring, cosmetic deformities) or flap surgery may be avoided [8].

Despite a large amount of skin and soft tissues resected in a skinny patient, we obtained a primary closure of the skin defect and abdominal contouring of the contralateral side through a miniabdominoplasty access, thus avoiding asymmetry of the abdominal wall or dog-ear deformities. Finally, given that DFPS lesions are mainly located on the trunk, miniabdominoplasty should be considered a valuable tool in the reconstructive armamentarium of this malignant tumor.

The need for a preoperative biopsy of these superficial skin masses also represents another issue that should be discussed. The usefulness of a biopsy in soft tissue sarcomas is indisputable especially when tumors are suspected to involve structures at or below the investing fascia. In such cases, if lesions are not adequately imaged o biopsied before excision they could be incompletely excised. Therefore, re-excision of the tumor bed is required, often resulting in a greater resection of tissue than would have been necessary had the tumor been adequately treated initially. If radiotherapy is subsequently required, then this also necessitates wider field coverage [9]. There is also the theoretical risk of inadequate re-excision of the tumor bed due to a scar distortion of the surrounding tissues. This likely results in increased morbidity for the patient. Conversely, primary excision of superficial lesions may represent a safe procedure without an increased risk of patient morbidity as the re-excision of a superficial lesion above the fascia could be easily performed without concerns about oncological radicality. In our case, the lump was completely mobile on the subcutaneous planes, without palpable attachments to deeper structures and a primary excision seemed a feasible option. Moreover, the clinical information provided by biopsy may not always be sufficient for an adequate diagnosis. Biopsy for the diagnosis of DFSP should include the subcutaneous layer as this sarcoma typically infiltrates subcutaneous tissue. If a biopsy is performed superficially or the specimen size is inappropriately small, histological diagnosis may remain challenging. Nevertheless, we believe that preoperative biopsy of such lesions is strongly recommended for the appropriate management of these lesions.

4. Conclusions

DFSP could be easily mismanaged and the differential diagnosis is challenging. A significant delay in diagnosis is more common in pre-adolescents and may lead to the development of large skin masses. Consequently, large skin defects resulting from wide local excision of misdiagnosed DFSP may be theoretically frequent and they represent a reconstructive endeavor. In our opinion, the use of miniabdominoplasty provides excellent outcomes and avoids functional and cosmetic deformities, particularly in young patients.

An increased awareness among primary care clinicians and dermatologists of the need for a prompt and early diagnosis of this malignant tumor remains mandatory. In this scenario, preoperative biopsies of these lesions are recommended to avoid dangerous diagnostic delays and to determine the most appropriate therapeutic strategy.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Ethical approval

Observational case studies are exempted from Ethical approval in our institution.

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Author contribution

Dr. Matteo Torresetti performed the surgical procedure, led the study design, data collection and interpretation and wrote the paper.

Dr. Donatella Brancorsini and Prof. Gaia Goteri performed histological evaluation and participated to the writing of the manuscript.

Prof. Giovanni Di Benedetto participated to study design, approved and drafted the final manuscript.

Guarantor

Dr. Matteo Torresetti.
Dr. Donatella Brancorsini.
Prof. Giovanni Di Benedetto.

Research registration number

N/A.

Conflict of interest statement

The authors declare that there is no conflict of interest.

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