Radiology Case Reports 20 (2025) 2604–2608

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Darier–Ferrand dermatofibrosarcoma protuberans of the right supraclavicular region in a child: A case report☆,☆☆

*Oukassem siham**, *Abourak Chaimae, Asmae Guennouni, Belkouchi Lina, Allali Nazik, Chat Latifa, El Haddad Siham*

Department of Radiology, Mother-Child, Faculty of Medicine and Pharmacy of Rabat, Children's Hospital, Ibn Sina University Hospital, Mohammed V University, Rabat, Morocco

ARTICLE INFO

Article history:
Received 2 December 2024
Revised 5 January 2025
Accepted 9 January 2025

Keywords:
Dermatofibrosarcoma protuberans
MRI
Recurrence

ABSTRACT

Dermatofibrosarcoma protuberans is a rare, locally aggressive sarcoma of the skin in children and adults, usually involving the trunk and extremities and less commonly the head and neck. Despite clinical reports in the literature on the management of dermatofibrosarcoma protuberans, there are limited articles describing its imaging features, we report a case of a child presenting with a swelling above the right clavicular for the past year. An MRI was performed, revealing a mass above the clavicle initially suggestive a vascular tumor. A surgical excision was conducted, identifying a dermatofibrosarcoma protuberans.

Introduction

Dermatofibrosarcoma protuberans (DFSP) is a fibrohistiocytic tumor of intermediate malignancy that is very rare in childhood. It accounts for approximately 1.0% of all soft tissue sarcomas and less than 0.1% of all malignant tumors globally. Although it is uncommon, DFSP is the most common sarcoma originating in the skin. This tumor predominantly occurs on the trunk and extreme [1]. Children and adults with DFSP have similar clinical presentations, histopathologic features, and molecular traits. However, a clinical diagnosis in children is more challenging and necessitates a high level of suspicion. Due to the tumor's rarity and lack of distinguishing characteristics, diagnosis is frequently delayed. To lower the chance of recurrence, the tumor must be completely surgically removed.

Case presentation

An 8-year-old boy presented with a weeping nodular mass in the right supraclavicular region, evolving over the past 9 months. The mass had progressively increased in size and had become painful. On clinical examination, a palpable mass measuring 4 cm × 3 cm was observed in the supraclavicular area, with a violaceous, multinodular, and ulcerated appear-

☆ Competing Interests: All authors declare no conflict of interest.
☆☆ Acknowledgments: No funding was given.
* Corresponding author.
E-mail address: siham.oukassem1995@gmail.com (O. siham).
https://doi.org/10.1016/j.radcr.2025.01.045

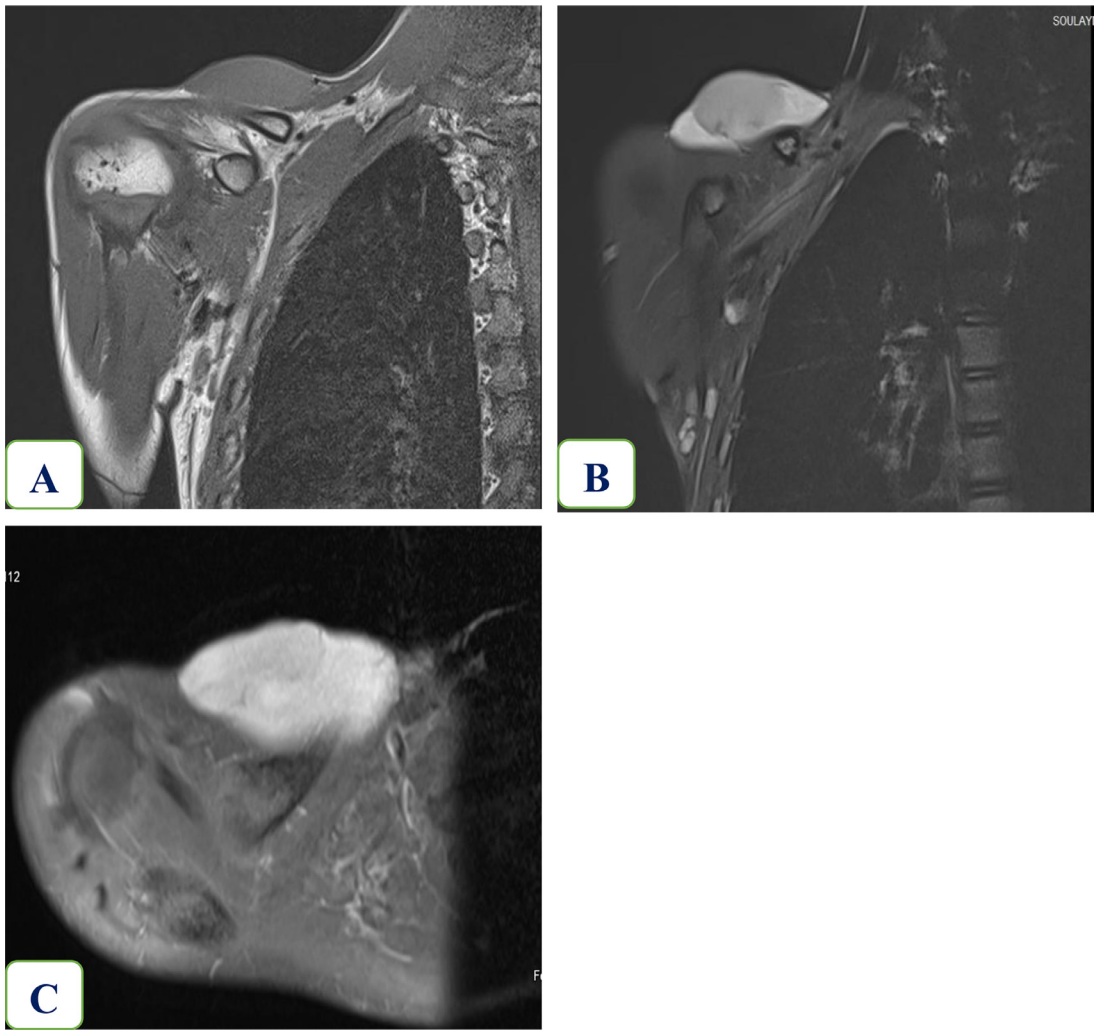

Fig. 1 – Coronal T1-weighted spin-echo image (A) and coronal DP FS (B) and axial section T1 post Gado (C)shows a subcutaneous mass above the clavicule, oval in shape, well-defined with regular borders, showing hypointensity on T1-weighted imaging, heterogeneous hypersignal on T2-weighted imaging. Post-contrast injection, the mass displayed homogeneous and intense enhancement (C).

ance. It was non-mobile, with no redness or warmth. Systemic examination revealed no abnormalities. The lymph nodes in the head and neck region and the axillae were nonpalpable. The patient does not have diabetes mellitus, asthma, tuberculosis (TB), hypertension, or any other known diseases.

The USG local region revealed a well-defined hypo-echoic lesion of size 4cm x 2 cm x 3 cm in the subcutaneous area in the right clavicular region. It showed posterior acoustic enhancement and multiple septa within. Color Doppler imaging revealed a significant vascularity. The patient was provisionally diagnosed with hemangioma, an additional MRI of the left shoulder revealed a subcutaneous mass above the right supraclavicular area, oval in shape, well-defined with regular borders, showing hypointensity on T1-weighted imaging, heterogeneous hypersignal on T2-weighted imaging, containing areas of signal void on susceptibility-weighted sequences. Post-contrast injection, the mass displayed homogeneous and intense enhancement. It is in contact with the deltoid muscle without invading it, maintaining respect for the clavicle and the surrounding soft tissues (Fig. 1).

Our patient underwent a surgical excision of the mass with a 2 cm clear margin. Histopathological analysis showed myxoid differentiation and spindle cell proliferation with a storiform pattern that extended past the surgical margins. A monomorphic, ovoid to elongated nucleus with variable low mitotic activity was seen in tumor cells, which also displayed eosinophilic and abundant cytoplasm (Fig. 2). Histological features suggestive of dermatofibrosarcoma protuberans were observed.

In our patient, the resection margins were clear in the initial excision. However, after 2 years, the patient returned with complaints of slight induration at the scar site, which had become painful, the clinician noted an indurated, erythematous plaque at the site of the scar. A follow-up MRI was per-

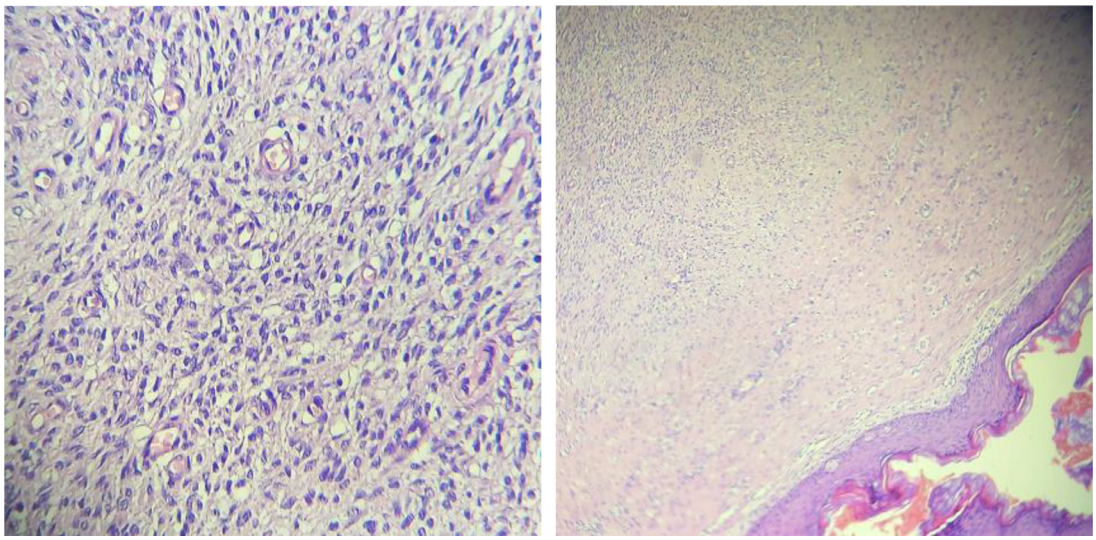

Fig. 2 – The cutaneous tissue is lined by a regular epidermis. The underlying dermis and hypodermis are the site of a densely packed spindle-cell tumor proliferation with a storiform architecture. It consists of slightly atypical, monomorphic spindle cells with oval nuclei and fine chromatin, exhibiting low mitotic activity.

formed, revealing an infiltration of subcutaneous fat at the scar site, appearing as a high signal on DP Fat SAT sequences, enhanced after contrast injection, suggestive of a recurrence (Fig. 3). Following a multidisciplinary consultation, the patient underwent a wide resection with a 5 cm safety margin. The patient underwent a wide resection with skin reconstruction. The surgical specimen was sent for pathological examination, which confirmed a recurrence, with clear resection margins. The patient progressed well over the following year with no signs of recurrence.

Discussion

Dermatofibrosarcoma protuberans (DFSP), a rare type of malignant skin tumor arising from fibroblasts. It comprises less than 0.1 percent of all malignant tumors and about one percent of soft tissue sarcomas [1]. DFSP most commonly occurs in early to middle adult life between 20 and 50 years of age, although its range of occurrence is from birth through the 80s. The tumor is relatively rare in children; 27 cases have been reported in persons younger than 16 years of age. DFSP at birth has been documented in five patients. The literature reveals an almost equal sex distribution with a slight male predominance in larger series [2].

According to most studies, about half of lesions present on the trunk, making it the most common site for DFSP. Though congenital forms may mimic the adult pattern and appear on the trunk and proximal limbs, the tumor typically manifests in pediatric patients on the legs and acral regions [3].

The precise cause of DFSP is not entirely known. There have been suggestions of prior trauma to the affected area, but this is probably coincidental. In our patient has never reported any history of trauma in the right supraclavicular region. The pathophysiology of DFSP is significantly influenced by genetic factors. About 90 % of cases have the chromosomal translocation t(17;22) (q22;q13), resulting in the formation of the COL1A1-PDGFB fusion gene, which drives tumor growth [4,5]. Fluorescence in situ hybridization (FISH) can identify this molecular abnormality, which can help with diagnosis and, in some situations, validate the use of targeted molecular therapies [6].

Depending on the developmental stage, the clinical presentation in children is comparable to that in adults. Deep-seated nodules are less common in the early stages, when lesions primarily take the form of single papules or plaques [7]. Induration is one of this tumor's most dependable characteristics. Until the disease reaches a late stage, when the underlying structures are invaded, the lesion typically travels freely over deep tissue structures [8]. The skin on top is colored fleshy, violaceous, brownish, or erythematous. A bluish tint could be the tumor's initial symptom, which could result in a false initial diagnosis of a vascular lesion [9]. Lesions typically measure 1 to 5 cm at diagnosis and are asymptomatic. These lesions typically get bigger over time.

Regardless of how DFSP initially manifests, it eventually begins to grow quickly with a nodule or nodules on its surface, moving on to the multi-nodular protuberant phase (which may occur even after 60 years). At this stage, pain, ulceration, and bleeding could make the tumor more complicated [10]. In our patient, the mass gradually increased over a period of 9 months and became ulcerative. Initially, it was a small nodule, which later developed into a multinodular mass.

The difficulty of diagnosing this tumor during the non protuberans phase should be emphasized. Because the tumor is so uncommon in children, the diagnosis might not be made until adulthood, when the lesion will have its distinctive bulging shape and begin to proliferate as usual [11].Although some children are not diagnosed until 14 or 15 years have passed, in certain pediatric series, the average time to reach a definitive diagnosis is 5 years [11].

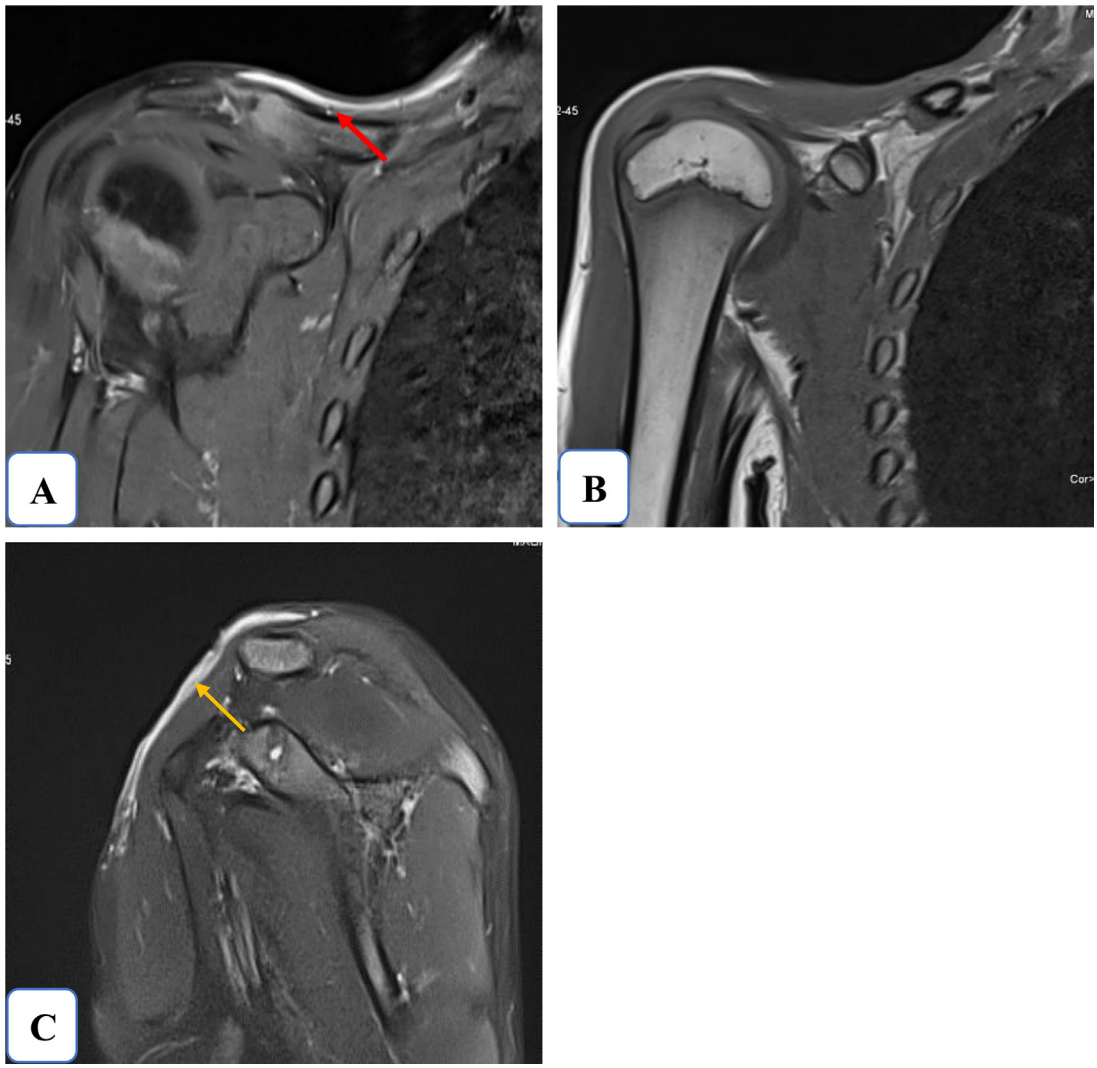

Fig. 3 – MRI sections: coronal DP fs (A), coronal T1 (B), Sagital T1 post gado (C) shows infiltration of subcutaneous fat at the scar site, appearing as a high signal on DP Fat SAT sequences (Red arrow), enhanced after contrast injection (yellow arrow), suggestive of a recurrence.

The initial size of the lesion at the early stage is 2-5 cm. However, this remains undiagnosed, since the patient doesn't ask for medical advice, so the lesion may reach even 25cm in diameter or larger. The tumor is initially firmly fixed to the overlying skin but not to underlying deeper structures, except for those lesions involving the scalp, where periosteal attachment occurs very early [4].

DFSP is often mistaken for malformations or vascular tumors, infantile fibromatosis or myofibromatosis, fibrosarcoma or fibrous hamartoma, aplasia cutis, subcutaneous fat necrosis, arthropod bites, or intrauterine trauma and hemangiomas (particularly in children) [12]. As in the case of our patient, the tumor was initially mistaken for a vascular tumor, and it was only after surgical resection that the diagnosis of DFSP was confirmed." Radiologic studies can be useful in these patients, but the gold standard for diagnosis is skin biopsy and immunohistochemical and molecular studies.

DFSP typically manifests macroscopically as a solid, fish-flesh-like mass in soft tissue that is poorly defined and white to yellow in color. Larger tumors may exhibit cystic changes or bleeding patches. According to histology, fibroblasts in the dermis or subcutaneous tissue are the source of DFSP. The upper dermis of DFSP has loosely arranged spindle cells in its early stages, which change into monomorphic spindle cells in a storiform pattern as the condition advances. The spindle cells typically exhibit CD34 positivity by immunohistochemical staining, whereas other markers, including protein S100, Factor XIIIa, alpha-smooth muscle actin, and melanA, are negative. Myxoid, pigmented, giant cell, giant cell fibroblastoma, granular cell, sclerotic, and fibrosarcomatous (FS) variants are among the histologic subtypes of DFSP [13,14].

Magnetic Resonance Imaging (MRI) is recommended for several indications in the assessment of Dermatofibrosarcoma Protuberans (DFSP). MRI is particularly valuable for large tumors, as well as those suspected to have a deeper component, which may not be adequately evaluated through other imaging modalities. Additionally, MRI plays a crucial role in the management of recurrent tumors and those located in

critical anatomical areas where precise surgical planning is essential. It is also beneficial in cases requiring re-excision of DFSPs with positive surgical margins, ensuring that all tumor tissue is effectively removed. Overall, MRI enhances the diagnostic and therapeutic approach to DFSP, facilitating better patient outcomes [15].

In children, recurrence is less common (9%) [16]. (15). Eight children (mean follow-up, 5 years) showed no local recurrences, according to Jafairan et al. [8]. Patients with fibrosarcomatous changes, positive microscopic margins, increased cellularity, high mitotic figures, or those over 50 are most likely to experience recurrence. [17,18]. The prognosis and recurrence do not appear to be significantly impacted by the size of the tumor. [19]. Although recurrences as long as 26 years after the primary tumor was removed have been documented, the majority occur within 3 years of surgery; Long-term monitoring of these patients is therefore necessary [20].

Surgical resection with negative margins is the first line of treatment for localized DFSP.The optimal surgical strategy depends on the tumor's size and location. Since there is little chance of metastasis, regional lymph node dissection is not required. Because of the tumor's capacity to spread to neighboring tissues and the delay in diagnosis, there is a significant chance of an insufficient initial resection. Simple excision will result in local recurrence in almost half of patients. It is more likely for recurrent tumors to spread to distant locations by invading bone, muscle, or fascia [19,21]. Pathological negative margins are required for these reasons.

Conclusion

This case underscores the diagnostic challenges of DFSP in pediatric patients, particularly in atypical locations. Early imaging and histopathological evaluation are vital for accurate diagnosis. Complete surgical resection with negative margins is essential to minimize recurrence risk. Given the potential for late recurrences, long-term follow-up is crucial for ensuring favorable outcomes.

Patient consent

Written informed consent was obtained from the legally authorized representative of the subject (minor patient) for the publication of this case report.

REFERENCES

[1] Gloster HM. Dermatofibrosarcoma protuberans. J Am Acad Dermatol 1996;35:355–74.

[2] Hao X, Billings SD, Wu F, Stultz TW, Procop GW, Mirkin G, et al. Dermatofibrosarcoma protuberans: uupdate on the diagnosis and treatment. J Clin Med 2020;9(6):1752.

[3] Mendenhall WM, Zlotecki RA, Scarborough MT. Dermatofibrosarcoma protuberans. Cancer 2004;101:2503–8.

[4] Jeremic J, Stefanovic A, Jeremic K, Jovic M, Pilic I, Cvetkovic A, et al. Giant dermatofibrosarcoma protuberans vulvae: rare clinical presentation and literature review. JBUON 2019;24:1289–95.

[5] Bogucki B, Neuhaus I, Hurst EA. Dermatofibrosarcoma protuberans: a review of the literature. Dermatol Surg 2012;38:537–51.

[6] Zhang Z, Lu Y, Shi C, Chen M, He X, Zhang H. Pediatric dermatofibrosarcoma protuberans: a clinicopathologic and genetic analysis of 66 cases in the largest institution in Southwest China. Front Oncol 2023;13:1017154.

[7] Iqbal CW, Peter SS, Ishitani MB. Pediatric dermatofibrosarcoma protuberans: multi-institutional outcomes. J Surg Research 2011;170:69–72.

[8] Jafarian F, McCuaig C, Kokta V, Laberge L, Ben Nejma B. Dermatofibrosarcoma protuberans in childhood and adolescence: report of eight patients. Pediatr Dermatol 2008;25:317–25.

[9] Lemm D, Mugge LO, Mentzel T, Höffken K. Current treatment options in dermatofibrosarcoma protuberans. J Cancer Res Clin Oncol 2009;135:653–5.

[10] Taylor HB, Helwig EB. Dermatofibrosarcoma protuberans. A study of 115 cases. Cancer 1962;15:717–25.

[11] Thornton SL, Reid J, Papay FA, Vidimos AT. Childhood dermatofibrosarcoma protuberans: role of preoperative imaging. J Am Acad Dermatol 2005;53:76–83.

[12] Weinstein JM, Drolet BA, Esterly NB, Rogers M, Bauer BS, Wagner AM, et al. Congenital dermatofibrossarcoma protuberans. Variability in presentation. Arch Dermatol 2003;139:207–11.

[13] Bague S, Folpe AL. Dermatofibrosarcoma protuberans presenting as a subcutaneous mass: a clinicopathological study of 15 cases with exclusive or near-exclusive subcutaneous involvement. Am J Dermatopathol 2008;30(4):327–32.

[14] Meguerditchian A, Wang J, Lema B, Kraybill WG, Zeitouni NC, Kane 3rd JM. Wide excision or Mohs micrographic surgery for the treatment of primary dermatofibrosarcoma protuberans. Am J Clin Oncol 2010;33:300–3.

[15] Gould SWT, Agarwal T, Boist S, Patel B, Gedroyc W, Darzi A. Resection of soft tissue sarcomas with intra-operative magnetic resonance guidance. J Magn Reson Imag 2002;15:114–19.

[16] Gooskens SL, Oranje AP, van Adrichem LN, de Waard-van der Spek FB, den Hollander JC, van de Ven CP, et al. Imatinib mesylate for children with dermatofibrosarcoma protuberans. Pediatr Blood Cancer 2010;55:369–73.

[17] Gerlini G, Mariotti G, Urso C, Brandani P, Reali UM, Borgognoni L. Dermatofibrosarcoma protuberans in childhood: two case reports and review of the literature. Pediatr Hematol Oncol 2008;25:559–66.

[18] Sandberg AA, Bridge JA. Updates on the cytogenetics and molecular genetics of bone and soft tissue tumors: dermatofibrosarcoma protuberans and giant cell fibroblastoma. Cancer Gen Cytogen 2003;140:1–12.

[19] Khatri VP, Galante JM, Bold RJ, Schneider PD, Ramsamooj R, Goodnight Jr JE. Dermatofibrosarcoma protuberans: reappraisal of wide local excision and impact of inadequate initial treatment. Ann Surg Oncol 2003;10:1118–22.

[20] Swan MC, Banwell PE, Hollowood K, Goodacre TE. Late recurrence of dermatofibrosarcoma protuberans in the female breast: a case report. Br J Plast Surg 2005;58:84–7.

[21] Bowne WB, Antonescu CR, Leung DH, Katz SC, Hawkins WG, Woodruff JM, et al. Dermatofibrosarcoma protuberans: a clinicopathologic analysis of patients treated and followed at a single institution. Cancer 2000;88(12):2711–20.