

# Rare myxoid dermatofibrosarcoma protuberans masquerading as a pilar cyst in a child

Tanya Greywal, BA,<sup>a,b</sup> Audrey S. Wang, MD,<sup>a,b</sup> Shang I. Brian Jiang, MD,<sup>a</sup> and Andrew C. Krakowski, MD<sup>a,b</sup>  
San Diego, California

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## INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a rare, low-grade malignant neoplasm that rarely presents in children. It is locally aggressive with a high rate of recurrence. We describe a boy who presented with a slow-growing nodule that was thought to be a pilar cyst. Histopathologic examination provided the diagnosis of dermatofibrosarcoma protuberans myxoid variant, which is one of the least common subtypes of DFSP.

## CASE REPORT

A 13-year-old boy presented to the Pediatric Dermatology Department with a 6-month history of an asymptomatic, enlarging pink nodule on his scalp. He could not recall any precipitating trauma to the area, and he denied any pain, pruritus, bleeding, or exudate. The lesion was not treated or biopsied previously by his primary care team. Physical examination found an otherwise healthy boy with a 1.2-cm by 1.5-cm smooth, firm, fairly mobile, pink nodule without a punctum on his scalp and no regional lymphadenopathy (Fig 1).

An excisional biopsy of the nodule was performed (Figs 2 and 3). Histopathologic examination found a poorly marginated, infiltrative dermal spindle cell neoplasm that subtly infiltrated the underlying subcutaneous fat in a honeycomb-like fashion. Relatively monomorphic spindle-shaped cells with tapering nuclei were present in a storiform fashion with a myxoid matrix. Histiocytes and blood vessels were also admixed within the lesion.

Immunohistochemical staining was positive for CD34 and negative for S-100, factor XIIIa, and smooth muscle actin (Fig 4). Fluorescence in situ hybridization analysis found no evidence of

### Abbreviations used:

DFSP: Dermatofibrosarcoma protuberans  
MMS: Mohs Micrographic Surgery  
PDGFB: Platelet-derived growth factor B

platelet-derived growth factor B locus (PDGFB) gene rearrangement.

The patient underwent Mohs micrographic surgery (MMS) to achieve optimal clearance of the margins; only 1 stage of MMS was required. The final defect was 2.4 cm by 4.6 cm (Fig 5). After confirmation of negative margins on formalin-fixed tissue, final closure was performed by plastic surgery under general anesthesia.

## DISCUSSION

DFSP is a rare cutaneous tumor that is locally aggressive with the potential for hematogenous metastasis.<sup>1-3</sup> This tumor accounts for 1.0% to 1.8% of all soft tissue sarcomas and less than 0.1% of all malignancies.<sup>2,4</sup> DFSP typically presents between the ages of 20 and 50, but it has been reported in children.<sup>4</sup> Although some studies have found male or female predominance, DFSP is generally thought to affect both sexes equally.<sup>2-4</sup> In addition, there are several subtypes of DFSP: atrophic, classic, fibrosarcomatous, granular cell, myxoid, pigmented (Bednar tumor), and sclerosing.<sup>1,3,4</sup> The myxoid subtype, with which our patient presented, is one of the least common variants.<sup>1,5</sup>

Clinically, DFSP often presents as an asymptomatic, indurated, violaceous plaque.<sup>3,4</sup> It is fixed to the skin and grows slowly for quite some time before growing rapidly, adhering to deeper

From the Department of Dermatology, University of California San Diego Health System<sup>a</sup> and the Division of Adolescent and Pediatric Dermatology, Rady Children's Hospital, San Diego.<sup>b</sup>  
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Correspondence to: Tanya Greywal, BA, 8010 Frost Street, Suite 602, San Diego, CA 92123. E-mail: [tgreywal@gmail.com](mailto:tgreywal@gmail.com).

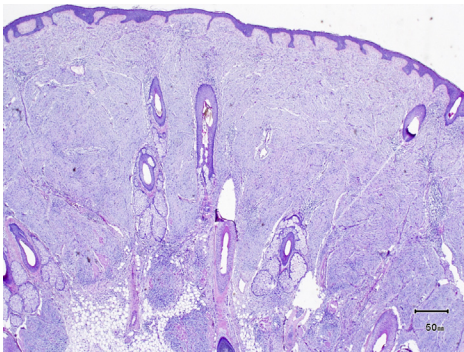
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**Fig 1.** DFSP. A 1.2-cm × 1.5-cm smooth, firm, fairly mobile, pink nodule without a punctum on the scalp of a 13-year-old boy; no regional lymphadenopathy present.

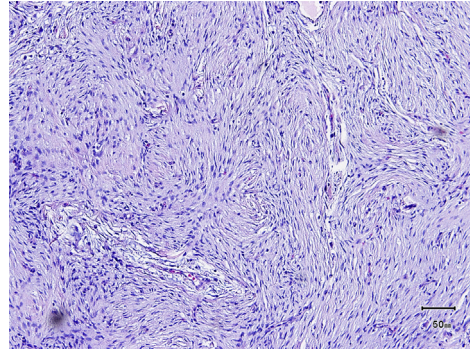


**Fig 2.** A poorly marginated, infiltrative dermal spindle cell neoplasm with a myxoid background. Note the infiltration into the underlying subcutaneous fat in a honeycomb-like fashion. (Hematoxylin-eosin stain; original magnification, ×2.)

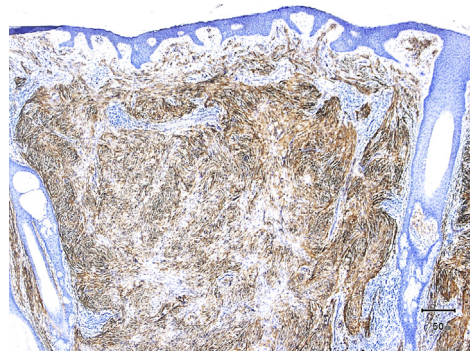
structures, and developing multiple nodules.<sup>2,3</sup> Size typically remains between 1 and 5 cm, although some lesions may grow in excess of 20 cm.<sup>2-4</sup> These lesions usually occur on the trunk, including chest and shoulders, limbs, and head and neck, in decreasing frequency.<sup>2-4</sup> In children, acral areas are most likely to be involved.<sup>3</sup> It has been hypothesized that prior trauma may trigger the development of DFSP; however, only 10% to 20% are associated with a definitive history of trauma.<sup>3,4</sup> Our patient's presentation was less typical, as it consisted of a single pink nodule on the scalp of a child.

The clinical differential diagnosis for DFSP on the scalp of a child may include pilar cyst, dermoid cyst, cylindroma, keloid, hypertrophic scar, melanoma, and metastatic cancer.<sup>2,6</sup> In general, the differential diagnosis of DFSP has also been reported to include mucinous cyst, myxoid mesenchymal neoplasms, pyogenic granuloma, and synovial cyst.<sup>1,2,6</sup>

The diagnosis of DFSP is classically based on histologic findings: spindle cells with a storiform growth pattern invading the subcutaneous tissue forming projections into the fat lobules.<sup>2-4,6</sup> The



**Fig 3.** Relatively monomorphic spindle-shaped cells with tapering nuclei present in a storiform pattern (Hematoxylin-eosin stain; original magnification, ×10.)



**Fig 4.** Most spindle cells are CD34<sup>+</sup> (Immunohistochemistry; original magnification, ×2.)

central areas of the lesion have greater cellularity, whereas the periphery is more diffuse with a honeycomb pattern.<sup>2</sup> These features are typically present, but less characteristic, in the myxoid variant.<sup>5</sup> Prominent features of myxoid DFSP include myxoid stromal changes, multinodular growth, and numerous branched or thickened vessels.<sup>6,7</sup> Immunohistochemical analysis is typically positive for CD34 (although 10%–20% may be negative), vimentin, and CD99 but negative for factor XIIIa, S-100, actin, and desmin.<sup>1,3</sup>

DFSP is thought to result from the reciprocal translocation t(17;22)(q22;q13), leading to fusion of the collagen, type I, alpha 1 and PDGFB genes that cause aberrant activation of the PDGF receptor and subsequent stimulation of cell growth.<sup>1,2,8</sup> Fluorescence in situ hybridization or reverse transcription polymerase chain reaction can isolate this translocation and help identify the corresponding collagen, type I, alpha 1 and PDGFB gene mutations.<sup>3,8</sup> In approximately 8% of DFSP cases, however, these mutations are not detected.<sup>8</sup>

The treatment of choice for DFSP is complete surgical resection.<sup>3,4</sup> Because of the high recurrence rate of these lesions, MMS is preferred over wide local



**Fig 5.** A 2.4-cm × 4.6-cm final defect after MMS.

excision for definitive treatment.<sup>3,4,9</sup> Since MMS was performed on our patient, he did not require the use of radiation therapy or imatinib mesylate (PDGFB inhibitor), which have been used as adjuvant therapy after surgery, primary therapy for unresectable lesions, and treatment for metastatic disease.<sup>7,9</sup>

Because DFSP does not typically present in children, it can be confused with other more common lesions such as a pilar cyst. Therefore, it is important that physicians consider the diagnosis of DFSP in the pediatric population because it is a malignant neoplasm that requires MMS for optimal excision.

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