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CASE REPORT

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Pediatric atrophic dermatofibrosarcoma protuberans

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1 | INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a locally aggressive, slow-growing, cutaneous tumor with a high local recurrence rate, but it rarely metastasizes. The histological morphology of DFSP can be divided into more than 10 subtypes. The atrophic variant of DFSP is rare, especially in children. The clinical diagnosis of atrophic DFSP remains difficult because the lesion often resembles an atrophic plaque, instead of having the typical protuberant morphology. It could be confused with morphea, anetoderma, morphea-like basal cell carcinoma, scar, lymphoma, lipoatrophy, and atrophic dermatofibroma. Now, we report a case of pediatric atrophic DFSP.

2 | CASE REPORT

The patient was a 6-year-old Chinese girl, presented for asymptomatic, oval, atrophic, slightly infiltrated lesions that had been present on the right lower abdomen for 5 years. Her mother mentioned that the lesion was initially a small painless macula, which grew slowly in the first 10 months of the patient's life. No trauma history was identified. Physical examination showed a brown, oval-shaped plaque on the patient's right lower abdomen. This lesion was 4 cm × 2.5 cm in size, depressed below the level of the surrounding skin, and had a smooth, regular surface (Figure 1). The clinical impression was consistent with morphea. Dermatoscopic evaluation of the lesion showed a purple, erythematous background. No telangiectasis or pigment networks were seen (Figure 2). A 4-mm punch biopsy was performed and histopathological examination revealed reduced dermal thickness and wavy spindle cells, which proliferated and irregularly extended into the upper to mid-dermis. Tumor cells reached the subcutaneous fat layer, along with the growth of fat lobules fibrous septa. No mitotic figures were found (Figure 3A-B). Immunohistochemical examination indicated that the cells were strongly positive for CD34 (Figure 3C) and vimentin, and negative for factor XIIIa, S-100 protein, CD31, CD99, CD117, CK, ALK, and Desmin. Detection of the *COL1A1-PDGFB* fusion gene and *PDGFB* gene rearrangement by fluorescence in situ hybridization (FISH) were positive (Figure 4A-B). The lesion was diagnosed as an atrophic variant of DFSP and treated with wide local excision, employing margins of at least 2.5 cm of clinically uninvolved skin and underlying fascia. During follow-up over the subsequent 6 months, there was no evidence of clinical recurrence.

3 | DISCUSSION

The atrophic variant of DFSP was first reported in 1985.[1] Since then, fewer than 50 cases of atrophic DFSP have been reported. In these reports, most of the patients were adults and only a few were

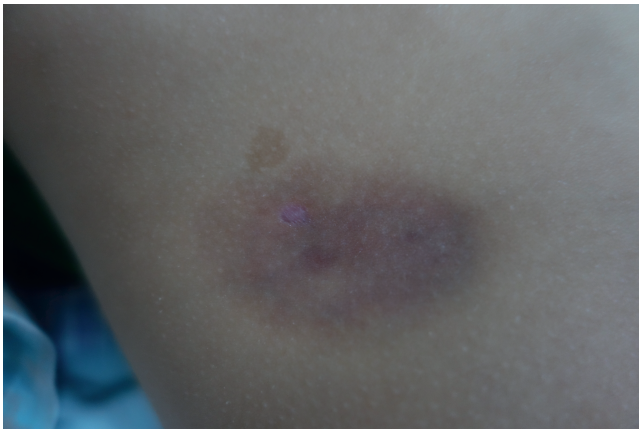

FIGURE 1 Oval brown depressed plaques on the patient's right lower abdomen

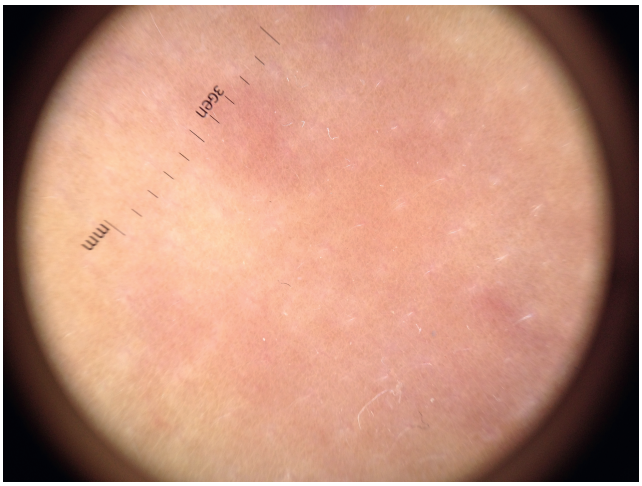

FIGURE 2 Dermatoscopic examination showed a purplish erythematous background. No telangiectasis and pigment network were seen

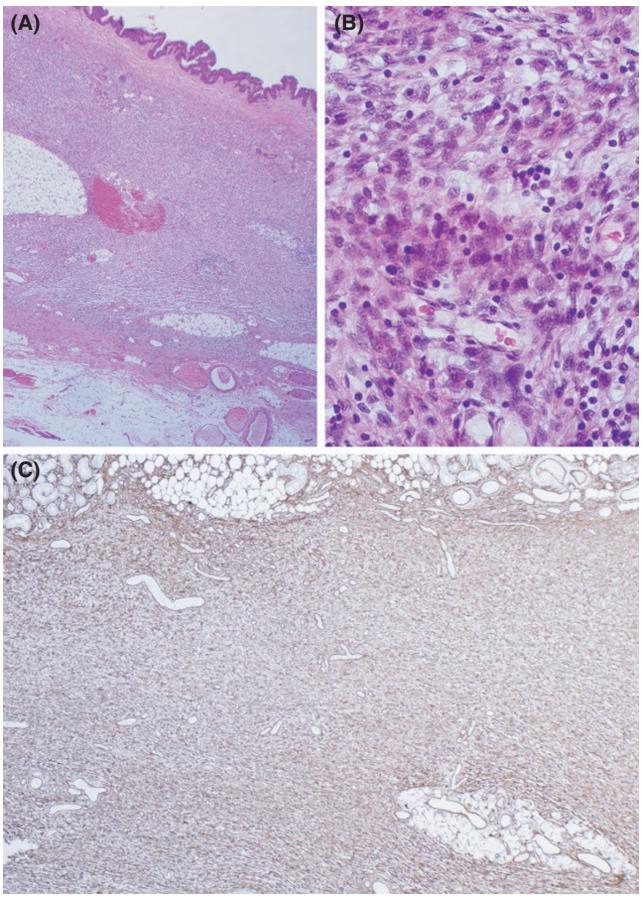

FIGURE 3 A, The thickness of the dermis was reduced, and a spindle cell proliferation replaced the dermis extending into the subcutaneous tissue (hematoxylin and eosin [HE] stain; original magnification, 25×). B, Wavy spindle-shaped cells with small oval nuclei arranged in a storiform pattern and tumor cells insinuated between fat cells in the subcutis (HE; original magnification, 400×). C, Tumor cells are positive for CD34 (CD34 immunostain; original magnification, 50×)

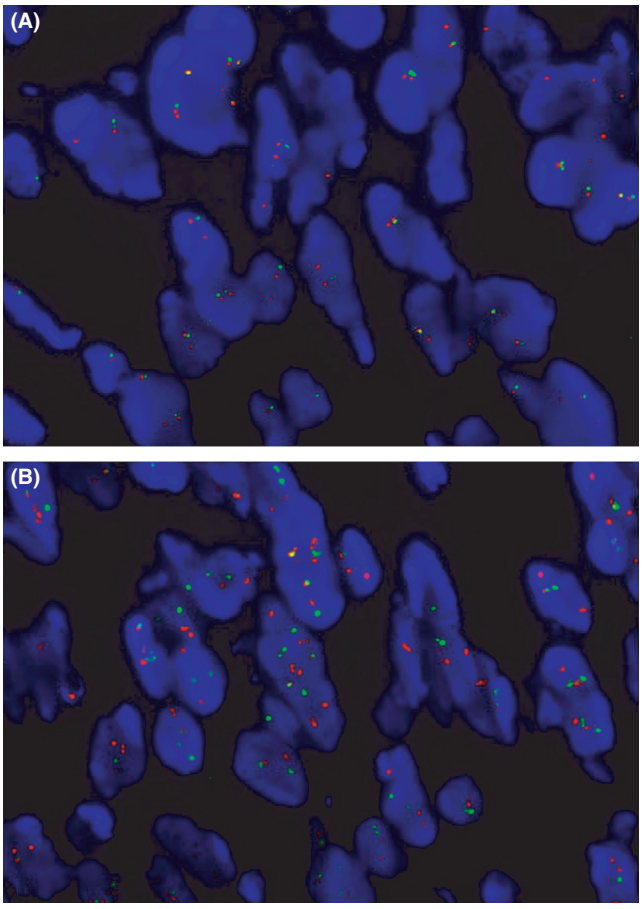

FIGURE 4 A, *PDGFB* gene rearrangement positive signal (one green and one red) (×1000). B, *COL1A1-PDGFB* fusion gene positive signal (red and green overlap) (×1000)

children born with congenital atrophic DFSP.[2] In the present case, the girl's abnormal lesion appeared within 10 months after birth. Atrophic DFSP is characterized by an asymptomatic, atrophic, and morphea-like plaque that persists for a long period of time before developing into a tumor. This silence period is actually highly variable, probably more than 20 years, or even longer. Hanabusa et al[3] reported a case of a patient who was initially diagnosed with morphea, which subsequently developed into protuberant tumors on the face 20 years later that were eventually diagnosed as atrophic DFSP. Makino et al[4] reported a case of a 19-year-old woman with a small congenital atrophic plaque, which remained quiescent for more than 10 years, followed by the development of small nodules on the right precordium. Therefore, dermatologists and pediatricians should be aware of the initial appearance so as to diagnose DFSP as early as possible.

Dermatoscopic studies of atrophic DFSP are very rare in the literature, with only three instances reported.[5-7] The results of these three studies varied in the presence of a pigment network. In our case, dermatoscopic examination revealed no obvious homogenous pigment network on the purple erythematous background. Accordingly, dermatoscopy is not the primary method of diagnosis, but may indicate the need for histopathological examination as the disease has prognostic significance.[6]

Immune markers such as CD34 can aid in the diagnosis of DFSP, but CD34 testing alone is not adequate because there are other soft tissue tumors that may also express CD34. At present, the *COL1A1-PDGFB* fusion gene has a higher sensitivity and specificity in the distinguishing DFSP from other benign or malignant neoplasms. The *COL1A1-PDGFB* fusion gene is detected in >90% of cases of DFSP.[8] The first atrophic DFSP with the *COL1A1-PDGFB* fusion gene was detected in 2008.[9] To our knowledge, only four cases of atrophic DFSP have been confirmed by detection of the fusion gene[4,9-11] and our patient represents the fifth case.

Among children, the workup of atrophic lesions should attempt to exclude the diagnosis of medallion-like dermal dendrocyte hamartoma (ML-DDH). This is a rare congenital lesion, comprising a benign dermal proliferation of fusiform cells that stain positive for CD34, often positive for factor XIIIa, and negative for S100. The characteristic clinical presentation of ML-DDH consists of a well-circumscribed atrophic and wrinkled patch located on the upper trunk or neck that remains stable with time.[12] However, the skin lesions of ML-DDH grew only in proportion to the body, which differs significantly from the progressive size increase observed in congenital atrophic DFSP. For individual cases of difficult identification, molecular testing for *COL1A1-PDGFB* can be used to distinguish ML-DDH from atrophic DFSP.

Conventionally, DFSP is treated with wide local excision or Mohs micrographic surgery. This approach is especially common for tumors on the head and neck. In addition, radiotherapy may be useful in patients with positive surgical margins, to reduce the risk of local recurrence, and in cases where surgery would result in major cosmetic defects. Several published reports have indicated that imatinib mesylate can be considered as an optimal therapeutic choice in patients with metastatic or locally advanced DFSP.[13,14] Tsai et al[15] reported 13 pediatric DFSP patients who received a wide local excision (25 mm-30 mm), and neither recurrence nor metastasis was observed from 20 months to 19 years after the surgery. Our present case remained disease-free for 6 months after the operation. Subsequent follow-up should be carried out every 6 months for the first 3 years and annually thereafter for life.

In conclusion, atrophic DFSP is an important differential diagnosis for atrophic and depressed skin lesions, particularly those seen on the trunks. Confirmation of the *COL1A1-PDGFB* fusion gene can be a useful tool for the diagnosis of DFSP.

CONFLICT OF INTEREST

The authors have no conflicts of interest relevant to this article.

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