Congenital Dermatofibrosarcoma Protuberans

Variability in Presentation

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**Background:** Dermatofibrosarcoma protuberans (DFSP) is an uncommon low-grade fibrohistiocytic tumor that usually occurs on the trunk or proximal extremities and typically appears during the second to fifth decade of life. It most commonly begins as a red-blue plaque that grows slowly and ultimately becomes nodular. The tumor is associated with a high recurrence rate but low metastatic potential. It rarely presents in childhood and is even more rarely present at birth. The clinical diagnosis of DFSP in infancy or childhood may be difficult because, in its early stages, the tumor often resembles a vascular birthmark.

**Observations:** We studied 6 patients with congenital DFSP who were initially thought to have other diagnoses, highlighting the potential clinical variability in presentation. Half of the cases in this series occurred in areas of the body outside of the typically reported distribution pattern of acquired DFSP and in locations that, therefore, may not arouse suspicion of congenital DFSP.

**Conclusions:** Given the aggressive local potential and high recurrence rate of DFSP, early diagnosis is preferable to facilitate appropriate excision. We recommend that any infant or child presenting with a cutaneous plaque or nodule, even congenital, that does not have characteristic or diagnostic clinical features undergo tissue biopsy for histologic evaluation.

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Dermatofibrosarcoma protuberans (DFSP) is an uncommon low-grade fibrohistiocytic tumor that occurs on the trunk or proximal extremities in most cases and most often appears during the second to fifth decade of life. The incidence of DFSP is estimated to be 0.8 to 5 cases per million persons per year, or 0.1% of all malignancies. Histologically, the tumor consists of a proliferation of spindle cells in a cartwheel or storiform pattern, with immunohistochemical stains showing positivity for CD34 antigen and negativity for desmin, S100, and factor XIIIa. The tumor is associated with a high recurrence rate but low metastatic potential. Standard treatment for DFSP has traditionally consisted of wide excision (ie, 3-cm margins). However, in recent years, Mohs micrographic surgery has evolved as the therapeutic standard of care.

Dermatofibrosarcoma protuberans rarely occurs in childhood (around 160 cases reported) and is even more rarely present at birth (21 cases reported). In its early stages, DFSP most commonly appears as a red-blue plaque that grows slowly over years and ultimately becomes nodular or protuberant. Thus, the clinical diagnosis of DFSP in infancy or childhood may be difficult because, especially in its early stages, the tumor may closely resemble a vascular birthmark. In most reported cases of congenital DFSP, the diagnosis was delayed until after several decades of life. Although congenital or childhood DFSP is often initially different from the adult form in appearance, the anatomic distribution of both groups parallels that of the adult form, with most lesions appearing on the trunk or proximal extremities.

We describe 6 patients with congenital DFSP who were initially thought to have other conditions, often including a vascular malformation or tumor, and describe their presentation and course. The goal of this report is to highlight the potential clinical variability in presentation of congenital DFSP and the importance of considering this diagnosis in pediatric patients with atypical cutaneous or subcutaneous tumors.
The clinical features and treatment course of 6 previously unreported cases of congenital DFSP are presented in Table 1. Two of the patients are described in more detail herein.

CASE 1

A 2-month-old male presented to the pediatric dermatology clinic with a 0.8-cm, firm to slightly cystic, flesh-colored subcutaneous nodule on his midparietal scalp (just to the right of midline; Figure 1) that had been present since birth. Long, darkly pigmented hair was present around the lesion, and there was no visible sinus pit or ostium. The clinical differential diagnosis at presentation included dermoid cyst, cephalocele, and glioma. After magnetic resonance imaging with contrast was performed, which showed no intracranial abnormalities or neurocutaneous tracts, the patient was referred to the pediatric plastic surgery department for excision. The surgeon also considered a mixed vascular and neural hamartoma in the differential diagnosis and performed an excisional biopsy, which showed a spindle cell proliferation extending to the deep dermis and forming irregularly woven fascicles in a matlike pattern. Immunohistochemical studies were positive for CD34 and negative for S100, desmin, HHF-35 (muscle-specific actin), and factor XIIIa. These findings were interpreted as being consistent with DFSP. A subsequent reexcision, with negative margins, was performed after tissue expansion, which enabled primary closure. There has been no sign of recurrence in 1 year of follow-up.

CASE 3

A 6-year-old boy was examined by a pediatric surgeon because of a 1 × 1.5-cm atrophic oval-shaped lesion with a central blue nodule. The lesion presented as a congenital, 0.5-cm atrophic blue plaque on the patient’s back and slowly enlarged, with the appearance of the central nodule at 3 years of age. A vascular malformation was suspected, but duplex scanning of the lesion demonstrated no flow. The entire lesion was removed at that time and was diagnosed initially as a fibrous histiocytoma.
Two years later, the lesion recurred, and the patient subsequently presented to the pediatric dermatology department at age 12 years with a 4 × 6-cm gray-blue atrophic plaque with a central scar and a blue nodule at the upper pole (Figure 2). The family refused biopsy at that time, but the patient returned at age 14 years, at which time the lesion measured 5 × 10 cm. Examination of a biopsy specimen disclosed a spindle cell tumor with a storiform pattern and immunohistochemical studies equivocal for smooth-muscle actin and S100 and negative for lysozyme, factor VIII, and desmin. These findings were interpreted as consistent with DFSP. A wide reexcision was performed with split-thickness grafting for repair of the defect. The excision was reported as “possibly incomplete,” and 1 year later there was an apparent recurrence at the suspect medial margin of the graft, which was excised but reported only as scar tissue. Three years later, at age 18 years, a 1-cm mobile subcutaneous tumor was found at the medial margin of the scar. Examination of a biopsy specimen showed a cystic cavity filled with a soft polyp of tissue, which was found on histologic evaluation to be consistent with recurrent DFSP. A further wide excision, with negative margins, was done with another skin graft for repair. There has been no further recurrence in 6 years of follow-up.

**COMMENT**

Dermatofibrosarcoma protuberans is a soft-tissue neoplasm of intermediate malignancy that is considered to be a low-grade sarcoma. Although DFSP lesions have low metastatic potential, they have a pronounced tendency to recur, and there are occasional reports of metastases and death. Metastases have been reported to occur in 1% to 6% of patients and are a poor prognostic sign, most often resulting in death within 2 years. Multiple and repeatedly recurrent tumors appear to be the greatest risk factors for metastatic disease, and the lung is the most common site of metastases (75%) via hematogenous spread. Most DFSPs present during mid adult life (20-50 years of age). Occurrence in pediatric patients is significantly less common (although the frequent delay in diagnosis may artificially deflate the figures for age at onset), and occurrence at birth has been described only rarely.

Six patients with congenital DFSP are described herein, to highlight the potential variability in presentation of these rare tumors. The most commonly considered diagnosis included in the differential diagnosis of these patients was vascular tumor or malformation (5 of the 6 patients). Other entities included in the differential diagnoses included myofibroma, subcutaneous fat necrosis, arthropod bites, fibrous hamartoma, aplasia cutis, and intrauterine trauma. In the patient who presented with a scalp DFSP, the differential diagnosis included midline embryologic defects, such as dermoid cyst, glioma,
In addition, although DFSP presenting in infancy or childhood is traditionally described as a plaque (in contrast to the more nodular tumors presenting in adulthood), 4 of the 6 children in this series presented with lesions that were either partially or completely nodular.

Of the 21 previously reported cases of congenital DFSP, 16 reports included details such as patient age and tumor location. Most of those tumors (14/16) occurred on the trunk or proximal extremity and the others (2/16) occurred on the distal extremity. In our series, half of the tumors (3/6) occurred on the trunk or proximal extremity, while the other half occurred on the distal extremity (1/6) and on the head and neck (2/6) (Table 2). Thus, while most of the previously reported cases of congenital DFSP follow the distribution pattern of acquired DFSP on the trunk or proximal extremities, half of the cases presented in this series occurred outside those areas and in locations previously not reported, which therefore may not arouse suspicion of congenital DFSP.

Both congenital and acquired DFSP tumors presenting in infancy or childhood may be misdiagnosed for many years because of their close resemblance to other lesions, especially vascular tumors or malformations. The growth characteristics of the lesion are important in making this distinction clinically, given the slow but steady growth typical of DFSPs. Hemangiomas, on the other hand, grow during the early proliferative stage, but eventually enter an involution period of spontaneous regression, which is not typical of DFSP. Vascular malformations exhibit relative stability in size, growing only in proportion to the patient’s growth. However, in some patients, growth characteristics alone are insufficient to distinguish clinically between vascular lesions and DFSP. In these patients, radiologic evaluations (ie, magnetic resonance studies) may be useful, and the gold standard diagnostic examination is tissue biopsy.

Patient 1 in the current series presented with a lesion on the midline scalp that had surrounding, darkly pigmented, long hair (“hair collar sign”), a finding reported in infants with cephaloceles, meningoceles, heterotopic brain tissue, and membranous aplasia cutis.20 His presentation suggested one of these developmental en-

Table 2. Tumor Location and Congenital Dermatofibrosarcoma Protuberans

<table>
<thead>
<tr>
<th>Location</th>
<th>Reported Cases &lt;sup&gt;1-4, 8-12, 14&lt;/sup&gt; (n = 16)</th>
<th>Current Series (n = 6)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No.</td>
<td>References</td>
</tr>
<tr>
<td>Trunk</td>
<td>12</td>
<td></td>
</tr>
<tr>
<td>Not further specified</td>
<td>1</td>
<td>7</td>
</tr>
<tr>
<td>Chest</td>
<td>1</td>
<td>11</td>
</tr>
<tr>
<td>Breast</td>
<td>4</td>
<td>1, 2, 6, 8</td>
</tr>
<tr>
<td>Abdomen</td>
<td>3</td>
<td>4, 11</td>
</tr>
<tr>
<td>Back</td>
<td>3</td>
<td>3, 10, 14</td>
</tr>
<tr>
<td>Upper extremity</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Shoulder</td>
<td>1</td>
<td>10</td>
</tr>
<tr>
<td>Hand/wrist</td>
<td>1</td>
<td>12</td>
</tr>
<tr>
<td>Lower extremity</td>
<td>2</td>
<td></td>
</tr>
<tr>
<td>Thigh</td>
<td>1</td>
<td>11</td>
</tr>
<tr>
<td>Leg</td>
<td>1</td>
<td>9</td>
</tr>
<tr>
<td>Ankle/foot</td>
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</tr>
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<td></td>
</tr>
<tr>
<td>Scalp</td>
<td>0</td>
<td>...</td>
</tr>
<tr>
<td>Neck</td>
<td>0</td>
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</tr>
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</table>

<sup>*No previous report of congenital dermatofibrosarcoma protuberans in this anatomic location.</sup>
bryologic defects, with the unexpected finding of DFSP on initial excisional biopsy. This patient’s presentation is unlike others reported to date and suggests another differential diagnosis that must be included in newborns with congenital scalp nodules with the hair collar sign.

Early diagnosis is desirable in any soft-tissue tumor and, in the case of DFSP, is useful in facilitating early excision, hence treating relatively smaller tumors and subsequently minimizing the extent of surgical scarring. Although the tumor location for patient 3 was more typical of DFSP, the initial misdiagnosis as a benign fibrous histiocytoma led to lack of attention to appropriate excisional margins and follow-up. Consequently, the patient was not adequately treated until 6 years after recurrence, when the tumor was nearly 10 times its original size, at which time the correct diagnosis of DFSP was rendered and wide excision with skin grafting was recommended. Similarly, in the case of patient 4 (Figure 3), the initial histologic diagnosis of dermatofibroma delayed correct diagnosis of DFSP until 21 months of age, when the lesion recurred.

In comparison with previously reported cases of congenital DFSP, which were diagnosed between 2 and 69 years of age with almost half (7 of 16 cases with sufficient data) diagnosed in adulthood (ages 25-69 years), the age at diagnosis for the current series ranged from 5 months to 14 years. This may relate to a higher index of suspicion and lower threshold for biopsy of children by pediatric dermatologists. Given the high recurrence rate of DFSP and the rare but existing potential for metastatic spread, early diagnosis is important to ensure appropriate and complete excision and reduce the potential for these complications. We recommend that any infant or child presenting with a cutaneous plaque or nodular lesion undergo tissue biopsy for histologic evaluation.

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REFERENCES


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