Childhood Dermatofibrosarcoma Protuberans, Giant Cell Fibroblastoma Variant: A Case Report and Discussion

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Abstract
Dermatofibrosarcoma protuberans (DFSP) is a fibroblastic tumor with low-grade malignant potential. DFSP is slow-growing, locally aggressive and has a high recurrence rate. It is most commonly seen in adults, but a small percentage is seen in the pediatric population. In children, there is typically a significant lapse between presentation and diagnosis. We present a case of a 14-year-old male with a longstanding lesion diagnosed as DFSP, giant cell fibroblastoma variant. This variant is commonly seen in the pediatric population and has specific histopathologic features. The cytogenetic profile and treatment options are similar for the giant cell fibroblastoma variant and other forms of DFSP, so they will be discussed as one entity. We present this case to raise awareness of the clinical presentation of DFSP in the pediatric population, with the goal of aiding in early diagnosis and prompt treatment to minimize functional and cosmetic disfigurement.

Introduction
Dermatofibrosarcoma protuberans (DFSP) is a slow-growing, locally aggressive, cutaneous fibroblastic mesenchymal neoplasm with low-grade malignant potential.¹ ² DFSP is most commonly seen in individuals 20 to 50 years of age, but 6 percent of cases present in the pediatric population.³ ⁴ Diagnosis is often delayed in the pediatric population, leading to increased lesion size and significant cosmetic disfigurement with treatment options. The delay in diagnosis may be due to the common clinical mimickers of early-stage DFSP in children, such as morphea or vascular lesions, which share the erythematous-to-blue hue of the atrophic plaque.⁵ We present a case of a 14-year-old male with a lesion that was present for 13 years before a diagnosis was made.

Case Report
A 14-year-old male presented with his mother for the evaluation of a skin lesion on the right aspect of the chest that had been present since 1 year of age. The mother stated that it started out as a dimple and continued to grow. The lesion was not painful, and there was no trauma to the area.

Clinical examination revealed a 4.9 cm x 4.5 cm atrophic, indurated, erythematous to violaceous plaque on the right superior aspect of the chest (Figure 1). Two punch biopsies were performed, showing a dermal proliferation of spindle cells that stained strongly positive for CD34 (Figure 2). Several multinucleate cells were seen at the periphery of the lesion, and mucinous stromal changes were noted. A diagnosis of dermatofibrosarcoma protuberans (DFSP), giant cell fibroblastoma variant was made.

The patient was referred to a tertiary center and underwent wide local excision with 2.0 cm margins. Pathology revealed residual DFSP extending to the deep margin of the specimen. Clear margins were obtained on re-excision, and the defect was repaired with a split-thickness skin graft. The patient will be followed every six months for skin examinations.

Discussion
Dermatofibrosarcoma protuberans (DFSP) is a slow-growing, fibroblastic neoplasm with low-grade malignant potential and high recurrence rates after excision.¹ ² The incidence of DFSP is 0.8 to 4.2 cases per million per year.³ It is most commonly seen in patients 20 to 50 years of age.³ The most common locations for DFSP, in decreasing order of frequency, are the trunk, proximal extremities and head and neck.² ³ Ten percent to 15 percent of DFSPs contain a fibrosarcomatous component, indicating a more aggressive nature and thus an increased risk of recurrence and metastasis.⁵ Clinically, a DFSP may present as a slow-growing, atrophic, indurated violaceous to red-blue plaque that develops nodules over time.¹ ³ This presentation broadens the differential diagnosis to include morphea, atrophoderma, vascular lesions and various scarring processes.⁴ There are currently several dermatoscopic features that are found in, but not specific to, DFSP. These include a delicate pigmented network, vessels, structureless light brown areas, white streaks, pink background and structureless hypopigmented areas.⁴ Cytogenetic studies have revealed a rearranged chromosomal sequence that is found in more than 90 percent of DFSP cases. This is a reciprocal translocation t(17;22)(q11;q11) that consists of a1 type I collagen (COL1A1; 17q21) and platelet-derived growth factor β-chain gene (PDGFB; 22q13).² ³ This translocation upregulates PDGFB, leading to activation and differentiation of mesenchymal cells, thus predisposing patients to DFSP. The genetic defects can be detected using FISH or reverse transcription polymerase chain reaction.² Diagnosis is confirmed with punch biopsy and corresponding histopathology.

Histopathology for DFSP shows a poorly-circumscribed, dense collection of spindle cells in the dermis arranged in a storiform pattern. The spindle cells can invade the subcutaneous tissue in a honeycomb or lace-like pattern. The cells are bland in appearance with few mitoses. Spindle cells stain positive for CD34 and vimentin but negative for factor XIIIa, S-100 and desmin.¹ ³ ⁵ The giant cell fibroblastoma variant of DFSP, as seen in our patient, is a juvenile form of DFSP characterized by the presence of multinucleated cells that stain strongly positive for CD34 and mucinous stromal changes. CD34 stain was strongly positive.

Figure 1
Figure 1. 4.9 cm x 4.5 cm atrophic, indurated, erythematous to violaceous plaque on the right superior aspect of the chest.

Figure 2
Figure 2. Punch biopsy showing dermal proliferation of spindle cells with several multinucleated cells at periphery and mucinous stromal changes. CD34 stain was strongly positive.
giant cells and myxoid stromal changes on histopathology. This variant of DFSP has the same chromosomal translocation as other variants of DFSP, and treatment options for the two are similar.

Treatment options for DFSP mainly rely on surgical excision with wide margins. There has been much debate on the appropriate margins to take when performing wide local excision. The goal is to maintain clear margins with excision, in order to decrease recurrence rate, while avoiding severe cosmetic disfigurement. Excision has been performed with anywhere from 1 cm to 5 cm margins. One study by Woo et al assessed the long-term outcomes of surgical treatment in DFSP according to width of gross resection margin. Their assessment recommended wide local excision with a margin of 1.5 cm to 2 cm, with intraoperative frozen section analysis for initial treatment. Suggested margins for wide local excision vary; to our knowledge, there is no definitive recommendation regarding margins for initial excision for DFSP. Mohs micrographic surgery has been implicated in the treatment of DFSP on the head and neck, but should be performed by an institution with a surgeon and pathologist proficient in treating this type of tumor. DFSP is responsive to radiation, which can be considered as an adjuvant therapy in certain cases. There are no randomized studies assessing the efficacy of adjuvant radiation therapy in DFSP, but Chen et al. performed a systematic review and meta-analysis to evaluate outcomes. Although further studies are needed, there is evidence suggesting that adjuvant radiation may be effective in controlling recurrent tumors that were excised with narrow or positive margins, as well as obtaining control when clear margins could result in severe functional or cosmetic impairment. The chromosomal translocation discussed previously allows for targeted therapy for DFSP. Reports have shown that imatinib mesylate, a tyrosine kinase inhibitor that works by blocking PDGF signaling, has beneficial activity in metastatic, inoperable, or pre-operative downstaging cases. Dose ranges from 400 mg to 800 mg once daily of imatinib mesylate have been used. Although promising, further studies are needed to determine when and at what dosage imatinib mesylate should be utilized.

Conclusion

This case of a 14-year-old male diagnosed with dermatofibrosarcoma protuberans (DFSP), giant cell fibroblastoma variant exemplifies the delayed DFSP diagnosis typical in the pediatric population. The giant cell fibroblastoma variant has specific histopathologic features but shares a cytogenetic profile and treatment options with other variants of DFSP. The mainstay of treatment is surgical excision with wide margin control. There are exciting treatment options underway that can be used in conjunction with surgical excision, including radiation therapy and imatinib mesylate, although further studies for their appropriate use are required. Physicians from multiple specialties that care for the pediatric population should be aware of the clinical presentation of DFSP to allow for early recognition and timely treatment to decrease morbidity and cosmetic disfigurement.

References