

Perianal Dermatofibrosarcoma Protuberans: A Case Report

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A 67-year-old man presented with a perianal lump that had increased in size. On examination he had a 3-cm irregular, mobile, elevated, red, polypoid lump at the edge of the anus at the 8-o'clock position. Biopsy results unexpectedly revealed a spindle cell lesion extending deep into the subcutaneous tissue with occasional mitoses. The lesion was positive for CD34 and negative for epithelial markers, consistent with dermatofibrosarcoma protuberans (DFSP). Magnetic resonance imaging of the pelvis showed the mass extending deep into the ischiorectal space with no involvement of the external or internal anal sphincter. He underwent excision of the lesion with circumferential margins of 1 cm and formation of a skin rotation flap to achieve primary closure. Histology confirmed DFSP. Both the deep and lateral resection margins were involved. He proceeded to have a wider excision of margins, which was free of any remaining tumor.

Dermatofibrosarcoma protuberans is a rare lesion. It most commonly occurs on the trunk; the perianal presentation in this case is unique. Surgical excision and preservation of functionality with cosmesis was an issue in this case, as DFSP is a locally aggressive tumor with a high recurrence rate.

Cutis. 2011;87:85-88.

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The authors report no conflict of interest.

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Case Report

A 67-year-old man presented with a perianal lesion that had been excised 15 years prior but grew back. On presentation the patient reported a bulky protuberance that had ulcerated the skin and bled. He had no associated symptoms of pain, no blood per rectum, and no change in bowel habit or weight.

The histology of the lesion excised 15 years prior was reviewed and showed a polypoid expansion of the keratoderma by a proliferation of fusiform to spindle-shaped cells arranged in a vague storiform pattern. Toward the superficial aspect, numerous multinucleated giant cells were present. At one edge there was evidence of myxoid change and the overlying epidermis was mildly hyperkeratotic but otherwise unremarkable. The lesion infiltrated deeply with tentacles of spindle cells extending through groups of subcutaneous fat cells. Overall the appearance was suggestive of a fibrous histiocytoma or dermatofibroma.

Diverticular disease was diagnosed on colonoscopy 2 years prior to presentation. At the time, a perianal pea-sized skin lesion was noted. Surgical history included excision of a basal cell carcinoma from the patient's back. Of note in his family history, his daughter had a sarcoma excised from her head; the margins were involved, requiring further excisions of this lesion. We were unable to obtain the exact histology.

On examination of the patient, no other skin lesions were found. His abdomen was soft and nontender with no masses. On rectal examination a 3-cm irregular lump was noted at the edge of the anus at the 8-o'clock position (Figure 1).

Routine full blood count, renal profile, and liver profile were normal. Magnetic resonance imaging of the pelvis was performed and showed a well-defined lesion with internal enhancement at the anal margin (Figure 2). The lesion extended from the right-hand side of the natal cleft for a distance of

6 cm deep into the ischioanal space coming to lie on the lateral aspect of the internal anal sphincter. There was no involvement of the external or internal anal sphincter. There was no evidence of communication with the anal canal or the rectum. The lesion was high signal on T2 compared to fat (Figure 2) and high signal following gadolinium contrast on T1-weighted magnetic resonance imaging.

An examination under anesthesia and biopsy showed that the anal mucosa and rectum were normal. A 3-cm dermal, subcutaneous, mobile nodule was obvious. It did not involve anal skin and radiated outward at the 8-o'clock position while the patient was in the lithotomy position. It was 3 to 4 cm in depth. A biopsy of the lesion was performed and histology showed a spindle cell lesion with a well-defined storiform pattern (Figure 3) extending deep into the subcutaneous tissue. Occasional mitoses were seen. It was positive for CD34 (Figure 3) and negative for epithelial markers AE1/AE3, S-100 protein, smooth muscle-specific actin, and factor XIIIa, consistent with dermatofibrosarcoma protuberans (DFSP).

The patient was electively admitted for excision of the DFSP and skin rotation flap to achieve primary closure. An elliptical incision with 1-cm margins was made around the lesion (Figure 4A). The lesion was excised using diathermy with macroscopic circumferential margins of 1 cm. Dissection extended down to the external anal sphincter with preservation of it. A second incision was made to rotate a skin flap of the inferior buttock medially to close the skin (Figure 4B). 3/0 Nylon was used to close the skin. Postoperatively, 3 doses of antibiotics, a low-residue diet, and laxatives were prescribed.

Histology demonstrated the presence of a spindle cell lesion showing a well-defined storiform pattern. There was infiltration of the adjacent connective tissue stroma and underlying fat with involvement of both deep and lateral resection margins. In addition, the tumor extended close to but appeared free of the anal soft tissue margin. Mitoses were relatively apparent. Features were diagnostic of DFSP. The sarcoma was of intermediate malignancy. The patient proceeded to have a wider excision of margins, which was free of any remaining tumor.

Comment

In 1924, Darier and Ferrand¹ first described DFSP as a distinct cutaneous lesion and referred to it as a progressive and recurrent dermatofibroma. Hoffman² officially coined the term in 1925. Dermatofibrosarcoma protuberans is a cutaneous malignancy that arises from the dermis and invades deeper subcutaneous tissue (eg, fat, fascia, muscle, bone).

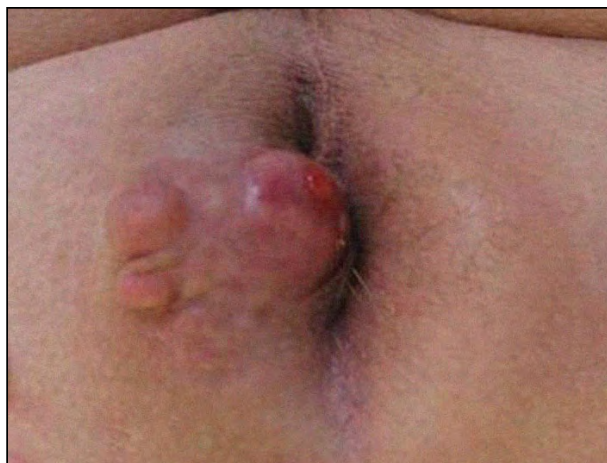


Figure 1. Perianal lesion with patient in lithotomy position.

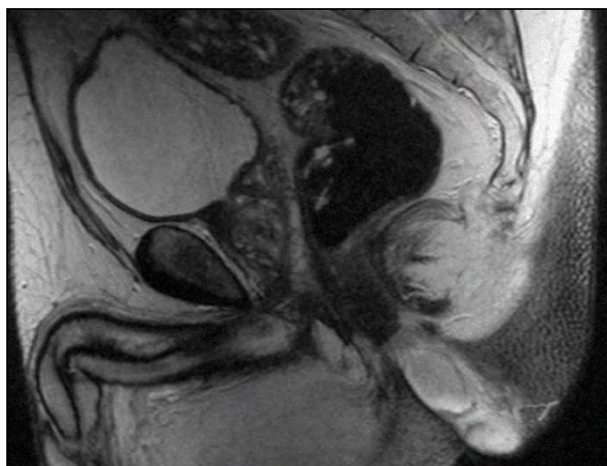


Figure 2. Magnetic resonance imaging sagittal section showed a well-defined lesion at the anal margin with high signal on T2 compared to fat.

It most commonly occurs on the trunk.³ Dermatofibrosarcoma protuberans is rare, with an incidence of 3 per 1,000,000 individuals per year, according to one study.⁴ It is a locally aggressive tumor with a high recurrence rate; however, despite the local invasiveness, it rarely metastasizes. Up to 90% of cases of DFSP are associated with a translocation between chromosomes 17 and 22 that places the platelet-derived growth factor-B under the control of the collagen 1A1 promoter.⁵ This process results in a collagen 1A1-platelet-derived growth factor-B fusion protein that acts as a continuous stimulator for DFSP cells by binding the platelet-derived growth factor receptor.⁶

Clinically, DFSP initially presents as a nonprotuberant plaque that behaves in an indolent fashion. The nonspecific clinical features often result

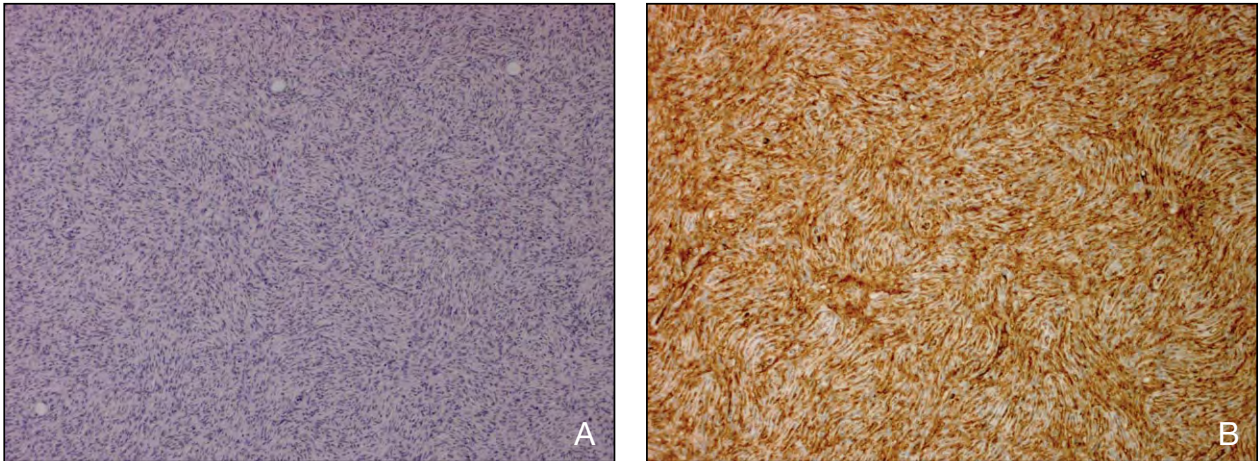


Figure 3. On histopathology the lesion was composed of spindle cells arranged in a storiform pattern (ie, intersecting fascicles appeared to radiate from an acellular collagenous focus, cart-wheel arrangement)(A)(H&E, original magnification $\times 100$). Spindle cells were positive for the immunohistochemical marker CD34 (B)(original magnification $\times 100$).

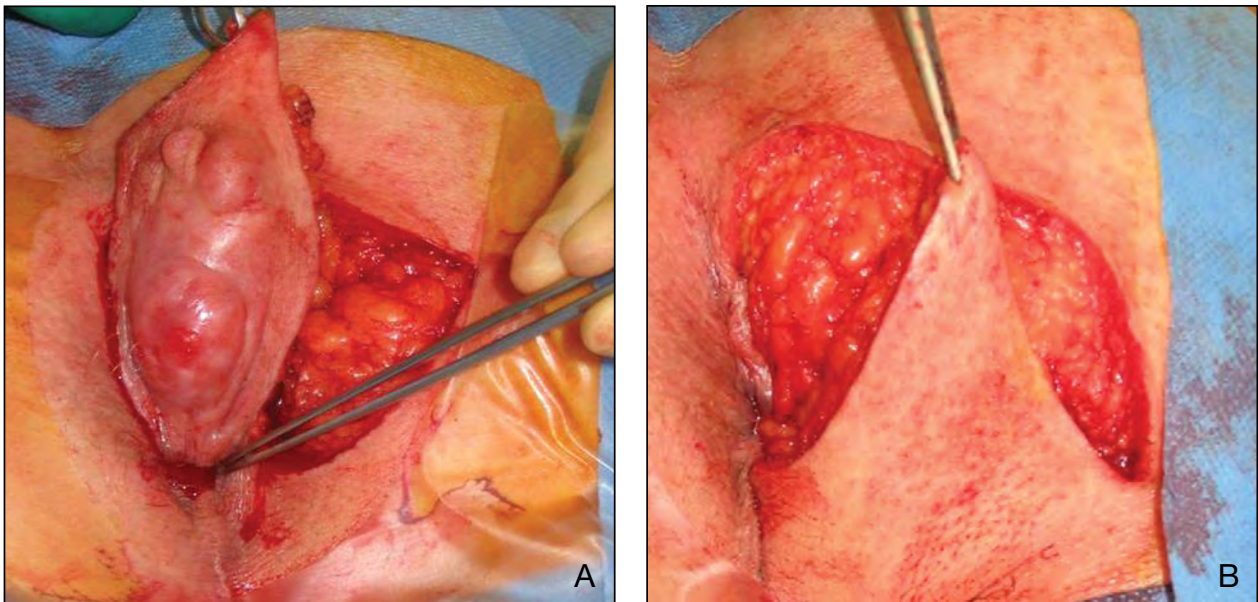


Figure 4. Excision of lesion with 1-cm margins (A). A flap was raised to close the skin (B).

in a delay in diagnosis. It is characterized by an asymmetrical growth pattern that often can result in local recurrence if the whole tumor is not adequately excised.

Magnetic resonance imaging is useful in identifying the extent and location of DFSP, especially regarding recurrence.⁷ A tissue biopsy is essential for diagnosis and characteristic histologic appearances include a dense uniform proliferation of spindle-shaped tumor cells arranged in a storiform pattern. Immunohistochemically these cells are positive for the human progenitor cell antigen CD34.

Surgical excision remains the mainstay of treatment of DFSP. In some surgical centers, margins of more than 3 cm are recommended to reduce the incidence of local recurrence.⁸ Because of the high recurrence rate, Mohs micrographic surgery has been increasingly accepted as the treatment of choice.⁹ The aim of Mohs micrographic surgery is to remove all of the tumor roots as accurately as possible by histologically confirming negative margins and to create the smallest possible defect by sparing tissue uninvolved by tumor.¹⁰ It is performed under local anesthetic. The visible tumor is excised with 2-mm margins and specimens are marked with dye for orientation. The

tissue is made into frozen sections and microscopically examined. Positive margins are mapped out and the process is repeated until negative margins are achieved.¹⁰ One study has shown a 5-year cure rate of 100% with Mohs micrographic surgery.⁹ Because of the drawbacks of Mohs micrographic surgery (ie, considerable training required, labor-intensive procedure, performed under local anesthesia), it is not widely available and some institutions have found that wide excision with histologic assessment of margins offers similar results.¹¹⁻¹³

Radiation therapy has been used as an adjunct to surgery.¹⁰ It is used in patients with involved margins or lesions that are unresectable. The development of molecularly targeted therapy, imatinib mesylate, has been found to have clinical activity against both localized and metastatic DFSP with t(17;22).¹⁴ Close surveillance is necessary and long-term follow-up is required, as recurrences can occur up to 5 years postoperatively.

This case report highlights a number of issues. Dermatofibrosarcoma protuberans is a rare lesion that most commonly occurs on the trunk; the perianal distribution in our patient was unique. This patient had a prior benign biopsy; however, DFSP is a slow-growing tumor. It may start as a small asymptomatic papule that is likely ignored and may gradually enlarge into a lumpy nodule or it may evolve into an atrophic or sclerotic plaque. Regarding family history, laboratory studies have shown that chromosomal aberrations may contribute to the pathogenesis of DFSP; however, no evidence of hereditary or family predisposition exists. Surgical excision and preservation of functionality with cosmesis was an issue in this case, as DFSP is a locally aggressive tumor with a high recurrence rate. Although surgical excision is the primary treatment, other therapies such as Mohs micrographic surgery, radiotherapy, and the monoclonal antibody imatinib mesylate are effective for this condition.

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