

Dermatofibrosarcoma Protuberans in a Male Infant



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Dermatofibrosarcoma protuberans is a rare cutaneous malignancy known to be locally aggressive. It is uncommonly seen in the pediatric population and can be difficult to distinguish from other benign skin lesions. We present a case of dermatofibrosarcoma protuberans of the penis in a 6-month-old child managed with surgical resection. This case highlights the challenges of diagnosis of genital lesions in children and the complexities of genitourinary reconstruction following surgical resection. UROLOGY 129: 206–209, 2019. © 2018 Elsevier Inc.

Dermatofibrosarcoma protuberans (DFSP) is a rare cutaneous malignancy with reported annual incidence of 4.2 per million (0.3 to 1.3 per million in pediatric patients) in the United States. Patients are typically 20–50 years old. DFSP most commonly occurs on the trunk, and is very rarely found on the genitalia.¹ To our knowledge, only four cases of penile DFSP have been reported.^{2–4} The tumor is locally aggressive, with few reported cases of metastasis.¹ There is a paucity of data concerning characteristics of disease and treatment strategies with only 2 published guidelines available to guide management.^{5,6} We present a case of DFSP of the penis in an infant, managed by surgical resection.

CASE PRESENTATION

A 6-month-old male presented for elective circumcision with his adoptive parents. Medical history included birth at 36 weeks gestational age, but further history was unknown. There was a 5 mm smooth, raised, erythematous lesion at 7 o'clock near the base of the penis (Fig. 1). The lesion was previously evaluated by five other physicians, including a dermatologist, and was suspected to be a mastocytoma (benign collection of mast cells that typically resolves over time). After discussion of management options, the patient's parents elected for excision of the lesion at the same time as an elective circumcision. Circumcision went without complication, and the lesion was ellipsed horizontally, with a resulting 10 × 5 × 2 mm specimen. Considering the lesion was presumed to be benign in nature, no orienting markings were placed

and no frozen section was sent intraoperatively. The foreskin was not sent to pathology per institutional practice.

Pathologic evaluation by a dermatopathologist revealed a CD34+ spindle cell neoplasm, favoring DFSP, with involvement of deep and “lateral” margins (again, the specimen was not orientated). FISH for the chromosomal translocation t(17,22) was negative. CT chest obtained for staging was negative for metastasis. After discussion at multidisciplinary tumor board, options for management proposed included Mohs surgery under local anesthesia by dermatology versus wide local excision with frozen section under general anesthesia by urology.

After discussion with the family, they agreed to proceed with wide local re-excision with urology. Bipolar excision was carried out to the circumcision incision distally and the base of penile shaft proximally using a complex margin legend (Fig. 2c). Initial frozen section of the 12 o'clock margin was positive, but all other margins were negative. Another distal margin was removed which was then negative on frozen section. Final resection included ~50% of penile shaft skin from base to collar with a final defect of 2 cm × 3 cm (Fig. 2). The defect was closed primarily using 5-0 monocril in interrupted horizontal mattress fashion (Fig. 3a). Final pathology was spindle cell neoplasm consistent with residual DFSP. The distal margin, which was initially positive for residual disease, was no longer involved, making all margins negative. The specimen was then sent out for second opinion expert review by the dermatopathologist and returned as perineuroma, another rare diagnosis that is benign in nature but requires complete excision with negative margins. Given the unclear diagnosis, multidisciplinary consensus was to manage as DFSP.

The patient will be followed clinically with examinations every 3 months. His first postoperative visit showed no sign of recurrence but a significantly contracted scar causing lateral penile curvature (Fig. 3b). This will be monitored and cosmetic reconstruction is planned for 6–18 months after initial resection.

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Figure 1. Preoperative examination showing 5 mm smooth, erythematous, and raised penile skin lesion. (Color version available online.)

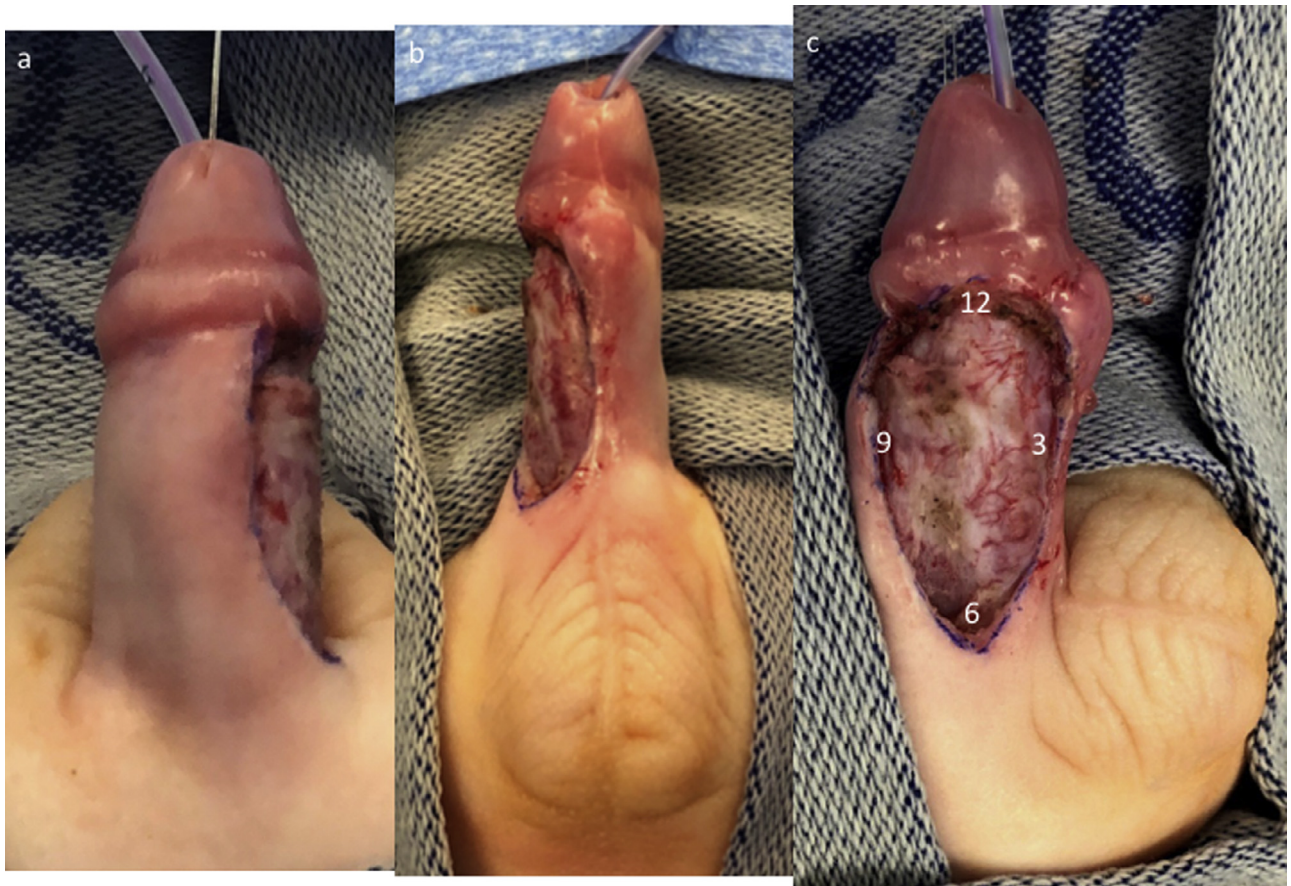


Figure 2. Intraoperative photos showing extent of surgical resection from dorsal (a), ventral (b), and right lateral (c) aspect of penis, including the complex orientation legend used intraoperatively (c). (Color version available online.)



Figure 3. Postoperative appearance at 10 days (a) and 3 months (b). (Color version available online.)

DISCUSSION

DFSP is a low-grade sarcoma of fibroblast origin arising from the dermis. The lesion typically presents as a firm, painless plaque with a violaceous, red-blue, or brown appearance. It is usually fixed to the skin, but not deeper. As the tumor enlarges, it can develop a multinodular appearance within the plaque.⁷ The tumor initially progresses slowly over time, then enters a rapid growth phase and can become locally aggressive. Thus, diagnosis tends to be late with tumors large in size. Misdiagnosis is common, leading to delayed treatment.⁸

The characteristic t(17,22) mutation has been identified in >90% of cases.⁷ This mutation results in overexpression of a platelet-derived growth factor receptor β fusion protein which acts on a tyrosine kinase receptor promoting local growth.⁹ The tumor spreads with asymmetric finger-like projections extending from the primary lesion in all directions.¹⁰ These extensions are often subclinical and difficult to identify macroscopically. Although known to be locally aggressive, metastases have been reported.⁷

National Comprehensive Cancer Network (NCCN) and European Dermatology Forum (EDF) guidelines agree initial management of suspected or biopsy proven DFSP revolves around local control with complete surgical resection.^{5,6} Considering the complexity of tumor extension and characteristic infiltrative growth, microscopic examination of all tissue margins is crucial to ensure complete removal. Local recurrence rates have been reported as high as 50% with incomplete resection.⁸ Surgical excision is also the mainstay of treatment for lesions with positive margins or recurrent tumors when possible. Options for surgical management currently include Mohs micrographic surgery or wide local excision with at least

2 cm to investing fascia with negative histologic margins.⁵ Wide surgical resection has been associated with higher rates of recurrence following excision in comparison to Mohs.¹¹ However, if proceeding with re-resection, as in this case, Mohs can be technically difficult as there is no initial lesion or pathology to follow with serial shavings and there are significant postoperative changes.

For initially unresectable tumors, metastatic disease, or unresectable recurrences, nonsurgical management options per NCCN and EDF guidelines include radiation therapy and systemic therapy with imatinib. Radiation is the preferred option of adjuvant treatment following resection with positive margins and unresectable disease.¹² Imatinib, a systemic tyrosine kinase inhibitor, is generally reserved for disease recurrence or metastases, and has shown up to 50% response rate.¹³ The drug has also been studied for neoadjuvant therapy with favorable results in a phase II clinical trial of 16 patients.¹⁴

Although the age of presentation varies widely, DFSP most frequently occurs in the middle-aged population and is a rare childhood malignancy.¹ Due to its relative infrequency, suspicious skin lesions are often confused with more common ailments including fibromas, vascular malformations, and atrophic plaques.¹⁵ Additionally, young patients may demonstrate atypical variations in clinical presentation such as solitary tumors, erythematous patches, and sclerotic nodular plaques further confounding the diagnosis. Considering the initial propensity for slow growth in DFSP, it is thought many tumors diagnosed later in life may have actually presented in childhood.¹⁵

DFSP most commonly arises on the trunk, and the reported incidence on the genitals is only 1%.^{1,8} There are few case reports of penile DFSP, but the site remains extremely uncommon.²⁻⁴ To our knowledge, there is one

other case reported of penile DFSP in a 9-month-old infant.⁴ Surgical resection with adequate margins and considerations for reconstruction can be technically difficult when concerning the genitalia.

CONCLUSION

DFSP is an uncommon soft tissue malignancy that can be difficult to distinguish from other benign pediatric skin lesions and is very rarely seen on the genitalia. The tumor is known to be locally invasive, and metastases are rare. The gold standard in management is complete surgical resection, which can be complex considering location and size, particularly in the pediatric patient. This case highlights the challenges in diagnosis and management of penile DFSP in the pediatric patient considering variability in presentation and complexities of genitourinary reconstruction in a child.

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