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Scrotal fibrous hamartoma of infancy: A case report and literature review of a rare tumor of the genitourinary tract☆

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ABSTRACT

Fibrous hamartoma of infancy (FHI) is a rare, benign soft tissue lesion observed in infants characterized histologically by triphasic appearance of bland fibroblastic fascicles, mature adipose tissue, and nodules of primitive myxoid mesenchyme. Preoperative and intraoperative recognition of FHI presents a significant diagnostic challenge due to nonspecific imaging findings and its histologic similarities to alternate benign and malignant entities. Management requires complete local excision and clinical follow-up to monitor for recurrence. Here, we present the diagnosis, management, and two-year follow-up of a 13-month-old boy with a scrotal FHI in addition to a comprehensive literature review of this entity.

1. Introduction

Fibrous hamartoma of infancy (FHI) is a benign but rare soft tissue lesion most commonly diagnosed in patients under 2 years of age, characterized by a classic triphasic histologic appearance of bland fibroblastic fascicles, mature adipose tissue, and primitive myxoid mesenchyme.[1] A case of an infant with this rare lesion in the scrotum is presented, including diagnostic workup, operative intervention, and 2-year follow-up. This is followed by literature review of this rarely-described entity.

2. Case presentation

A 13-month-old previously healthy boy was referred to pediatric urology for a right scrotal mass. Concern for development of a second testis in the right hemi-scrotum that was not present at birth was first noted at 9 months of age. There was no history of scrotal or abdominal pain, scrotal wall changes, trauma, or infection. At one year, the pediatric urologist palpated a non-tender, non-erythematous, subcentimeter, cord-like mass in the right mid-scrotum with overlying induration. The right and left testis were normal and descended. Ultrasound demonstrated a scrotal tubular soft tissue structure with internal vascularity inferior to the right testis (Fig. 1a/1b). Pelvic MRI identified an elongated structure with homogenous enhancement, extending to the distal scrotum with overlying skin thickening (Fig. 1c).

In the operating room, the right testis was delivered through a transverse inguinal incision. A 5mm gubernacular mass was adjacent to but non-contiguous with scrotal skin, testis, or spermatic cord. Wide excision via scrotal inversion to the inguinal incision was performed without scrotal skin violation, followed by orchiopexy. Two frozen sections were examined, with one reported as benign and the second inconclusive. Pathologic examination revealed bland fibroblasts/myofibroblasts, mature adipocytes, and primitive myxoid mesenchyme, consistent with FHI. No atypical features were identified (Fig. 1d).

Two-week postoperative exam demonstrated orthotopic bilateral testes, without palpable scrotal mass. Postoperative follow-up at 3 months, 1 year, and 2 years revealed no palpable masses on exam with normal testicular echotexture and volume on serial ultrasounds (Table 1).

3. Discussion

This case demonstrates a rare paratesticular mass etiology that exists within the differential diagnosis of infantile scrotal lesions. Regardless of age, the majority of paratesticular masses are benign.[3] Less is known regarding the relative frequency in infancy. In a review of 1133 cases

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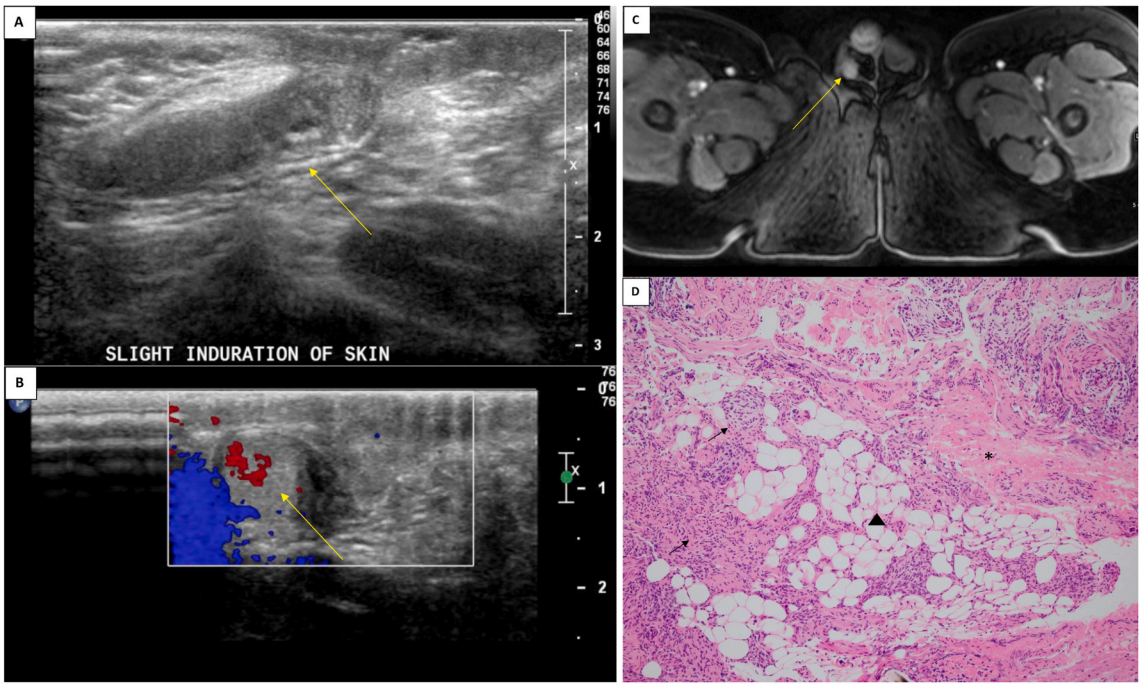

Fig. 1. Radiologic and pathologic images of 13-month-old patient with scrotal fibrous hamartoma of infancy.
a.) Longitudinal greyscale scrotal ultrasound demonstrating tubular soft tissue structure in the scrotal sac inferior to the right testis.
b.) Transverse ultrasound image of the right scrotum at the palpable abnormality demonstrates an echogenic lesion with internal Doppler flow.
c.) Axial T1 FS postcontrast image demonstrates homogenous enhancement of the lesion with thickening of the adjacent skin.
d.) Hematoxylin-eosin (H&E)-stain reveals classic triphasic morphology composed of bundles of bland fibroblasts/myofibroblasts (asterisks), admixed with mature adipocytes (triangle) and primitive myxoid mesenchyme (arrow).

Table 1
Patient's ultrasound findings pre- and post-operatively, demonstrating no evidence of recurrence.

Time	Left Testis (Unaffected)		Right Testis (Affected)		Other Findings
	Testis Length (cm)	Testis Volume[a] (mL)	Testis Length (cm)	Testis Volume[a] (mL)	
Pre-operative	1.3 × 0.7 x 0.7	0.5	1.8 × 0.8 x 0.9	0.6	Right scrotal sac with thickened, tubular soft tissue structure with internal vascularity; overlying skin thickening
3 months postoperatively	0.7 × 0.9 x 0.4	0.7	1.5 × 0.8 x 1.2	1.0	Normal with no focal mass
1 year postoperatively	1.6 × 0.8 x 0.8	0.7	1.6 × 0.7 x 1.1	0.9	Normal with no focal mass
2 years postoperatively	1.1 × 1.6 x 0.7	0.9	0.9 × 0.6 x 1.7	0.7	Normal with no focal mass

[a] Testicular volume was calculated utilizing the formula of Lambert.[2]

across all ages, 67% were benign with the most common etiologies being lipomas and adenomatoid tumors.[3] Malignant paratesticular etiologies (33%) included rhabdomyosarcoma, leiomyosarcoma, and fibrosarcoma.[2] Evaluation of an infantile scrotal mass must consider potential malignant and benign etiologies to provide optimal surgical and postoperative management.

The largest case series of pediatric FHI to date details 145 cases from four institutions.[1] The mean age of diagnosis was 15 months (range birth-14 years) and the mean tumor size was 3cm (0.4–17cm). There was a male predominance (2.7:1). The most common sites were axilla (17%), back (16%), upper arm (14%) and scrotum (9%). Following primary excision, only two recurrences (3.8%) occurred among the 52 patients who had follow-up (median follow-up 8 months).[1]

To comprehensively identify FHI cases in the GU tract, "Fibrous hamartoma of infancy in the genitourinary tract" was searched in PubMed, Science Direct, Scopus, and Google Scholar. Eleven peer-reviewed, English language or translated articles reporting FHI in the GU tract were identified. This included 1 case series (15 cases) and 10 case reports. Altogether, 96.2% of cases identified were benign without reported recurrences.[3] However, locally aggressive features have been described,[3] highlighting the importance of complete resection and postoperative local clinical monitoring for recurrence. Table 2 outlines these GU FHI cases.[4–14]

FHI is characterized by classic triphasic histologic appearance of bland fascicles fibroblastic/myofibroblastic cells, mature adipose tissue, and nodules of primitive myxoid mesenchyme.[1] Twenty-five percent of cases show areas resembling giant cell fibroblastoma with hyalinized zones with cracking artifact which form slit-like spaces lined by flattened tumor cells.[1] Immunohistochemistry shows variable expression of smooth muscle actin in the fibroblastic and primitive mesenchyme areas with S100 protein expression in the adipocytes.[1] Primitive mesenchyme and areas with giant cell fibroblastoma-like morphology are CD34 positive.[1] Intraoperatively, sufficient tissue sampling is important to capture all three features, supporting a diagnosis of FHI versus other fibromatoses. Primitive myxoid mesenchyme may resemble rhabdomyosarcoma, a malignant tumor treated aggressively with radical

Table 2
Case reports yielded from search of "fibrous hamartoma of genitourinary tract" in PubMed, Google Scholar, Scopus, and Science Direct.

Authors	Location	Age of Diagnosis	Therapy	Follow-up	Local recurrence
Popek et al[3]	5 Inguinal region 5 Scrotum 1 Spermatic cord 1 Perineum 1 Labium majus 1 Suprapubic region 1 Pubic region	Median age 10 months	Local excision	Not specified	1 (6.7%)
Sengar et al[4]	Scrotum	12 months	Orchiectomy	1 year	None
Monajemzadeh et al[5]	Vulva	18 months	Local excision	2 years	None
Groisman et al[6]	Scrotum	8 months	Local excision	1 year	None
Stock et al[7]	Scrotum	15 months	Local excision	3 months	None
	Labium majora	6 years old	Local excision	18 months	None
Thami et al[8]	Scrotum	15 months	Local excision	6 months	None
Harris et al[9]	Scrotum	12 months	Local excision	8 months	None
Ritchie et al[10]	Spermatic cord	10 months	Local excision	6 months	None
Stepančec et al[11]	Scrotum	8 months	Wide local excision	1 year	None
Kim et al[12]	Scrotum	5 months	Local excision	None	Not assessed
Kilitci et al[13]	Testis	7 years old	Orchiectomy	None	Not assessed

orchiectomy followed by retroperitoneal lymph node dissection. However, the two other histologic features and absence of myogenic markers like myogenin and MyoD1 aids in distinguishing the two entities.[15] Pathologic diagnosis represents a diagnostic challenge due to similarities to other benign and malignant lesions.

Preoperative imaging likewise provides a diagnostic challenge, and is nonspecific to this entity. On ultrasound, FHI can demonstrate heterogeneous echogenicity similar to other soft tissue tumors; however, a "serpentine" pattern of intervening hypoechoic portions in the hyperechoic mass has been described.[16] MRI demonstrates areas of low-signal intensity in fibrous components as well as high-signal intensity on T1-and T2-weighted images in fatty components; the differential for this appearance includes lipoma, lipoblastoma, and involuting hemangioma.[16] Imaging findings of FHI are often nonspecific and require further validation within the GU tract.

Intraoperative diagnosis based on gross appearance and frozen section is likewise limited. Due to its benign nature, for most paratesticular cases a local complete resection may be an adequate treatment strategy. However, uncertainty of the diagnosis pre- and intra-operatively can result in unnecessary radical orchiectomy. Postoperatively, unless locally invasive or an incomplete resection was performed, monitoring for local recurrence may be a viable strategy with annual ultrasounds and physical examinations after an initial postoperative analysis within the first year. However, with no current practice guidelines for obtaining intraoperative pathology and monitoring postoperatively, additional multi-institutional case series for FHI in the GU tract are needed to guide management.

4. Conclusion

FHI is a rare, benign entity that should be considered in the differential diagnosis of infantile scrotal and paratesticular masses. Inconclusive preoperative and intraoperative findings present a diagnostic challenge that can be mitigated through increased awareness of the distinctive imaging and pathologic features of FHI. While excision is curative in most cases, local recurrences have been reported. Further study of FHI in the GU tract is needed to guide management of this entity.

Consent

Informed consent was obtained from parents of patient described.

Declaration of competing interest

Authors report no conflicts of interest.

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