

Case – Antenatal ultrasound diagnosis of a giant penile inclusion cyst

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KEY MESSAGES

- Penile and preputial cysts are uncommon and although unlikely to cause functional impairment, can be psychologically distressing.
- Penile and preputial cysts may be congenital or acquired.
- Prenatal ultrasound can detect genitourinary anomalies, which warrants further counselling and evaluation.

INTRODUCTION

Penile and genitourinary abnormalities are common, accounting for approximately one-third of all congenital malformations.¹ Congenital cutaneous penile cysts are usually identified postnatally, localized to the prepuce, and are frequently asymptomatic; however, in some instances, they may impair urination or sexual function or create psychological distress or cosmetic concerns for patients and families.² The majority of congenital preputial cysts are self-limited and may be observed for spontaneous resolution, although some may ultimately benefit from surgical intervention.³ Herein, we present the first documented case of a newborn who on antenatal ultrasound was found to have a giant cystic structure of the distal penis, which was confirmed as a benign congenital cyst.

CASE REPORT

A five-day-old healthy male, born full-term via Cesarean section, was evaluated for a large cystic mass at the end of the phallus, first identified at the 23-week prenatal ultrasound, where it initially measured 8 mm. By the 38-week ultrasound, it had enlarged significantly to 22.2 × 20.8 × 16.6 mm (Figures 1a, 1b). At clinical presentation, he was voiding spontaneously without difficulty, and there was no evidence the structure was causing discomfort. Examination revealed an otherwise healthy neonate, with the only demonstrated anomaly being a 2.5 cm circumscribed cystic structure at the distal, dorsal aspect of the foreskin just left of midline (Figure 1c). The foreskin was easily reducible. The meatus was orthotopic, and both testes were normally descended. Ultrasound revealed a mildly complex cystic

structure at the tip of the penis with a difficult-to-visualize distal urethra (Figure 1d).

After counselling, the family elected to proceed with observation. By history, the mass continued to enlarge before rupturing spontaneously at one month of age. The parents reported drainage of white fluid from the cyst. At two-month followup, spontaneous rupture was confirmed, and the family elected to proceed with elective circumcision.

At nine months of age, cyst excision and phalloplasty were performed. Upon initial dissection, the cyst was pedunculated (Figures 2a, 2b). The cyst and stalk were external to the corpora and urethra. Although the cyst was dorsal, the stalk appeared to connect to the urethra ventrally. It was excised and sent for pathological analysis. A leak test was performed, without evidence of communication between the stalk and the urethra. Retraction of the foreskin revealed a 90-degree, counter-clockwise penile torsion. This was fully identified after the penis was degloved due to mass effect from the cystic structure (Figure 2c). On further evaluation, torsion was noted to be due to rotation of the left corporal body posterior to the spongiosum. The dartos fascia was fixed to the corporal bodies at the penoscrotal junction to correct the torsion and penile concealment. Subsequently, the dorsal foreskin was incised to the level of the mucosal collar, and the preputial flaps were rotated ventrally and approximated at the midline to recreate the median raphe. Finally, the skin

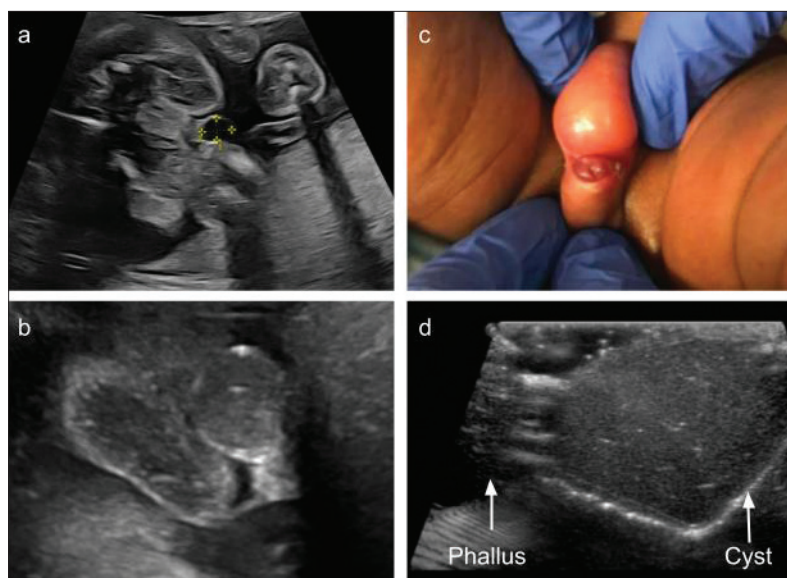


Figure 1. Preputial cyst detected on (a) 23-week prenatal ultrasound; (b) 38-week prenatal ultrasound. (c) Exam on day of life 5 demonstrated a 2.5 cm dorsal cyst and orthotopic meatus. (d) Ultrasound on day of life 5 with cystic structure at distal end of penis.

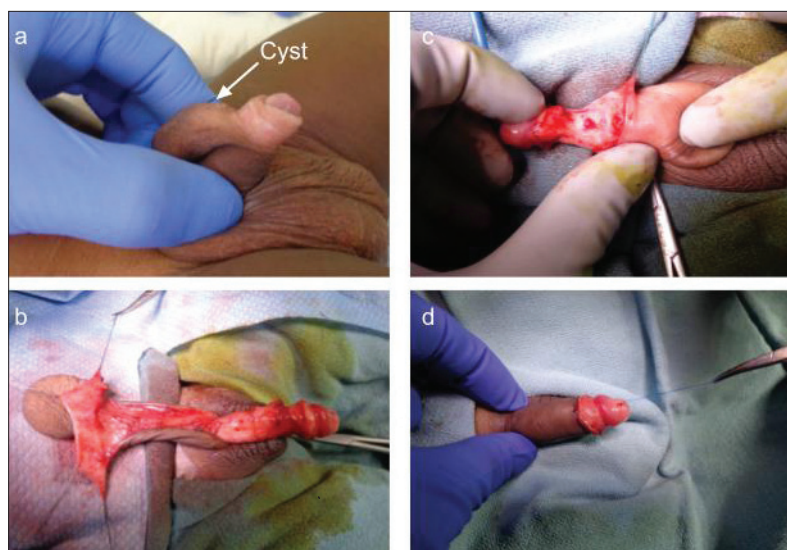


Figure 2. (a) Preoperative examination. (b) Dissection and penile degloving demonstrated the cystic structure to be on a stalk and (c) with a 90-degree, counter-clockwise penile torsion. (d) Postoperative images with a straight phallus and circumcised appearance.

was anastomosed to the mucosal collar to complete the phalloplasty (Figure 2d).

At his six-week postoperative visit, incision sites were healing well, with no evidence of recurrence, and he was voiding without issue (Figure 3). Final pathology demonstrated squamous epithelial lining and chronic inflammatory changes consistent with an inclusion cyst. The stalk contained urothelial lining. All specimens were benign (Figure 4).

DISCUSSION

Penile cysts are uncommon genitourinary malformations that are often not detected until adolescence or adulthood and are hence acquired rather than congenital.³ Such cysts are typically located on the ventral midline of the penis. Acquired (or false) cysts include preputial Epstein pearls, trichilemmal cysts, epidermal inclusion cysts, and smegma cysts.⁴ Smegma cysts, or 'smegmomas,' are the most common penile cystic lesion. They form due to an accumulation of smegma beneath an unretractable foreskin and are often yellow in color.⁵ While smegma cysts typically occur secondary to physiological adhesions present at birth, epidermal inclusion cysts typically form following penile surgeries, including circumcision and hypospadias repair. Epidermal inclusion cysts may also be congenital due to the sequestering of epidermal rests during development.³

Congenital penile cysts include dermoid cysts, median raphe cysts, parametarial cysts, mucoid cysts, pilosebaceous cysts, and juvenile xanthogranulomas.⁴ Dermoid cysts are typically asymptomatic, small, and firm.⁶ Median raphe and parametarial cysts exist along the same spectrum and are thought to result from incomplete closure of the genital or urethral folds; they can present anywhere from the urethral meatus, along the ventral penile shaft, or more proximally towards the scrotum.² Mucoid cysts are also primarily present on the ventral surface, although they occur more distally on the glans and originate from ectopic urethral mucosa.⁷ Fortunately, penile cystic lesions are not suspected of having malignant potential, allowing observation as a potential management course if the patient is not bothered by the lesion.⁸ For those managed surgically, complete excision should be pursued, as there is a risk of recurrence.³

On our patient's initial presentation, he had no discomfort or voiding concerns due to this cystic mass. Hence, given the high prevalence of smegma cysts and their spontaneous resolution, a period of observation was deemed appropriate to avoid unnecessary surgical intervention. The cyst's subsequent enlargement and spontaneous rupture may have resulted from the accumulation of smegma.

Ultimately, the parents elected to proceed with phalloplasty due to the size, location, and persistence of this structure and its potential psychological distress. Since penile torsion may be related to the abnormal attachments of the skin and dartos fascia to the corporal bodies, it stands to reason that the co-presence of this problem may therefore be the result of the torsional forces exerted by this cyst during the development of the prepuce and underlying

corpora.³ It is interesting to note that the location of the cyst was off midline, to the left (Figures 1c, 2b). Pathological analysis noted urothelial lining in the stalk, with squamous epithelial lining of the cyst. Since the urothelial lining was localized to the stalk, external to the corpora and urethra, and exhibited no communication with the urethra, we hypothesized this was due to metaplastic changes rather than a median raphe cyst, duplicated urethra, or prior communication with the urethra.

Unique to this patient was the antenatal detection of his phallic abnormality. In our literature review, we could only identify a single case describing a prenatal penile cystic lesion on ultrasound, although this resolved before birth and confirmatory exam.⁸ With the increasing use and quality of antenatal ultrasound, the detection of similar congenital genital malformations will likely occur with increased frequency.

The results of anatomy ultrasound scans of fetal genitalia in the second and third trimesters can assist in guiding the counselling of families and optimize postnatal evaluation and management.⁹ For example, hypospadias is diagnosed with increasing frequency during the third trimester, with findings including chordee and a deviated urinary stream on Doppler ultrasound, or in the most severe cases, the 'tulip sign'; isolated chordee has also been identified on prenatal imaging.^{10,11} One study by Epelboym et al demonstrated a greater than 80% positive predictive value for antenatally diagnosed hypospadias.¹²

CONCLUSIONS

Congenital penile cysts make up a small proportion of genitourinary malformations that present at birth. Most of these lesions have minimal to no effect on function, along with no known malignant potential. The high prevalence of smegma cysts and their spontaneous resolution warrants an initial observation period. In instances where the anomaly persists, or in the case of our patient, where circumcision was desired, surgical intervention can be offered; however, care must be taken to proceed with complete excision to decrease the risk of recurrence. The prenatal ultrasound detection of an isolated congenital penile cyst is a rarity that requires appropriate, individualized evaluation and parental counselling antenatally and postnatally.

COMPETING INTERESTS: The authors do not report any competing personal or financial interests related to this work.

This paper has been peer-reviewed.

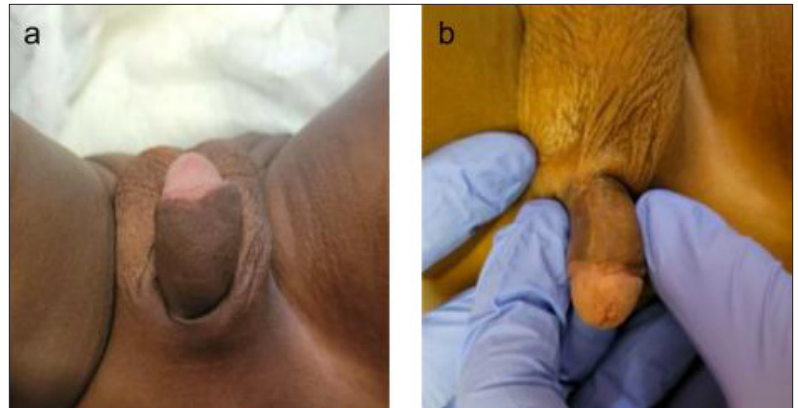


Figure 3. Evaluation 5 weeks postoperatively with well-healing incision (a) dorsally; and (b) ventrally.

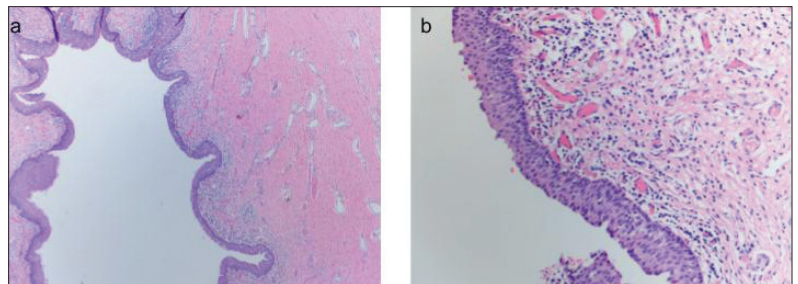


Figure 4. (a) Benign cyst lined by squamous epithelium and mild chronic inflammation (H&E x 100). (b) Stalk of the cyst lined by urothelium (H&E x 200).

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