VISUAL DIAGNOSIS

Apparent Clitoromegaly in Healthy Female Newborn

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ABSTRACT

Cysts of the external female genitalia are uncommon findings and can be either congenital or acquired. Congenital cysts of the external female genitalia may be discovered from the initial newborn examination and cause concern for clitoromegaly or ambiguous genitalia. We describe a clitoral cyst presenting as apparent clitoromegaly in a newborn; this is the third reported case in the literature. We also review the differential and evaluation for clitoromegaly in a newborn. We present a case of a 1-day-old newborn, with concern for clitoromegaly on initial newborn examination. Multiple anatomy ultrasounds were performed during pregnancy and no genital abnormality was noted. On physical examination, a 1 × 1 cm cyst was found on the clitoral hood obstructing the clitoris and vaginal opening. There were no signs of virilization or hyperandrogenism on examination. The cyst was aspirated, with clear, serous fluid removed. Normal female anatomy and normal clitoris size were noted after aspiration. She had no recurrence of the cyst in the months after discharge. Clitoral cysts of the female external genitalia are rare findings. Barring signs of virilization of hyperandrogenism, conservative management and watchful waiting or cyst drainage are reasonable approaches.

PRESENTATION

A 1-day old female newborn, weighing 2,400 grams and born by spontaneous vaginal delivery at 38 weeks' gestation presents with an apparent enlargement of her clitoris and no other abnormal physical examination findings. The pediatrician is concerned about ambiguous genitalia (see Fig 1).

The infant was born to a 22-year-old mother with no known medical problems and a history of 2 previous unremarkable pregnancies. The mother reports a history of substance abuse, including marijuana and 4-Methylenedioxymethamphetamine (MDMA), commonly known as ecstasy, that was reportedly stopped when she discovered she was pregnant. She had appropriate prenatal care with frequent anatomy scans during pregnancy due to fetal growth restriction. There were no concerns for clitoromegaly, ambiguous genitalia, or genitourinary anomalies noted on prenatal ultrasound scans. The patient's mother does not report any unexpected changes in her body during pregnancy, such as virilization or hirsutism, and she was not

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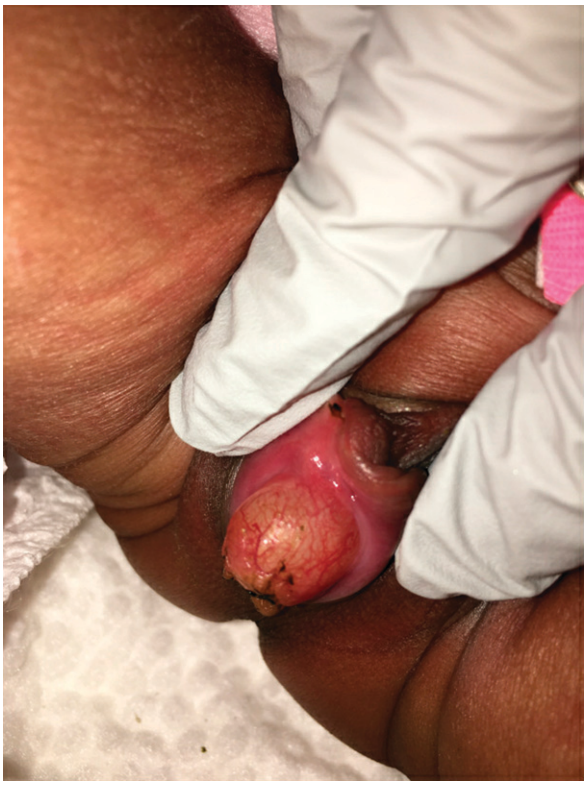

Figure 1. 1 × 1 cm, round cystic lesion anterior to the clitoris, distorting the clitoral hood. The vaginal orifice is unable to be visualized.

taking birth control or progesterone early in her pregnancy. Her only medication was a prenatal multivitamin. The newborn had normal Apgar scores and was admitted to the newborn nursery. She voided spontaneously within the first 24 hours after birth. Physical examination confirms the diagnosis.

DIAGNOSIS

A physical finding of isolated clitoral enlargement with no other sign of virilization or hyperandrogenism is likely nonhormonal in nature. The differential diagnosis of nonhormonal clitoral enlargement in this infant includes apparent clitoromegaly, neurofibromatosis, hemangioma, or cystic lesions of the external genitalia. After examination by a pediatric endocrinologist and a pediatric urologist, the patient was diagnosed with a congenital clitoral cyst.

DISCUSSION

Lesions of the female external genitalia in neonates are rare findings that are reported in the literature by different pediatric subspecialties including urology, gynecology, surgery, and endocrinology. The most common reported masses are vaginal (hymenal) cysts and paraurethral gland cysts. (1) A prospective review evaluating the incidence of congenital cysts of the external genitalia of female newborns over a 2-year period found an incidence of 0.6%, or 1 in 166 female infants. This incidence appears to be higher than what is seen in our clinical experience. It could be simply due to under-reporting by general practitioners as it may not be common practice to thoroughly examine the genital area. (1) Herein, we discuss a case of a congenital, spontaneous clitoral cyst, which has rarely been described in the literature. (1)(2)

Clitoral cysts do not typically involve the urethra or cause anatomic problems with voiding. They are not associated with physical examination findings that may suggest androgen exposure, such as rugated labia, hyperpigmented genitalia, an abnormal vaginal opening or urethral position, or an increased anogenital ratio (>0.5). If signs of androgen exposure are noted, the patient should be evaluated for congenital adrenal hyperplasia immediately as this condition can be life-threatening. Moreover, a pediatric urologist should be consulted if there any anatomic concerns on examination or if the infant is unable to void within 24 hours after birth.

The differential diagnosis for clitoromegaly in a newborn encompasses hormonal and nonhormonal causes. (3) Hormonal causes include congenital adrenal hyperplasia, disorders of sexual development, and exogenous or endogenous androgen exposures. Exogenous androgen exposure occurs when mothers take oral contraceptives or progestins during early pregnancy or when mothers are exposed to exogenous hormones by a partner, as might be the case with a family member using testosterone gel. Endogenous androgen exposure can happen in cases such as luteoma of pregnancy or a steroid secreting tumor.

Nonhormonal causes include apparent clitoromegaly, neurofibromatosis-1, and cystic or epidermoid cysts of the clitoris. Another common entity is "apparent clitoromegaly," common in preterm females and small for gestational age (SGA) infants. In these cases, the clitoris appears relatively large secondary to the small pubic fat pad and labia majora in SGA infants. Notably, even in SGA infants, a normal clitoral length should still be less than 1 cm. Infants with neurofibromatosis-1 can have clitoromegaly due to a neurofibroma of the clitoris. Hemangiomas and cystic lesions of the clitoris can also cause apparent clitoromegaly.

Most clitoral cysts described in the literature are secondary to trauma, typically due to female genital mutilation. However, cases of spontaneous nontraumatic and

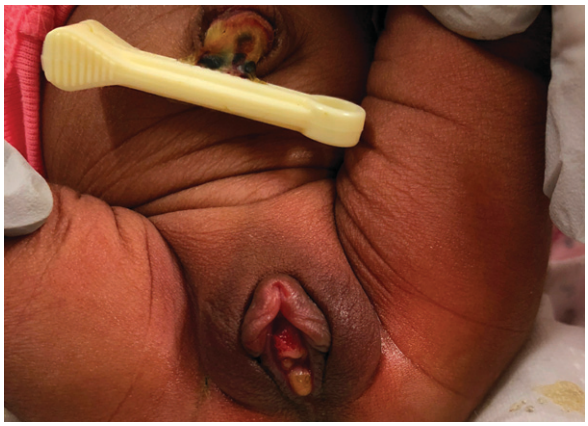

Figure 2. Normal female external genitalia after cyst drainage. Note no hyperpigmentation, no clitoromegaly, and normal vaginal opening.

congenital clitoral epidermal inclusion cysts are rare. (4)(5) Previously reported congenital clitoral cysts were epidermoid in nature, whereas our patient's cyst was simple, round, fluid filled in appearance and on palpation. Surgical intervention has been reported, but there is no consensus on the optimal management of these lesions. One described clitoral cyst treated with surgical resection had not resolved after 9 months of observation. (2) In addition, most patients reported in the literature underwent additional evaluation including imaging, endocrine labs, and karyotyping to rule out other pathologies. Watchful observation and limited laboratory testing may be reasonable options in recognized uncomplicated clitoral cyst of the newborn.

PATIENT COURSE

The pediatric endocrinologist evaluated the child and suspected a simple clitoral cyst, and the pediatric urologist agreed with the diagnosis. Needle aspiration was performed at bedside by the pediatric endocrinologist. A small-gauge needle was inserted into the superior left aspect of the cyst, and 1 mL of serous fluid was drained. The newborn urinated at the start of the procedure and during cyst decompression. Normal external genitalia, including clitoral tissue and size, were easily visualized after the procedure (Fig 2). Given the normal appearance of the external genitalia after drainage of the cyst, no further imaging or evaluation was indicated.

Our patient was lost to follow-up by both pediatric endocrinology and pediatric urology. However, she was admitted to the hospital at 6 months of age for a respiratory infection. At that time, there had been no recurrence of the cyst. The grandparents reported no changes in her genitalia, and examination at that time was normal (Fig 3).

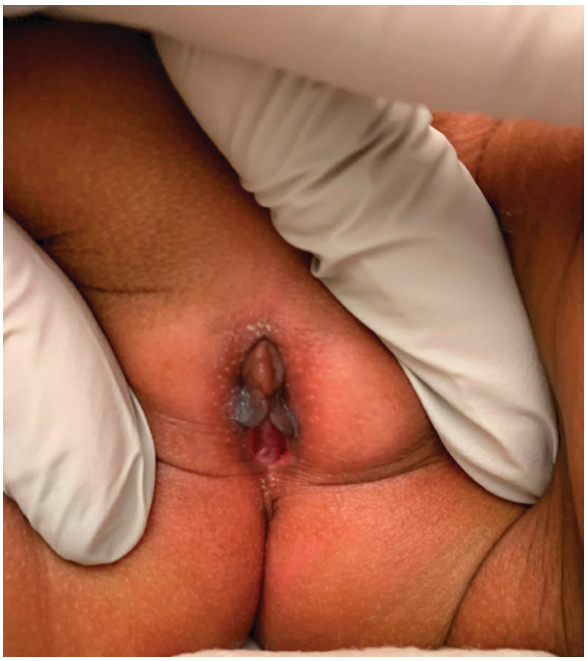

Figure 3. Normal female external genitalia at 6 months of age.

Summary

- Cysts of the female external genitalia are likely under reported in the literature. A contributing factor may be lack of routine genital examination of the female neonate.
- Congenital cysts of the female external genitalia are uncommon, and many cases will resolve spontaneously.
- Careful inspection of the genitalia is an important aspect of a routine neonatal physical examination. Congenital malformations can be diagnosed quickly and should have early subspecialty evaluation.
- Consult subspecialists for physical examination findings suggestive of abnormal androgen exposure (hyperpigmentation, rugae of the labia, or anogenital ratio > 0.5), anatomic anomalies, or inability to void in the first 24 hours after birth.

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