A newborn female infant presented to the outpatient pediatric office for an initial weight visit on her third day of life. She was born full term, appropriate for gestational age, via spontaneous vaginal delivery to a 21-year-old mother. The mother was blood type A+, HIV negative, rubella immune, hepatitis B surface antigen negative, rapid plasma reagin nonreactive, and Group B streptococcus positive. The mother received antepartum antibiotics prior to delivery. The baby’s APGAR scores were 8 at 1 minute and 9 at 5 minutes. The baby did well in the newborn nursery, and had been breast-feeding and voiding and stooling several times per day. It was noted in the nursery that she had an unusual appearance of her labia.

On examination, the baby was awake and appropriate for age. She had no dysmorphic features, and her palate was intact. There was no heart murmur. Her abdomen was soft and nontender, with no organomegaly or mass. Her umbilical stump was still attached. Her genitourinary examination revealed normal labia majora. There appeared to be mucosal tissue from the fossa navicularis extending posteriorly through the posterior fourchette ending just anterior to the anal opening. There was no clitoromegaly. There was a sleeve hymen. The anal sphincter surrounded the anal opening normally (Figure 1).

For diagnosis, see page e9

Editor’s note: Each month, this department features a discussion of an unusual diagnosis. A description and images are presented, followed by the diagnosis and an explanation of how the diagnosis was determined. As always, your comments are welcome via email at pedann@Healio.com.
DISCUSSION

Failure of midline fusion, also known as a perineal groove, can be a mimic of pediatric sexual abuse. It is a congenital defect leading to mucosal exposure anywhere along the midline from the fossa navicularis to the anus. The embryologic pathogenesis is unclear, but is thought to arise from failure of fusion of the medial genital folds in early fetal development. This defect typically resolves spontaneously over a period of years, so conservative management is recommended. It is important to identify this defect during early preventive care visits to eliminate any future concerns of trauma, especially in the absence of any relevant history. Accurate documentation is vital to avoid misdiagnosis of sexual abuse.

Child sexual abuse remains a significant and underreported problem. In 2013, there were approximately 3.5 million referrals in the United States to child protective services for alleged maltreatment of a child. Of these referrals, approximately 2.1 million were ultimately screened as appropriate for a child protective response, and of these, 9% of victims were sexually abused. Abused children are more often victimized by family members than strangers. Awareness regarding normal genital variants, including this child’s failure of midline fusion, and congenital anomalies is important to avoid erroneously identifying sexual abuse.

If in doubt about a midline fusion defect, reexamination of the child after 1 to 2 weeks should reveal unchanged examination results.

CONCLUSION

This child has been followed in our primary care pediatric practice since birth. Her genital examination has remained unchanged, and she remains asymptomatic. Shortly after birth, she was evaluated by physicians in Pediatric Surgery, who concurred that this was a midline fusion defect and recommended no specific therapy.

REFERENCES