Superior vesical fissure concealed by giant omphalocele: A case report

Danial Hayek*, Eric Massanyi, Melissa Mancuso, Katherine Krepkovich, Aaron Garrison

Akron Children’s Hospital, Akron, OH, USA

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ABSTRACT

Omphalocele is an abdominal wall defect characterized by exteriorization of abdominal viscera that is covered by a membrane. Omphalocele has a well-known association with chromosome abnormalities. Here we present a case of a superior vesical fissure (SVF) initially disguised as a giant omphalocele in a newborn with trisomy 18. We also show unique prenatal imaging and discuss the importance and limitations of prenatal genetic testing.

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The pathogenesis of omphalocele is still controversial and is associated with chromosomal abnormalities including trisomies 18, 13, 21, and 45X as well as other genetic conditions [1]. Omphalocele has been associated with a wide range of exstrophy conditions ranging from mild pseudoxestrophy to severe cloacal exstrophy [2]. Exstrophy variants are rare, but represent a wide spectrum of urogenital anomalies. SVF is a urogenital anomaly typically characterized by the exstrophy of a normal bladder located in the subumbilical region [3,4]. Like other exstrophy variants, SVF is typically associated with both an open bladder and a pubic diastasis. Some, however, believe that SVF represents a completely distinct entity given that patients with SVF have a normal bladder neck and external genitalia [5]. While SVF may be underdiagnosed during the prenatal period, postnatal diagnosis is usually readily apparent secondary to presence of an open bladder on the abdominal wall. The case presented here is unique in that diagnosis of SVF was delayed secondary to the involvement of a large omphalocele. We will review the relevant prenatal imaging and discuss the importance and limitations of prenatal genetic testing.

1. Case report

A 31-week 2700 g male infant with a prenatal diagnosis of giant omphalocele was referred from an outside hospital for surgical management after delivery. A prenatal ultrasound revealed evidence of a giant omphalocele containing liver, bladder, and bowel. In addition, bilateral choroid plexus cysts were visualized. A fetal MRI was significant for polyhydramnios and a large omphalocele containing most of the liver and multiple loops of bowel. The dome of the bladder was seen extending just into the omphalocele (Fig. 1, MRI). Of note, non-invasive (cell-free DNA) prenatal screening results were low risk for trisomy 18. Invasive testing was declined by the family. Postnatal physical examination revealed a 10 × 12 cm omphalocele and undescended testes, but otherwise normal appearing male external genitalia. He had a normal anal opening and passed meconium, ruling out OEIS with imperforate anus. Initially, the patient underwent application of silver sulfadiazine in order to epithelialize the giant omphalocele. On day of life 7, he became acidotic and developed a profound leukocytosis (42.1 × 10^9 per liter). He was promptly taken to the operating room for exploration and planned excision of his omphalocele due to presumed rupture of the membrane with sepsis. At that time, the area of leakage was explored by removing the omphalocele, and we became concerned that the bladder was exteriorized into the inferior portion of the omphalocele. Urologic consultation was obtained. Cystoscopy revealed a

*Corresponding author. 1 Perkins Sq., Ste. 8400, Akron, OH 44308, USA.
E-mail address: dhayek1@gmail.com (D. Hayek).
normal urethra and bladder neck. A guidewire was passed through the urethra and exited through the bladder, confirming a superior vesicle fissure at the base of the omphalocele (Fig. 2). Closure of the bladder was performed in two layers. The abdomen was closed with a large silo sutured to the fascia circumferentially as a temporary dressing, with plans to sequentially obtain abdominal domain prior to more definitive coverage. On post-operative day 16, we observed succes in the silo, so he was taken back for exploration and found to have an intestinal perforation which was repaired. At this time, karyotype results returned consistent with a diagnosis of trisomy 18. The patient remained in the NICU until the family finally elected to withdraw support on day of life 17.

2. Discussion

SVF is a rare urogenital anomaly which some consider to be a part of a large spectrum of bladder extrophy variants [4]. The classical presentation of SVF is an extrophied bladder in the sub-umbilical region concurrent with musculoskeletal abnormalities such as pubic diastasis and low set umbilicus [5]. Our patient was found to have a 25 mm pubic diastasis on subsequent plain film, significantly larger than the width or separation of the symphysis pubis in a healthy neonate, which should be less than 9 mm [2]. SVF is rare but has appeared in both sexes and has been associated in the literature with a number of other congenital malformations or conditions including omphalocele, trisomy 18, and urethral duplication [4,6,7]. SVF is a variant in which pelvic deformity occurs, but usually only the upper bladder is open near the umbilicus. The case presented here is unique because the diagnosis of SVF was delayed due to an overlying giant omphalocele which prevented the visualization of an SVF on physical exam. Furthermore, the umbilicus was not low set, which is typical of SVF, and the distance between the SVF and pubis was larger than normal. These factors, in conjunction with the giant omphalocele, contributed to the delay in diagnosis.

Bladder extrophy and extrophy variants are typically readily diagnosed by the experienced clinician due to their hallmark characteristics. Classic bladder extrophy is characterized by extrophy of the bladder and epispadias of the urethra. Cloacal extrophy, a more significant birth defect, is characterized by extrophy of both bowel and bladder along with omphalocele, imperforate anus, and oftentimes neurospinal abnormalities [8]. Pseudox extrophy, a mild variant of bladder extrophy, is characterized by the musculoskeletal abnormalities of extrophy (i.e. widened pelvis), but the bladder may be covered with skin, making the diagnosis less obvious [9]. In this case, SVF was not considered until the omphalocele was explored. An OEIS complex was felt to be unlikely given normal bowel function and anatomy.

Additionally, the case herein was a difficult one that highlights the increasing complexity of prenatal genetic screening and testing options and underscores the important role it plays in clinical practice. Given the multiple abnormalities noted on the initial ultrasound, this mother was counseled prenatally that the most likely etiology (also considering her age-related risks) is aneuploidy, specifically trisomy 18. Initial non-invasive (cell-free DNA) prenatal screening results were low risk for trisomies 21, 18, and 13 (via counting-method technology). Given the possibility of, although expected to be rare, false negative results, she was counseled and chose to have blood obtained for repeat cell-free DNA screening (via single nucleotide polymorphism method technology); results were also low risk. She was counseled about the residual risk for trisomy 18 given the clinical findings and continually declined invasive testing via amniocentesis. Due to the complexities in management of this case postnatally, karyotype results were not received until two weeks after initial treatment. Knowledge of the limitations of these types of prenatal screening options are paramount and continued multidisciplinary management essential in the care of infants with complex medical issues, as was seen in this case.

3. Conclusion

SVF is a urogenital anomaly readily diagnosed postnatally upon physical exam. In this case, a concomitant omphalocele prevented visualization of SVF and consequently delayed diagnosis. Prenatal genetic testing is an integral part of clinical practice but does have limitations which need to be considered when counseling patients.

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References