CASE REPORT

Bladder agenesis, ectopic ureters and a multicystic dysplastic horseshoe kidney in one twin newborn with normal amniotic fluid index in utero

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SUMMARY

A monochorionic-diamniotic twin baby presented with intrauterine growth restriction and anuria. The baby was found to have bladder agenesis, a pelvic dysplastic horseshoe kidney, vertebral anomalies, a ventricular septal defect and facial dysmorphisms. It was surprising to find no abnormalities in amniotic fluid indices prenatally, suggesting the possibility of urine output that declined as the pregnancy proceeded. Some degree of twin-to-twin transfusion of amniotic fluid was also possible, which could have rescued the oligohydramnios known to be associated with kidney and urinary tract abnormalities. It was also notable that there was no abnormality in respiratory function, especially since further investigations revealed close to no kidney function. The intrauterine growth restriction (IUGR) along with the multiple anomalies found made the baby unsuitable for dialysis and transplant, and the decision of transition to palliative care was made.

BACKGROUND

Bladder agenesis is a very rare finding, especially in viable male neonates.^{1 2} The presence of ectopic ureters in such a setting is not unusual.² Other common associations with bladder agenesis include the presence of oligohydramnios, cystic kidneys, penile anomalies and pulmonary hypoplasia.³ Over 90% of the reported cases of bladder agenesis have been female neonates,³ and a 30:1 female-to-male ratio has been recorded.⁴ We report a case of one twin live infant without oligohydramnios who was found to have bladder agenesis, ectopic ureters and a multicystic dysplastic horseshoe pelvic kidney.

CASE PRESENTATION

The baby (twin A) was referred to our institute on day of life 2 with anuria, intrauterine growth restriction and no bladder on renal ultrasound. Our patient was the product of a monochorionic-diamniotic twin gestation and was delivered via caesarean section at 35 weeks due to preterm labour, non-reassuring fetal heart tones and breech presentation of twin A. The pregnancy had been complicated by twin gestation, obesity and a concern for bilateral dysplastic kidneys of twin A; however, the amniotic fluid indices remained within normal limits throughout the pregnancy. The sibling, twin B, was reportedly normal, and the maternal history was otherwise insignificant.

At birth, twin A was pink and responsive, with APGARs of 7 and 8 at 1 and 5 min of life,

respectively. Our patient underwent routine resuscitation and was stable on room air for the rest of the hospital course. On examination, our patient was stable and weighed 1717 g (the third percentile on the WHO growth chart). The neonate appeared small, but was well developed and appropriately nourished. Some notable facial dysmorphisms included up-slanting palpebral fissures, a long philtrum and a thin upper lip. A harsh high-pitched murmur was heard at the left lower sternal border, and breath sounds were equal and clear bilaterally. A small phallus was also noted, with no identifiable meatus and undescended testes.

INVESTIGATIONS

A renal ultrasound did not identify a bladder, and attempts to catheterise the neonate failed. An MRI of abdomen and pelvis revealed a dysplastic horseshoe pelvic kidney with no corticomedullary differentiation and cysts of varying sizes, bilateral ectopic ureters and no bladder (figure 1). An X-ray of the chest and abdomen revealed multiple hemi and butterfly vertebral bodies extending through T2–T9 (figure 2), and an echocardiogram revealed a ventricular septal defect and tricuspid valve insufficiency. Our patient was found to be hyponatraemic

Figure 1 MRI of the abdomen and pelvis suggestive of a dysplastic horseshoe pelvic kidney(outlined), a bladder was not identified.

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(120 mmol/L), hyperkalaemic (6.2 mmol/L), and the initial creatinine was 2.4 mg/dL, which increased to 3.1 mg/dL by day of life 3.

OUTCOME AND FOLLOW-UP

The option of peritoneal dialysis followed by need for kidney transplantation and revitalisation of the bladder was discussed. It was not seen to be possible nor in our patient's best interest due to the severe IUGR, renal dysfunction and multiple other anomalies. Our patient was transferred to a hospice facility and passed away a few days later.

DISCUSSION

Bladder agenesis is an extremely rare entity, with 63 cases reported worldwide, with a female-to-male ratio of ~30:1.¹⁻⁴ There have been 24 live births reported as of 2016,¹ and to the best of our knowledge only 2 of these have been male neonates. It has been predicted that ureteric drainage into the female genital tract allows for passage of urine and hence helps maintains renal function.⁴ Bladder agenesis is thought to be compatible with life in a male neonate only if the ureters ectopically drain into the rectum or a patent urachus.⁵ Our patient's radiographic findings were consistent with the absence of a bladder, and ectopic ureters were found to be most likely terminating into the seminal vesicles, if not a blind pouch. Surprisingly, our patient was not reported to have the antenatal ultrasound

Figure 2 X-ray of the chest and abdomen highlighting multiple hemi and butterfly vertebrae in the thoracic spine, extending from at least T2 till T9. The arrows highlight the butterfly vertebrae; the square bracket with the asterisk identifies T2 through T9.

finding of oligohydramnios that is commonly associated with this condition.³

Adequate amniotic fluid production requires normal renal function,⁶ which is difficult to imagine in our patient. Twin A was born with a severely dysplastic horseshoe pelvic kidney with serum creatinine levels indicating close to no renal function. This along with our patient's IUGR lead us to our hypothesis that there must have been either some urine output or some element of twin-to-twin amniotic fluid transfusion, which rescued our patient's in utero oliguria and hence did not manifest as oligohydramnios.

It is well known that amniotic fluid is required for fetal lung maturation. It has also been noted that pulmonary hypoplasia accompanies bladder agenesis in a neonate.⁷ In our patient, the lack of respiratory signs raises the suspicion that there could have been urine output early in gestation, sufficient for lung maturation, which diminished as the pregnancy proceeded. The MRI findings were consistent with a multicystic dysplastic horseshoe pelvic kidney, which strengthens this suspicion as the current literature suggests that this indicates that the fusion of the kidneys happened later in gestation.⁸

It has been suggested that the lack of distension by urine leads to atrophy of the urogenital sinus that results in bladder agenesis.⁴⁻⁹ Other embryologic theories of bladder agenesis have also been suggested, such as disruption of the cranial part of the urogenital sinus, or the lack of fusion of the mesonephric ducts and ureters into the trigon, but all of these still remain unclear.⁵⁻¹⁰

Our patient was also found to have a small phallus and an unidentifiable meatus. These findings correspond with previous reports of penile anomalies in male neonates with bladder agenesis.⁷⁻⁹ Furthermore, our patient could also be diagnosed to have VACTERL association as three of the core anomalies had been identified which therefore meets the definition of the association:¹¹ vertebral (butterfly vertebrae), cardiac (ventricular septal defect) and renal (multicystic dysplastic horseshoe pelvic kidney).

The rarity of this condition and the low long-term survival rates of those neonates that survive the newborn period raise the question of possible interventions in this setting. Our patient had several factors that were not compatible with a favourable outcome. Intrauterine growth restriction, along with the severe renal dysplasia, and the VACTERL anomalies described, made the idea of peritoneal dialysis unsuitable, which therefore made our patient ineligible for renal transplant in the future. In those instances where renal transplant is possible, urinary diversion post-transplant is essential.³⁻⁴ Recently, studies have been published presenting continent urinary diversions with ileal conduits, the results of which have not been described

Learning points

- ▶ Bladder agenesis is an extremely rare condition, with only 63 reported cases worldwide.
- ▶ Most of the reported cases have been female neonates (90%, ~30:1 ratio).
- ▶ Pulmonary hypoplasia and renal dysfunction are the major factors determining the mortality and morbidity of bladder agenesis.
- ▶ The embryology of bladder agenesis is still uncertain.
- ▶ To survive transplant one must be eligible for dialysis.
- ▶ There is a low long-term survival rate for even those neonates that survive the newborn period.

in detail to the best of our knowledge. These conduits are being considered definitive treatment for some cases of bladder agenesis.³

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Competing interests None declared.

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