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Long-term Survival in a Patient with Bilateral Renal Agenesis

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Abstract

Bilateral renal agenesis is a rare condition where live births occur but death ensues shortly after, usually from pulmonary hypoplasia. A fetus with bilateral renal agenesis can avoid pulmonary hypoplasia in situations of a monoamniotic monochorionic (MoMo) twin. Herein we describe a MoMo infant surviving past 3 years. Bilateral renal agenesisis often part of a syndrome.Our patient had VATER (vertebral, anal, tracheo-esophageal and renal anomalies) syndrome, including vertebral abnormalities and imperforate anus. Surviving cases of bilateral renal agenesis are exceptionally rare. A search showed seven cases of infants with bilateral renal agenesis with one surviving beyond 2 months of age. We provide clinical course details of a MoMo infant surviving over 3 years, currently on the transplant wait list. During her first year, in addition to three episodes of peritonitis, she experienced sequelae common to infants with end stage renal disease. One exception was the extent of secondary hyperparathyroidism, which improved only after cinacalcet was begun. Despite this, her general development has been excellent.

Keywords

bilateral renal agenesis; anephric; peritoneal dialysis; neonatal end stage renal disease

Abbreviations

VATER: vertebral, anal, tracheo-esophageal and renal anomalies; DOL: day of life;
Momo: monoamniotic monochorionic

Introduction

Congenital renal anomalies are the most common cause of pediatric end stage renal disease, accounting for 39% of pediatric transplants in 2010 [1]. Bilateral renal agenesis occurs at a rate of 1 in 4,000 births and is incompatible with extended life. Lack of amniotic fluid results in Potter's sequence, including bowed legs, club feet, and widened epicanthic folds with a flat nose and low-set ears as well as other organ malformations like pulmonary hypoplasia, the primary cause of death [2].

Although live births occur, infants with bilateral renal agenesis rarely live more than a day which is why the condition has been described as perinatal lethal renal disease [3]. One case report describes an infant whose mother underwent serial amnio transfusions surviving to at least 9 months post-gestational age [4]. Under the additional condition of a monoamniotic monochorionic (MoMo) twin with normal kidneys, viable lungs may form [4-9]. In MoMo twins, one fetus with kidneys providesthe critical amniotic fluid for both. Although the fetus without kidneys can survive up to the time of delivery and

avoid Potter's sequence anomalies, we found no cases of bilateral renal agenesis in MoMo neonates surviving beyond 2 months of age (see Table) [9]. We describe a bilateral renal agenesis MoMo infant who has thrived because of developments in peritoneal dialysis.

Case Presentation

A 31-year-old, gravida 5, para 3 woman had a MoMo twin gestation. Prenatal ultrasound at 30 weeks showed bilateral renal agenesis with hemivertebrae in twin A. Omphalocele, bladder exstrophy, vertebral anomalies, and congenital heart disease were evident in twin B. The twins were delivered at 34.2 weeks gestation. Twin A had a 2-vessel umbilical cord, imperforate anus and ambiguous genitalia. Twin B had ambiguous genitalia and patent ductus arteriosus. Both twins were chromosome 46 XX with normal chromosomal microarray (Combimatrix, 170k). Twin B expired on day of life 95 from respiratory failure related to pulmonary hypertension.

After delivery, Twin A easily transitioned to room air without signs of pulmonary hypoplasia. On day of life (DOL) 3,surgeons placed a colostomy in the left lower quadrant and a Tenckhoff peritoneal dialysis catheter on the right. On DOL 13 she started low volume continuous peritoneal dialysis.

Our patient experienced typical comorbidities of renal disease. In her first 12 months she had three episodes of peritonitis (*Enterococcus faecalis, Enterobacter aerogenes,* and *Enteroccocusfaecalis*on DOL 151, 190, and 271 respectively).This exceeds typical culture-positive peritonitis in the first 12 months of life according to NAPRTCS (The North American Pediatric Renal Trials and Collaborative Studies), where the 2011 annualized rate is 0.79 infections per year for 0- to1-year-olds [1]. We treated intermittent hypertension with isradipine and daily amlodipine was begun on DOL 225. Perinatal echocardiogram was unexceptional. Metabolic acidosis was treated with supplemental bicarbonate starting DOL 68. The patient required epoetin alpha beginning DOL 4.Her secondary hyperparathyroidism resisted traditional therapy with calcitriol. Secondary hyperparathyroidism improved with cinacalcet, which was started on DOL 248 with serum parathyroid hormone (iPTH) of 1834 pg/ml. Our patient's electrolyte abnormalities included hyponatremia, hyperkalemia, and metabolic acidosis. We supplemented sodium chloride and bicarbonate as necessary. Hyperkalemia resisted low potassium formula and was treated by chelating her formula with sodium polystyrene sulfonate.

Nutritionally she reached full enteral feeds on DOL 60, having been fed with donor breast milk, then Similac 60/40 and Good start Gentle (as serum potassium permitted). A gastrostomy tube was placed on DOL 101.

Birth parameters at birth were weight 1.95 kg (10-50th percentile), length 41 cm (below 3rd percentile), and head circumference 30 cm (3rd-10th percentile) according to the Fenton growth chart [9]. With appropriate nutrition, her weight reached 8.8 kg (10-25th percentile) at 12 months of age. At that time her length was 65 cm and head circumference 43 cm, both below the 2nd percentile (according to World Health Organization growth chart) [11].

Development has been promising. Motor skills were assessed by Peabody Developmental Motor Scales [12]. At 7 months (6 months corrected age), her grasp was at the age equivalent of 5 months while visual motor integration was at 7 months, and gross motor function was at a 5-month level. As for social

social development, she performed many skills at the 6-9 month level, but language expression and comprehension were at a 3-6 month age level. She was reassessed at 13 months (12 months corrected age), and both grasp skills and visual motor integration were at 10 months. Gross motor function, reflexes and stationary skills were at the level of 9 months of age, locomotion at the 6-month level, object manipulation at 12 months, and speech at 9 months. Our patient demonstrated age-appropriate social interaction.

After 1 year of age, our patient developed a seizure disorder at 23 months of age controlled with levetiracetam. She was treated by orthopedics for osteopenia with ankle/foot orthosis and began ambulating with the help of a walker at 45 months old. She underwent ileal conduit creation in preparation for renal transplant and has had 4 cases of peritonitis over the past 30 months since initial hospital discharge.

Discussion

Historically, bilateral renal agenesis is fatal in the perinatal period. In the rare case of MoMo twins, where production of urine by one twin protects the other from the effects of inadequate amniotic fluid (especially pulmonary hypoplasia), neonates have a chance for long-term survival. Frequently bilateral renal agenesis patients in the setting of MoMo twins still exhibit features of Potter's sequence, and, sometimes, extra renal syndromic abnormalities such as VATER. The cause of VATER syndrome is believed to be multifactorial, including environmental factorssuch as maternal diabetes and genetic factors as familial clustering is noted [13].

Our literature review (Table) showedincremental improvement in bilateral renal agenesis survival. In 2014 a patient survived beyond 9 months, this infant did not have VATER syndrome and their mother underwent serial amniotic fluid transfusions (Table 1). The infant described in our case is the first MoMo bilateral renal agenesis to survive, currently past 3 years and listed for transplant.Many cases of bilateral renal agenesis in MoMo twins demonstrate the following pattern: normal lung tissue, lack of Potter's sequence, and extra renal comorbidities.

As an infant with VATER association findings, our patient's history fits with these reports of MoMo twins. However, she has survived on peritoneal dialysis. Similar to other infants with end stage renal disease, our patient has demonstrated growth and developmental delay, anemia, hypertension, and peritonitis. During her first year of life, persistent hospitalization and the presence of a gastrostomy tube are two risk factors that could account for her increased rate of infection [14]. Her course also deviated from the typical end-stage pattern with respect to difficulty of control of her secondary hyperparathyroidism. Recommendations for infants and children on peritoneal dialysis suggest serum iPTH goals of 1.7 to 3 times the upper limits of normal, or 100-200 pg/ml [14]. Despite aggressive therapy with calcitriol and cinacalcet, her serum iPTH exceeded has been difficult to control and she has evidence of metabolic bone disease. Dietary factors, bilateral renal agenesis, or syndromic extra renal manifestations could contribute to the poor control of secondary hyperparathyroidism. Because the patient remained hospitalized for the duration of the first 12 months of life, adherence to her medication regimen is not in question.

Table

Author	Gestational age	Gender	Renal abnormalities	Other abnormalities	Mechanical ventilation	Urine production	Outcome
Mauer*et al.*, 1974	38 wks	Male	Bilateral renal agenesis, absent renal arteries	No urethral meatus, 2 vessel cord	None	Unspecified	Death at DOL 12
Koffler *et al.*, 1978	40 wks	Male	Multicystic left kidney, hypoplastic right kidney, ureters present	Anal atresia, absence of external genitalia, 2 vessel cord	None	Unspecified	Death at DOL 9
Cilentoet *al.*, 1994	36 wks	Male	Right renal agenesis, small dysplastic left kidney, bladder agenesis	TOF, esophageal atresia, 13th rib, peno-scrotal transposition with bilateral gonadal agenesis, redundant rectum, absent left thumb, 2 vessel cord	None	None	Death at DOL 2
McNamara *et al.*, 1995	33 wks	Male	Nonfunctioning dysplastic kidneys	Hemivertebrae, imperforate anus, intestinal duplication, GU anomalies	While septic	Unspecified	Hemodialysis attempted; death at DOL 7 (sepsis)
Klinger *et al.*, 1997	37 wks	Female	Bilateral renal agenesis, absent renal arteries, absent collecting system	Absent external genitalia, imperforate anus, left club foot, 2 vessel cord	None	Unspecified	Death at DOL 4
Perez-Brayfield*et al.*, 2004	35 wk	Male	Bilateral renal agenesis	None	None	None	Death at 2 months from peritoneal dialysis complications
Beinstock *et al.*, 2014	28 wk	Female	Bilateral renal agenesis, absent bladder	None	None	Unspecified	PD since DOL 3
Current Report	34 wk	Female	Bilateral renal agenesis, absent renal arteries, absent ureters	Hemivertebra, imperforate anus, ambiguous genitalia, 2 vessels chord	None	None	PD since DOL 13

Table 1: Demographics and characteristics reported in cases of renal agenesis/abnormalities

Conclusion

In summary, we report an infant with bilateral renal agenesis who has survived more than three years and has been listed for kidney transplant. Without modern day perinatal care, peritoneal dialysis, highly trained nursing, capable medical practitioners, and competent home care, our patient's prospects for survival would have been much lower. The complex care, including peritoneal dialysis, during the perinatal period has evolved so that even those born with bilateral renal agenesis can survive.

References

1. North American Pediatric Renal Trials and Collaborative Studies (NAPRTCS) 2011 Annual Transplant Report. EMMES Corporation, Rockville.

2. Potter EL. Bilateral absence of ureters and kidneys: A report of 50 cases. Obstet Gynecol.1965; 25:3-12.

3. Cilento BG, Jr., Benacerraf BR, Mandell J. Prenatal and postnatal findings in monochorionic, monoamniotic twins discordant for bilateral renal agenesis-dysgenesis (perinatal lethal renal disease). J Urol.1994; 151:1034-1035.

4. Bienstock J, Birsner M, Coleman, F, Hueppchen N. Successful In Utero Intervention for Bilateral Renal Agenesis. Obstet and Gynecol. 2014; 124 (S): 413-415.

5. Klinger G, Merlob P, Aloni D, Maayan A, Sirota L. Normal pulmonary function in a monoamniotic twin discordant for bilateral renal agenesis: report and review. Am J Med Genet. 1997; 73:76-79.

6. Koffler H, Aase JM, Papile LA, Coen RW. Persistent cloaca with absent penis and anal atresia in one of identical twins. J Pediatr. 1978; 93:821-823.

7. Mauer SM, Dobrin RS, Vernier RL. Unilateral and bilateral renal agenesis in monoamniotic twins. J Pediatr. 1974; 84:236-238.

8. McNamara MF, McCurdy CM, Reed KL, Philipps AF, Seeds JW. The relation between pulmonary hypoplasia and amniotic fluid volume: lessons learned from discordant urinary tract anomalies in monoamniotic twins. Obstet Gynecol. 1995; 85:867-869.

9. Perez-Brayfield MR, Kirsch AJ, Smith EA. Monoamniotic twin discordant for bilateral renal agenesis with normal pulmonary function. Urology. 2004; 64:589.

10. Fenton TR. A new growth chart for preterm babies: Babson and Benda's chart updated with recent data and a new format. BMC Pediatr. 2003; 3:13.

11. Pastores SM, Katz DP, Kvetan V . Splanchnic ischemia and gut mucosal injury in sepsis and the multiple organ dysfunction syndrome. Am J Gastroenterol. 1996; 91:1697-1710.

12. Folio MR, Fewell RR.Peabody Developmental Motor Scales. 2nd ed. Pro-ED Inc., Austin, TX. 2000.

13. Bartels E, Schulz AC, Mora NW, Pineda-Alvarez DE, Wijers CH, Marcelis CM, Stressig R, et al.VATER/VACTERL association: identification of seven new twin pairs, a systematic review of the literature, and a classical twin analysis. Clin Dysmorphol. 2012; 21:191-195.

14. Ramage IJ, Harvey E, Geary DF, Hebert D, Balfe JA, Balfe JW. Complications of gastrostomy feeding in children receiving peritoneal dialysis. Pediatr Nephrol.1999; 13:249-252.

15. Haffner D, Schaefer F. Searching the optimal PTH target range in children undergoing peritoneal dialysis: new insights from international cohort studies. Pediatr Nephrol 2013; 28:537-545.

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