Ped Urol Case Rep 2017; 4(6):387-390 DOI: 10.14534/PUCR.2017632143

Ped Urol Case Rep

PEDIATRIC UROLOGY CASE REPORTS

ISSN 2148-2969

http://www.pediatricurologycasereports.com

Bilateral renal agenesis, a severe anomaly in a premature infant with VACTERL association: A case report

Erol Basuguy[1], Sevinc Akdeniz[1], Sabahattin Ertugrul[2]

Dicle University, Medical School, [1]Pediatric Surgery and [2]Newborn Intensive Care Unit, Diyarbakir, Turkey

ABSTRACT

We report on a preterm male (birth weight 1,100 g) with bilateral renal agenesis, a lethal malformation. Additionally, the child suffered from an atrial septal defect, ventricular septal defect, right aortic arch anomaly, a high type of anal atresia, vertebral anomalies, limbs defects (VACTERL association). The infant during first day of life was treated with an emergency sigmoid ostomy and peritoneal dialysis because of increasing abdominal dilatation and high urea and creatinine levels in blood. Important congenital anomalies associated with VACTERL association and prematurity are very serious causes of mortality in the early period.

Key Words: VACTERL association; renal anomalies; bilateral renal agenesis; preterm

Corresponding Author: *Dr. Erol Basuguy*
Dicle University, Medical School, Pediatric Surgery, Diyarbakir, Turkey
E-mail: erbas.80@hotmail.com
Accepted for publication: 15 September 2017

Introduction

The vertebral defect, anal atresia, cardiac defect, tracheoesophageal fistula/esophageal atresia, renal defect, and limb defect (VACTERL) association are rare conditions. Even with optimal surgical corrections of malformations, patients affected by the VACTERL association can face medical challenges throughout life such as back pain (scoliosis), fecal incontinence (anal atresia,), and functional impairment (limb anomalies) [1,2]. The medical challenges are highly dependent on type and severity of the specific malformation [1]. Urinary tract abnormalities are seen in approximately 60–90% of patients with VACTERL association [3]. There can be a wide range of severity and type of urinary tract anomalies, which can include unilateral renal agenesis (or bilateral in severe cases), horsehoe kidney, and cystic and/or dysplastic kidneys, vesicoureteral reflux (VUR) and neurogenic bladder [2,3,4]. These problems can cause end stage renal failure (ESRF) and may require kidney transplant [3]. Bilateral renal agenesis is a lethal malformation in human and in the majority of cases, no specific etiology can be established, although teratogenic, syndromal and single gene causes can be assigned to some cases [5]. We report on a preterm male (birth weight 1,100 g) with bilateral renal agenesis, a lethal malformation.

Additionally, the child suffered from an atrial septal defect, ventricular septal defect, right aortic arch anomaly, a high type of anal atresia, vertebral anomalies, limbs defects (VACTERL association).

Case report

A preterm male born at a gestational age of 28 weeks with a birth weight of 1100 g presented with anal atresia and limb anomalies [**Fig. 1**].

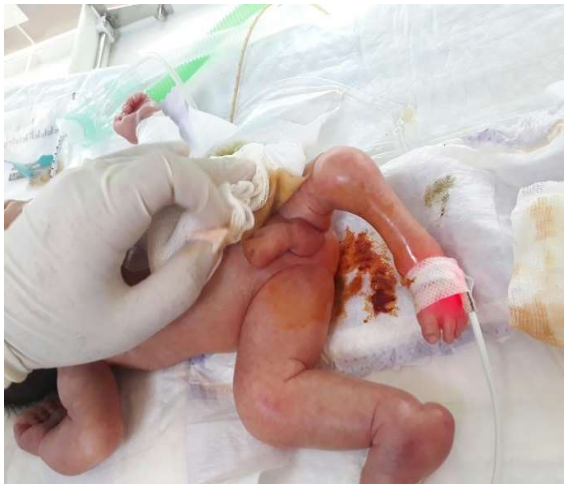

Fig. 1. VACTERL infant.

His family history was without known inborn abnormalities and consanguinity. The pregnancy was uneventful. Ultrasonography showed bilateral renal agenesis. Contrast material delivered by urethral pathway was observed to pass directly to the sigmoid colon [**Fig. 2**].

The infant was admitted to newborn intensive care unit for treatment. During first day of life, he was treated with an emergency sigmoid ostomy and peritoneal dialysis because of increasing abdominal dilatation and high blood urea (68 mg/dl) and creatinine (3.7 mg/dL) levels.

Additionally, the child suffered from an atrial septal defect, ventricular septal defect, and right aortic arch anomaly, a high type of anal atresia, vertebral anomalies (hemivertebrae, dysplastic vertebrae) and limbs defects.

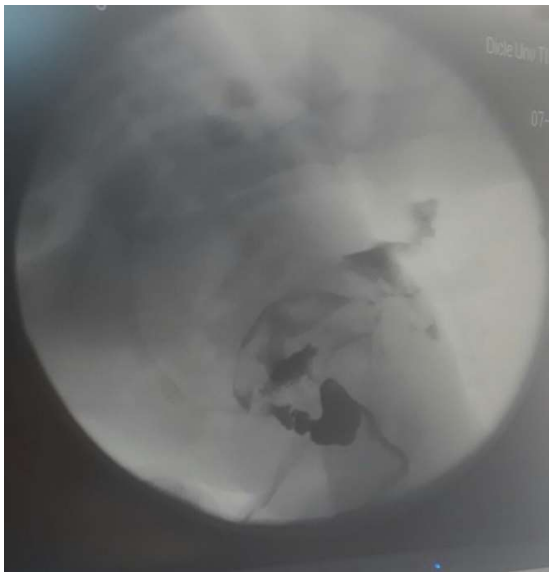

Fig. 2. Retrograde urethrogram: contrast material passes directly to the sigmoid colon.

Color Doppler echocardiography demonstrated cardiac anomalies such as patent ductus arteirosus, atrial septal defect, ventricular septal defect, right aortic arch anomaly and atrial septal aneurysm.

Computed tomography (CT) scanning revealed bilateral renal agenesis, hemivertebrae, vertebral fusions and dysplastic vertebrae.

He fulfilled the criteria for VACTERL association with vertebral anomalies, anal atresia, cardiac anomalies, bilateral renal agenesis and lower limb anomalies.

After a treatment period of 2 weeks, the baby died of a sepsis and multiple organ failure.

Discussion

Like other congenital malformations demonstrated in VACTERL association, there are a wide range of severity and type of renal anomalies, which can include unilateral renal agenesis (or bilateral in severe cases), horsehoe

kidney, and cystic and/or dysplastic kidneys, VUR and neurogenic bladder, sometimes accompanied by ureteral and GU anomalies [4,5,6]. Renal anomalies are reported in approximately 50-80% of patients with VACTERL association [6,7,9,10]. Cunningham et al [11] suggested that the most frequent renal manifestation in their VACTERL cohort was VUR in conjunction with a structural renal anomaly in addition to a structural defect (present in 27%), followed by unilateral renal agenesis (24%), and then dysplastic/multicystic kidneys or duplicated collected system (18% for each).

Although genitourinary anomalies are not considered one of the core component features in patients with VACTERL association, ascertainment of such anomalies can be critical, as they may lead to significant morbidity and mortality [4,12]. These problems can cause end stage renal failure and may require kidney transplant. Additionally, bilateral renal agenesis is a lethal malformation in humans [3,5].

As we have here, important congenital anomalies associated with VACTERL association and prematurity are very serious causes of mortality in the early period. In other cases, developments in surgical techniques, neonatal specialization and post-surgical facilities revealed a much better prognosis in the diagnosis and treatment of VACTERL association. However, even with ideal surgical corrections of this malformations, patients can face major medical challenges throughout life [2].

Acknowledgements

The author(s) declare that they have no competing interest and financial support.

References

[1]Bjørsum-Meyer T, Herlin M, Qvist N, Petersen MB. Vertebral defect, anal atresia, cardiac defect, tracheoesophageal fistula/esophageal atresia, renal defect, and limb defect association with Mayer-Rokitansky-Küster-Hauser syndrome in co-occurrence: two case reports and a review of the literature. J Med Case Rep. 2016;10(1):374.

[2]Solomon BD. VACTERL/VATER Association. Orphanet J Rare Dis. 2011;16;6:56.

[3]Telkes G, Reusz G, Szabó AJ, Langer RM. A single-center experience with kidney transplantation in the verteberal, anal, cardiac, tracheoesophageal, renal, and limb birth detects (VACTERL) association. Transplant Proc. 2011;43(4):1250-1.

[4]Solomon BD, Raam MS, Pineda-Alvarez DE. Analysis of genitourinary anomalies in patients with VACTERL (Vertebral anomalies, Anal atresia, Cardiac malformations, Tracheo-Esophageal fistula, Renal anomalies, Limb abnormalities) association. Congenit Anom (Kyoto) 2011; 51(2):87-91.

[5]Harewood L, Liu M, Keeling J, Howatson A, Whiteford M, Branney P, Evans M, Fantes J, Fitzpatrick DR. Bilateral renal agenesis/hypoplasia/dysplasia (BRAHD): postmortem analysis of 45 cases with breakpoint mapping of two de novo translocations. PLoS One. 2010;5(8):e12375.

[6]Rittler M, Paz JE, Castilla EE. VATERL: an epidemiologic analysis of risk factors. Am J Med Genet. 1997;73(2):162-9.

[7]Weaver DD, Mapstone CL, Yu PL. The VATER association. Analysis of 46 patients. Am J Dis Child. 1986 ;140(3):225-9.

[8] Solomon BD, Pineda-Alvarez DE, Raam MS, et al. Analysis of component findings in 79 patients diagnosed with VACTERL association. Am J Med Genet A. 2010;152A(9):2236-44.

[9] Botto LD, Khoury MJ, Mastroiacovo P, et al. The spectrum of congenital anomalies of the VATER association: an international study. Am J Med Genet. 1997;71(1):8-15.

[10] Mapstone CL, Weaver DD, Yu PL. Analysis of growth in the VATER association. Am J Dis Child. 1986;140(4):386-90.

[11] Cunningham BK, Khromykh A, Martinez AF, Carney T, Hadley DW, Solomon BD. Analysis of renal anomalies in VACTERL association. Birth Defects Res A Clin Mol Teratol. 2014;100(10):801-5.

[12] Rich MA, Brock WA, Peña A. Spectrum of genitourinary malformations in patients with imperforate anus. Pediatr Surg Int. 1988; 3(2-3):110–113

Access this article online

http://pediatricurologycasereports.com

Quick Response Code