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

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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

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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), horseshoe 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.

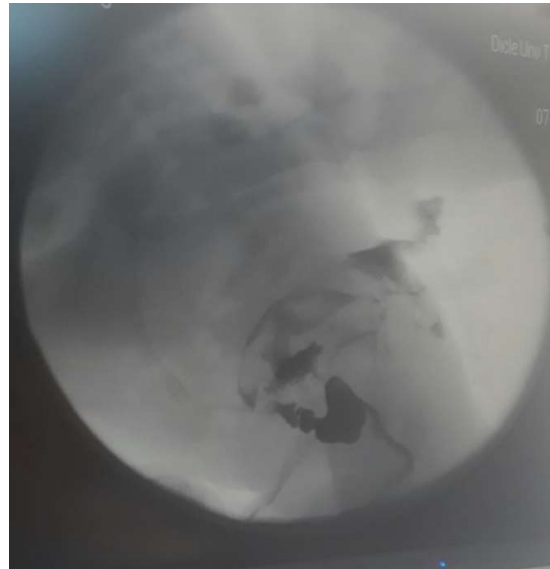


Fig. 2. Retrograde urethrogram: contrast material passes directly to the sigmoid colon.

Color Doppler echocardiography demonstrated cardiac anomalies such as patent ductus arteriosus, 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), horseshoe

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].

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