VACTERL ASSOCIATION WITH POTTER SEQUENCE

VACTERL BİRLİKTELİĞİ VE POTTER SEKANSI

Abdullah Barış AKCAN[*], Seyhan ERİŞİR OYGUCU[*], Ercan MİHCİ[*], Güzide Ayşe GÖKHAN[**], Nihal OYGÜR *

ABSTRACT

Upper urinary system involvement is common in VACTERL association. However cases, reported in literature, have nonserious renal abnormalities and generally have a good outcome. We report a neonate who died shortly after birth, having both VACTERL association and Potter sequence due to cystic renal dysplasia. This unusual combination is extremely rare. We recommend investigating Potter sequence in the spectrum of congenital anomalies with the VACTERL association.

***Key words*:** VACTERL association, potter sequence, neonate

ÖZET

VACTERL birlikteliğinde üst üriner sistem tutulumu sık görülür. Buna karşın literatürde ki vakalarda ciddi olmayan ve genelde prognozu iyi olan böbrek tutulumu görülmektedir. Bu yazıda, doğumdan hemen sonra ölen, kistik renal displazi ve Potter sekansı ile VACTERL birlikteliği gösteren bir yenidoğan vakası sunulmuştur. Bu birliktelik oldukça nadir görülmektedir. VACTERL birlikteliği olan konjenital anomalilerde Potter sekansının da araştırılmasını önermekteyiz.

Anahtar kelimeler: VACTERL birlikteliği, potter sekansı, yenidoğan

INTRODUCTION

VATER is a mnemonically useful acronym for vertebral defects (V), anal atresia (A), tracheoesophageal fistula with eusophageal atresia (TE), renal, and radial dysplasia (R). This association was first reported in 1973 and later expanded to the VACTERL association, adding cardiac (C) and limb malformations (L) to the previous features. Potter syndrome, which causes a typical physical appearance, is the result of oligohydramnios secondary to renal diseases such as bilateral renal agenesis. Other common causes of neonatal renal failure associated with the Potter phenotype include cystic renal dysplasia and obstructive uropathy (1,7).

Upper urinary system involvement is common in VACTERL association. Renal anomalies are found in approximately 60-90% of VACTERL patients. Uehling et al. reported genitourinary involvement in 21 of 23 children with VATER association, including renal agenesis, severe reflux, crossed fused ectopia, and ureteropelvic junction obstruction (8,9). However in large series, reported in literature, almost all cases have nonserious renal abnormalities such as multiple renal cysts, ectopic and/or supernumeray kidney, horseshoe kidney. Potter sequence is not expected in such kind of renal abnormalities and these infants generally have a good outcome (2,3,5,6). Now we report a neonate having spectrums of VACTERL association and Potter sequence due to cystic renal dysplasia which is an unusual abnormality seen in this association.

CASE

A female neonate was born vaginally at 36 weeks of gestation to a 20-year-old secundigravida mother and a 27 year old father. Their first child has died two months old with an unknown etiology however, her phenotype was normal. Parents were first degree consanguineous. The mother did not have a history of antenatal teratogen exposure, infection, fever, rash or any drug intake and did not smoke or drink alcohol during her pregnancy. She had regular antenatal check ups. Her ultrasonographic examination, performed at 31st weeks of gestation, revealed oligohydramniosis and renal development anomalies. The newborn was delivered with caeserian section as her fetal heart rates showed bradycardia. She was born with the low Apgar scores of 4/7 at 1st and 5th minutes. She was resuscitated with bag and mask in the delivery room and transferred to NICU after the intubation.

On her physical examination, birth weight was 2350 g (50-90 th percentile), length 39 cm (10th percentile) and head circumference was 34 cm (50-90th percentile). She had a characteristic Potter face with compressed flat nose, bilateral webbing, separated eyes, prominent epicanthal folds, micrognathia and low set ears. She had also short neck and clinodactyly. Upper limbs were malformated and lower limbs showed pes equinovarus deformity of the left foot and rocker bottom of the right foot (Figure 1) . There were scoliosis of lumbar vertebrae and oligodactyly at her right foot .

Her cardiac examination revealed a pansystolic murmur,

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* Akdeniz Üniversitesi Tıp Fakültesi, Çocuk Sağlığı ve Hastalıkları Anabilim Dalı, Antalya
(İletişim kurulacak yazar: barisakc@hotmail.com)

** Akdeniz Üniversitesi Tıp Fakültesi, Patoloji Anabilim Dalı, Antalya

heard at the left upper sternal border. Her abdomen was distended and external genitalia was normal. Imperforated anus was also detected on her genitourinary examination.

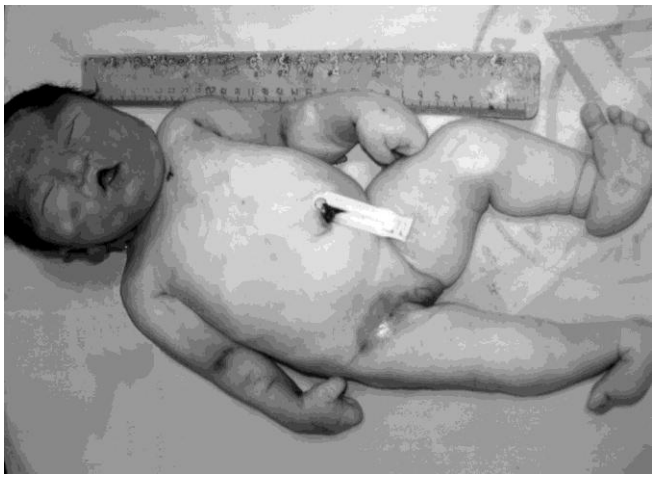

Figure 1. The general appearance of the infant

Chest x-ray showed coiling of the nasogastric tube with presence of gas in the stomach, suggesting a diagnosis of tracheoesophageal fistula with eusophageal atresia. X-ray of vertebrae showed, anomalies of lumbar vertebrae and a rudimentary sacrum. The urine output of the patient was only 3 ml/day on the first day. She gained much weight despite volume restriction. Peritoneal dialysis was planned but, her vital signs, blood gas values deteriorated and died on the second day of life.

Postmortem examination revealed normally developed central nervous system. There was cardiomegaly, especially right atrium was larger than normal. The right lung had two lobes. There was tracheoesophageal fistula with esophageal atresia. Both kidneys were smaller than original size and showed cystic renal dysplasia (Figure 2). Both ureters were normal. Descending colon ended blindly. The gonads were normal ovaria, normal fallopian tubes and a bicornuate uterus. Karyotype analysis was normal (46, XX).

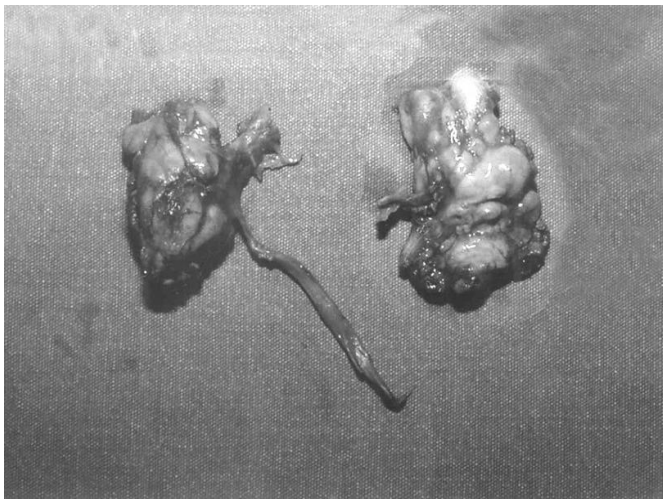

Figure 2. Both kidneys were smaller than original size and showed cystic renal dysplasia

DISCUSSION

The acronym VACTERL refers to the symbols of anomalies of different organ systems due to developmental missteps in intrauterine life. Every year the spectrum and components of associated defects in every organ system is going to increase in number due to the new cases published in literature. In one of the recent literature, published by Keckler, a review of 20-year experience of patients has shown that the anomalies of the urinary system were the third most common associated birth defect occuring in 17% of patients with VACTERL association and vesico-ureteral reflux was the most common renal anomaly (42% of renal anomalies) (2). In another documentation done by Kolon *et al* vesico-ureteral reflux was also the most common renal anomaly (93.2% of renal anomalies) in VACTERL patients with renal involvement (3).

Although the upper urinary system is the third most common organ, involved in VACTERL association, most of the cases, reported in literature had nonserious renal abnormalities such as multiple renal cysts, ectopic and/or supernumeray kidney, horseshoe kidney and these infants generally had a good outcome (2,3).

Our patient had all the characteristic features of VACTERL association. Besides she had also the clinical findings of Potter sequence such as compressed flat face and limb deformities. To our knowledge the association of VACTERL and Potter sequence has been reported only once in a child and ours is the second patient in the literature. The other case, presented by Lukusa *et al*, was a combination of Potter sequence, Prune Belly anomaly and VACTERL association (4).

In general at least one blastogenetic defect can be found in approximately 10% of liveborn malformed infants and these defects of blastogenesis are highly lethal. The type of defects that comprise the VACTERL association suggest that most of them occur during early blastogenesis such as renal agenesis. Sometimes any of the VACTERL anomalies may appear as being of nonblastogenetic origin such as renal dysplasia (2,5). In the series of Martínez-Fríaz the proportion of VACTERL anomalies observed in the group of patients with nonblastogenetic defects was higher than in those with blastogenetic defects (5). According to his hypothesis, since blastogenetic defects are highly lethal , they may produce miscarriages more than nonblastogenetic defects thus making them less frequent. The clinical findings of our patient seems to be related with developmental malformations of both nonblastogenetic and blastogenetic origin.

The features of oligohydramniosus as Potter face and limb deformities suggest that the cause of death should be the result of respiratory insufficiency due to pulmonary developmental defect. However the pulmonary system did not have the histologic findings of pulmonar hypoplasia at postmortem examination.

In conclusion we can not say whether the combination of VACTERL association and Potter seguence is coincidental or a constant finding. There fore we recommend investigating Potter sequence in the spectrum of congenital anomalies of the VACTERL association.

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