Bilateral multicystic renal dysplasia with potter sequence

A case with penile agenesis

Ahmet Dursun, *MD, PhD,* ***Bahri Ermis,*** *MD, PhD,* ***Varim Numanoglu,*** *MD, PhD,* ***Burak Bahadir,*** *MD, PhD,* ***Ilker Seckiner,*** *MD, PhD.*

ABSTRACT

Hereditary renal adysplasia (HRA) is a rare autosomal dominant condition. Patients have several other anomalies including Potter facies, thoracic, cardiac, and extremity deformities. The case present dysmorphic facial features such as hypertelorism, prominent epicanthic folds, a flat and broad nose, choanal stenosis, low-set ears, and a receding chin. He had femoral bowing, hypoplastic right tibia and agenesis of the right foot. He had rich and thick skin. He had also a dysplastic empty scrotum, penile agenesis, and anal atresia. The autopsy revealed pulmonary hypoplasia, ventricular septal defect, bilateral multicystic renal dysplasia, agenesis of both ureter and bladder, intraabdominal testicles, and a single umbilical artery. The penile agenesis was first reported, and including the consanguinity in the parents might further delineate the bilateral multicystic HRA. Vater/caudal regression anomalies, Mullerian duct/aplasia, unilateral renal agenesis, and cervicothoracic somite anomalies association, and Coloboma, heart anomaly, choanal atresia, retardation, genital and ear anomalies syndrome has been considered in differential diagnosis.

Saudi Med J 2006; Vol. 27 (11): 1745-1747

Hereditary renal adysplasia (HRA) is a rare and probable autosomal dominant condition first described in sibs.[1] In 1973, it was hypothesized that a spectrum of renal malformation ranging from bilateral renal adysplasia (BRA) to unilateral renal adysplasia had the same genetic origin and proposed the term "Hereditary Renal Adysplasia".[2] Patients have several other anomalies including potter facies, thoracic, cardiac, and extremity deformities. We present a case born to consanguineous parents with new features of bilateral multicystic renal dysplasia.

Case Report. A baby boy was born as the first child of healthy consanguineous parents (first cousin). This stillborn boy was delivered at 35 weeks of gestation due to the oligohydramnios. The mother had no prior antenatal care. Birth weight was 1422 g, length 43.5 cm, head circumference (OFC) 31 cm. He exhibited dysmorphic facial features (Potter facies), such as hypertelorism, prominent epicanthic folds, a flat and broad nose, choanal stenosis, low-set ears, and a receding chin. He had also femoral bowing, hypoplastic right tibia, and agenesis of the right foot (**Figure 1**). The other

From the Department of Medical Genetics (Dursun), Division of Neonatology (Ermis), Department of Pediatric Surgery (Numanoglu), Department of Pathology (Bahadir), and the Department of Urology (Seckiner), ZKU Medical Faculty, Zonguldak, *Turkey.*

Received 4th December 2005. Accepted for publication in final form 3rd June 2006.

Address correspondence and reprint request to: Dr. Ahmet Dursun, Department of Medical Genetics, ZKU Medical Faculty, Kozlu, 67600 Zonguldak, *Turkey*. Tel. +90 (372) 2610243 Ext.4332. Fax. +90 (372) 2610251. E-mail: dursuna@karaelmas.edu.tr

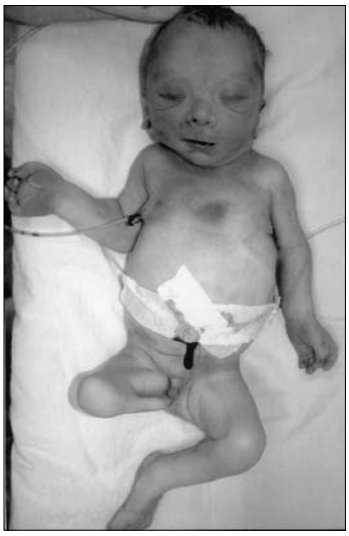

Figure 1 - The baby had potter facies and extreme deformity.

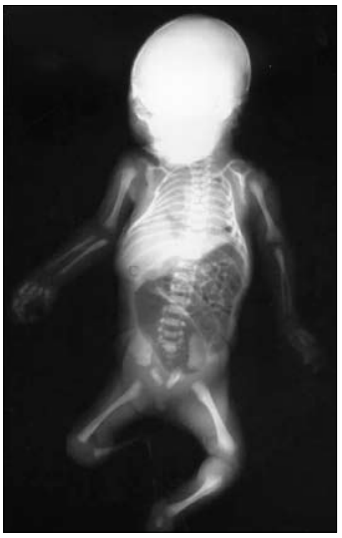

Figure 3 - Radiological findings of limb defects and pulmonary hypoplasia.

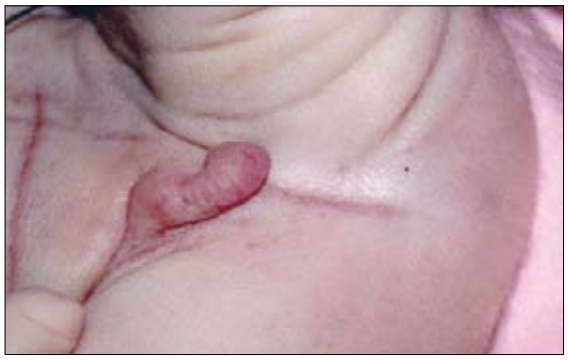

Figure 2 - Penile agenesis and anal atresia.

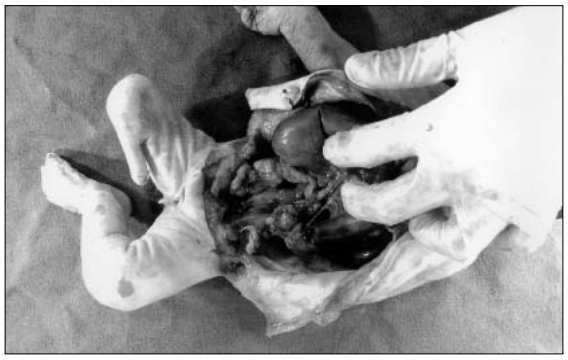

Figure 4 - Renal adysplasia and agenesis of both ureter and bladder.

extremities were normal. He had rich and thick skin. The examination of the genitalia revealed that he had a dysplastic empty scrotum, penile agenesis, and anal atresia (**Figure 2**). He also had a respiratory problem causing cyanosis as expected, and the thorax was hypoplastic (circumference were 28 cm). Radiological examination of the body confirmed the limb defect and pulmonary hypoplasia (**Figure 3**). The autopsy confirmed pulmonary hypoplasia (right lung 4 x 3.5 x 1.5 cm, left lung 4.3 x 1.5 x 1.5 cm diameters). He had also a ventricular septal defect. The examination of the abdomen revealed a multicystic BRA. The measurement of the right kidney was 3.5 x 1.8 x 0.5 cm, and the left kidney was 2.8 x 1.8 x 0.4 cm (**Figure 4**). The left kidney was situated lower than its normal position. Agenesis of both ureter and bladder were noted, and testicles were seen in the abdominal cavity. Dilated sigmoid colon was filled with meconium and ended with an imperforate anus. An umbilical cord had a single umbilical artery. Malrotation of the abdominal organs was not noted. The karyotype analysis has not been completed due to technical problems.

Discussion. Sporadic is the most cases of renal agenesis and dysplasia.[3] However, in 1973, Buchta et al[2] first suggested the term HRA and reported families, in which both unilateral and bilateral renal agenesis and severe aplastic dysplasia were segregated. Multicystic dysplasia, which differs from aplastic dysplasia only in a degree of cyst formation, was regarded until recently as a sporadic entity. In 1987, Squiers et al[4] reported the occurrence of unilateral multicystic dysplasia in an infant, whose mother and maternal aunt had unilateral renal agenesis, suggesting that non-syndromal multicystic

renal dysplasia can occur as a part of the spectrum of HRA. Hereditary renal adysplasia was hypothesized, resulting from failure of the ureteric bud to make contact with the metanephric blastema prior to the end of the fourth week of development.[5,6] In addition, it has been suggested that developmental defects in the mesonephric and paramesonephric ducts may have a common genetic basis.[7] Same authors also proposed the term hereditary urogenital adysplasia for the association of anomalies of the urinary tract and of the Mullerian duct. Opitz, reviewed the relation between HRA and mullerian anomalies, and concluded that the 2 defects may result from the variable expression of a single autosomal dominant gene.[8]

This case provides most symptoms of bilateral multicystic HRA. The penile agenesis was first reported, and including the consanguinity in the parents might further delineate the bilateral multicystic HRA. Vater/caudal regression anomalies, Mullerian duct/aplasia, unilateral renal agenesis, and cervicothoracic somite anomalies (MURCS) association, and Coloboma, heart anomaly, choanal atresia, retardation, genital and ear anomalies (CHARGE) syndrome has been considered in differential diagnosis. Although, Vater association includes the anal atresia, choanal atresia, renal dysplasia and ventricular septal defect, our case possessed the Potter sequences and had no skeletal anomalies except the limb defect mentioned above. The MURCS association has also renal agenesis, and share some of the facial features of the Potter facies, nevertheless, our case had more distinctive feature suggesting HRA. The CHARGE syndrome also features choanal atresia, cardiac, and genitourinary system anomalies, which are similar to our case, however, limb defects, Potter facies, and bilateral multicystic renal dysplasia had distinguished from our suggested HRA case from that of CHARGE. Due to low sociological level, we could not persuade the parents for further examination to test for renal dysmorphology. Genetic counseling, has been given for the next pregnancy informing that their next baby might be at risk for the same condition due to consanguinity and detailed ultrasound, examination, and antenatal care will be needed.

References

1. Madisson, H. Ueber das Fehlen beider Nieren (Aplasia renum bilateralis). ***Zentralbl Allg Pathol*** 1934, 60: 1-8
2. Buchta RM, Viseskul C, Gilbert EF, Sarto GE, Opitz JM. Familial bilateral renal agenesis and hereditary renal adysplasia. ***Z Kinderheilkd*** 1973; 115: 111-129.
3. Duke V, Quinton R, Gordon I, Bouloux PM, Woolf AS. Proteinuria, hypertension and chronic renal failure in X-linked Kallmann's syndrome, a defined genetic cause of solitary functioning kidney. ***Nephrol Dial Transplant*** 1998; 13: 1998-2003.
4. Squiers EC, Morden RS, Bernstein J. Renal multicystic dysplasia: an occasional manifestation of the hereditary renal adysplasia syndrome. ***Am J Med Genet Suppl*** 1987; 3: 279-284.
5. Potter EL. Normal And Abnormal Development Of The Kidney. Chicago: Year Book Medical Publishers; 1972. p. 98.
6. Wood SG, Skandalakis JE. Embroyology For Surgeons: The Embryological Basis For The Treatment Of Congenital Defects. Philadelphia: WB Saunders; 1972. p. 459.
7. Schimke RN, King CR. Hereditary urogenital adysplasia. ***Clin Genet*** 1980;18: 417-420.
8. Opitz JM. Vaginal atresia (von Mayer-Rokitansky-Kuster or MRK anomaly) in hereditary renal adysplasia (HRA). ***Am J Med Genet*** 1987; 26: 873-876.