Bilateral multicystic renal dysplasia with potter sequence

A case with penile agenesis

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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.

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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
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. However, in 1973, Buchta et al 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 reported the occurrence of unilateral multicystic dysplasia in an infant, whose mother and maternal aunt had unilateral renal agenesis, suggesting that non-syndromal multicystic
Bilateral renal dysplasia ... Dursun et al

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. In addition, it has been suggested that developmental defects in the mesonephric and paramesonephric ducts may have a common genetic basis. 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.

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