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Cloacal dysgenesis sequence with bilateral renal agenesis, bladder agenesis, pulmonary hypoplasia and left choanal atresia: A case report

Esra Ozcakir, Sefer Tolga Okay, Ipek Guney Varal, Mete Kaya
Department of Pediatric Surgery, University of Health Sciences, Bursa Yuksek Ihtisas Training and Research Hospital, Bursa, Turkey

ABSTRACT

A case of posterior urethral valve is described with multiple urethroperineal fistulas. This association is extremely rare. The crucial point in the diagnosis is to distinguish these fistulas from the urethral duplications that actually guide the treatment. The presence of these fistulas in patients with posterior urethral valve may have a beneficial effect on renal function as it reduces the pressure on the bladder. On the other hand, incomplete valve ablation may contribute to the recurrence of the fistula.
Key Words: Cloacal dysgenesis sequence, bilateral renal agenesis, bladder agenesis, pulmonary hypoplasia, left choanal atresia.

Dr. Esra Ozcakir,
Department of Pediatric Surgery, University of Health Sciences, Bursa Yuksek Ihtisas Training and Research Hospital, Bursa, Turkey
E mail: dresramermer@hotmail.com
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Introduction

Cloacal dysgenesis sequence (CDS) is a rare congenital anomaly seen in 1: 50000 to 250000 newborns [1,2]. It is characterized by a phallus-like structure, smooth perineum, and absence of genitourinary and anal orifices as primary malformations [1-3]. The coexistence of bilateral renal agenesis with CDS is still very rare, and the resulting severe oligohydramnios also causes pulmonary hypoplasia [4]. In addition, megacystis, hydronephrosis, cystic kidneys, and absent uterus are considered as secondary malformations associated with CDS [3]. Very few cases of CDS with pulmonary hypoplasia and bilateral renal agenesis have been reported in the English language literature. A case of pulmonary hypoplasia, bilateral renal agenesis and choanal atresia associated with CDS is reported in the current study.

Case report

The baby of a 30-year-old multiparous mother (gravida 3, para 2) who was born by cesarean section for 32 weeks was brought with a smooth perineal anomaly showing absence of anal, genital and urinary orifices. The patient's history revealed that the fetus had bilateral renal agenesis, pericardial effusion and bilateral hypoplastic lungs with anhydramnios on prenatal ultrasonography at 17 weeks of

gestation. Visualization of external genitalia was impossible due to severe oligohydramnios. According to these results, the parents were informed about the poor prognosis and the possibility of pulmonary hypoplasia. It was understood that termination was offered to the family, but the family wanted the pregnancy to continue. After delivery, the baby was cyanotic with no respiratory effort and decreased heart rate. She was intubated and responded well with improved color and heart rate. The newborn was admitted to the intensive care unit and the treatment started. Right lung sounds were found to be severely decreased, and right thorax tube drainage was started. Peritoneal dialysis treatment was started (Fig. 1). Eventually, the baby's condition deteriorated, and the patient died the same day. The parents were recommended autopsy but did not agree to it.

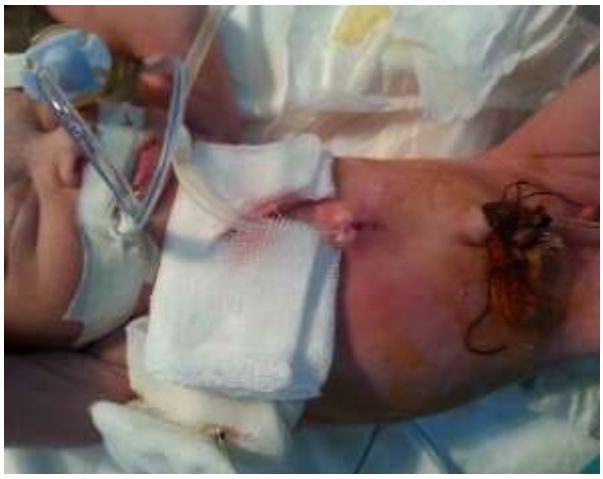

Fig. 1. A photograph showing the newborn with CDS who placed a chest tube and a peritoneal dialysis catheter.

On examination, there was no formation of female or male genitalia and the anal orifice was also absent. Only a small nubbin of tissue was present in the perineum (Fig. 2). There was the left choanal atresia. Abdominal USG showed absence of both kidneys and a dilated bowel loop on the left side of the abdomen. In addition, the bladder was not observed.

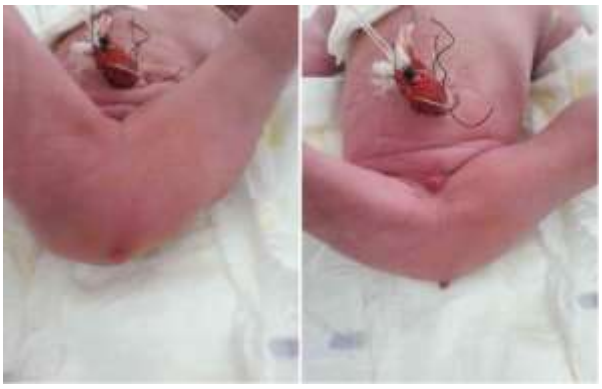

Fig. 2. In the photographs, no opening is seen on a flat perineal surface and urogenital and sacrococcygeal nubbin formation is observed.

Discussion

Cloacal dysgenesis sequence (CDS) is a collection of anomalies resulting from the failure of the division of primitive cloaca, which can cause fetal obstructive uropathy (FOU), a complex malformation of complicated embryological origin [1-4]. CDS is almost entirely seen in females but has also been described in male patients [3,5]. The cloacal membrane has an important role in the development of external genital organs, urethral orifice and some adjacent structures. Failure to develop a cloacal membrane result in the absence of genital folds, pars phallic of the urogenital sinus, anal tubercles, and the connection between the cloacal derivatives and the outer part. The typical manifestations of CDS are the absence of anal, urethral and vaginal holes and abnormal labioscrotal structures along with some secondary deformities [2,3,6,7]. The severity of fetal outcomes is generally correlated with the duration of exposure to urinary outflow obstruction as well as pulmonary immaturity [3].

In the Cloacal dysgenesis sequence, urinary outflow obstruction causes secondary urinary tract problems such as hydronephrosis, hydroureter or dysplastic kidneys. Vaginal atresia, rectovaginal fistula, dysplastic ovaries and uterine anomalies are associated with genital anomalies seen in CDS [3,8,9]. High pressure in the urinary tract and severe oligohydramnios in CDS leads to severe renal dysfunction and pulmonary immaturity and ultimately, a fatal picture [9,10].

Prenatal diagnosis of CDS by ultrasound is possible. Megacystis, oligohydramnios, perineal mass, hydronephrotic or dysplastic renal anomalies seen in CDS can be detected as prenatal ultrasound findings [3,11,12]. However, the bladder may not be visualized due to renal agenesis, dysplastic kidneys, and agenesis of the bladder or perforation of the bladder [3]. In addition, intracolonic calcifications due to calcified meconium may be seen [3,11]. The communication between urine and gastrointestinal tract causes calcified meconium formation [13]. In rare cases, as in the present case, there will be no perineal patency diagnosed with cloacal dysgenesis. This is the most severe type of cloacal malformation spectrum, and the presence of complete anhydramnios, prominent urinary acids and hypoplastic fetal lungs without fluid is the leading diagnosis in this case [14]. It is important to differentiate CDS from other treatable conditions such as the fetal obstructive uropathies and persistent cloaca [3].

The prognosis of newborns diagnosed with CDS is extremely poor. The majority of known cases were stillborn, or the pregnancy was terminated. Only a few cases were born alive and died in the neonatal period [3,15]. As mentioned above, severe renal anomaly and pulmonary hypoplasia are the two most important factors limiting the survival of babies born with CDS. Five cases of CDS over the age of 1 have been reported with long-term survival [10,16].

In conclusion, the CDS is a very rare series of congenital malformations characterized by a phallus-like structure, smooth perineum and absence of urethral, vaginal and anal openings as primary malformations. This malformation is a defect in the formation of cloaca during the first 50 days of pregnancy. It is associated with abnormalities of the pulmonary and genitourinary system, which significantly affect prognosis. Serial sonographic examinations are very important in the prenatal diagnosis of CDS.

Compliance with ethical statements

Conflicts of Interest: None.

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Consent: All photos were taken with parental consent.

ORCID iD of the author (s)

Esra Ozcakir /0000-0002-0773-7430
Sefer Tolga Okay /0000-0002-2716-6006
Mete Kaya /0000-0002-8877-5737
Ipek Guney Varal /0000-0002-3298-066X

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