The cloaca is a single canal from which the urinary, genital, and intestinal tracts arise around gestational weeks 5-6. Developmental defects in septum formation lead to persistent cloaca, a condition associated with a wide spectrum of complex anatomical abnormalities (Hendren 1992). Cloacal abnormalities are relatively rare with an overall incidence of 1 in 50,000 neonates (Pena 1989), and can be detected by prenatal ultrasonography as abnormalities in fetal ascites, cystic
hydronephrosis at 30 weeks gestation, which worsened during the course of pregnancy. The pelvic cyst size had increased to a volume of 9.8 × 8.2 × 6.2 cm by 36 weeks gestation.

Because the right hydronephrosis had worsened substantially, the baby was delivered at 36 weeks of gestation by caesarean section. The infant weighed 2,414 g and measured 43 cm in length at birth. Her Apgar scores were 6 at 1 min and 9 at 5 min post-delivery. The infant was intubated because of dyspnea caused by abdominal distension. Computed tomography confirmed esophageal atresia with tracheoesophageal fistula, right hydronephrosis (grade 2-3), a non-functioning left kidney, and persistent cloaca. Also, postnatal ultrasonography confirmed the presence of a VSD.

The tracheoesophageal fistula was repaired and a defunctioning transverse colostomy was performed 12 hrs after birth. Approximately 140 ml of cloudy urine was drained by urethral catheter, leading to disappearance of the pelvic cyst. Urethrocystography revealed a single perineal/anal opening that drained into a common cloaca, as well as the lack an anus suggesting hydrocolpos. Intestinal barium examination showed a rectal fistula ending in the urogenital sinus with no vesicoureteric reflux. Three weeks after delivery, the baby was discharged.

DISCUSSION

During embryogenesis, urorectal septal formation occurs around the same time that VATER association develops. Axial mesodermal dysplasia spectrum (AMDS) is thought to arise as a consequence of aberrant mesodermal migration during early development, resulting in bronchial, pulmonary, cardiovascular, gastrointestinal, renal, urogenital and skeletal abnormalities (Russell et al. 1981). Both cloacal malformation and VATER association are likely to be associated with AMDS, as they are among the continuous spectrum of AMDS anomalies (Henmi et al. 2000). Consistent with this hypothesis, Liu and Hutson (2000) showed in an adriamycin-induced rat model of cloacal and urogenital anomalies, similarities to VATER association.
Fig. 1. Prenatal ultrasound image of the fetus at 26 weeks of gestation. Gastrointestinal and urogenital abnormalities were found. a) imperforate anus (arrow) behind the vagina (V), b) ascites, c) pelvic cystic mass (C) with debris behind the bladder (B), d) polycystic left kidney. Also shown are explanatory drawings.
our patient could be diagnosed as a combination of an intermediate form of the cloacal-VATER spectrum, although details of the internal genitalia were unclear. Prenatal diagnosis of this spectrum by ultrasonography is important and could reveal cardiac anomalies, imperforate anus, as well as vertebral anomalies. Esophageal atresia with tracheoesophageal fistula is difficult to diagnose prenatally. However, magnetic resonance imaging (MRI) may be used to detect these anomalies (Goldberg et al. 2006). Pelvic cysts and debris are typical features of persistent cloaca, and ultrasonography and MRI can be used to diagnose persistent cloaca.

Functional studies of the bowel, genital, and urinary tracts have been scarce historically. However, recent technological advances have improved the ease with which these results can be collected from most patients (Hendren 1992). Cloacal malformation and VATER association lead to elevated risk of neurovesical damage, and renal failure is the most serious problem. Renal failure can result from persistent cloaca and VATER association by at least two mechanisms. First, unilateral renal agenesis, which occurs in approximately 20% of patients with cloacal malformations (Brock and Pena 1992), may induce poor renal function when the opposite renal unit exhibits reflux or obstruction (Hendren 1998; Liu and Hutson 2000). However, some patients diagnosed as unilateral renal agenesis after birth might have gone undetected before birth. Secondly, a long common cloacal channel leads to urine drainage and accumulation in the vagina, causing hydrocolpos and bladder dysfunction (Appignani et al. 1994; Shimada et al. 2005).

Urine escapes from the hydrocolpos via fallopian tubes into the abdominal cavity leading to ascites pooling. Later, chronic inflammation caused by urine and meconium accumulation blocks the fallopian tubes increasing hydrocolpos, as in our patient. In these patients, the infant should be delivered before function is lost from both kidneys.

Here, we measured renal arterial blood flow and renal size by ultrasonography. On the basis of kidney size and renal arterial blood flow, we diagnosed failure of the left kidney. During the first visit to our outpatient, the size and morphology of the right kidney, as well as arterial blood flow, appeared normal. However, right kidney became polycystic and the size increased during pregnancy. While determining the appropriate time for infant delivery can be difficult, when fetal renal failure is predicted and the estimated fetal body weight is sufficient to support survival outside of uterus, early delivery is essential. In addition, measurement of the amount of urine production and the Creatinine concentration in the amnion are effective methods for monitoring fetal renal function.

Early diagnosis and treatment can lead to near normal urogenital and digestive function. If hydronephrosis exists, hydrocolpos must be ruled out before performing an ureterostomy or a nephrostomy, as the hydrocolpos may interfere with ureter drainage. Since failure to accurately detect these abnormalities can lead to serious complications, fetuses with persistent cloaca with multiple anomalies should be examined by careful prenatal screening to prepare for prompt surgery after birth.

References


