Persistent urogenital sinus presenting as hydrometrocolpos and urinary ascites

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Received - 08 September 2019 Initial Review - 27 September 2019 Accepted - 16 October 2019

ABSTRACT

Persistent urogenital sinus (UGS) is a rare congenital cloacal anomaly whereby the urinary and genital tracts fail to separate during embryonic development. It can present as hydrometrocolpos (HMC) with severe vaginal stenosis/atresia, ambiguous genitalia, and/or urinary ascites. A term neonate born to a primigravida showed on antenatal ultrasound scan features suggestive of gross fetal ascites with severe oligohydramnios and hydronephrosis. Postnatally, the neonate presented with respiratory distress, urinary ascites, and abdominal mass and unilateral buphthalmos. An ascitic tap was done and HMC was drained. The cystogenitoscopy revealed vaginal stenosis for which vaginoplasty was done. The child was discharged and on follow-up is gaining weight. Our report shows an unusual presentation of a persistent UGS and also emphasizes the importance of early antenatal diagnosis and prompt post-natal management to reduce morbidity.

Key words: *Cloaca, Hydrometrocolpos, Hydronephrosis, Urinary ascites, Urogenital sinus*

Urogenital sinus (UGS) is a congenital cloacal anomaly comprising of a solitary common passage between lower parts of the urinary and genital tract that is urethra and vagina [1-3]. It can present as hydrometrocolpos (HMC), which is enlargement of the vagina along with the uterus due to accumulation of fluid or hydrocolpos, which is the enlargement of vagina due to accumulation of fluid or fetal ascites [4,5]. HMC could occur as a result of a cloacal anomaly or due to syndromic association or vaginal atresia, transverse vaginal septum, and imperforate hymen [5]. We report a rare case of persistent UGS with fetal ascites, HMC, severe vaginal stenosis, and unilateral buphthalmos. Our report essentially underlines the necessity of early antenatal diagnosis and prompt post-natal management to reduce morbidity.

CASE REPORT

A 25-year-old primigravida presented with an antenatal ultrasound scan (USG) at 32 weeks suggestive of gross fetal ascites with the right hydronephrosis and an 8×5 cm isoechoic mass with an external para- and post-spinal component and an intrapelvic component. There was no area of calcification or cystic area. This was misdiagnosed as a sacrococcygeal teratoma or neurogenic tumor. Oligohydramnios with an amniotic fluid index of two was noted.

A full-term neonate was delivered through caesarean section with birth weight of 2.7 kg. The child did not cry immediately after birth with APGAR scores of 2, 5, and 8 at 1, 5, and 10 min. The neonate had severe respiratory distress and required intubation and mechanical ventilation. General examination revealed retrognathia with low set ears and an 8 mm large left corneal opacity. Abdominal examination revealed gross ascites with transillumination positive and a pear-shaped mass 5×3 cm was palpable in hypochondriac region (Figs. 1 and 2). There was bilateral labial edema (left more than right) which was drained with incision to the left labia majora. Cardiovascular examination revealed ejection systolic murmur over the left upper sternal border. The rest of systemic examination was normal.

An ascitic tap was done that showed clear pale-yellow fluid, creatinine level of 0.9 mg/dl with a corresponding serum creatinine of 0.8 mg/dl suggestive of urinary ascites. Renal and liver function tests were normal. A post-natal USG of the abdomen revealed UGS with HMC, ascites, and right-sided hydronephrosis.

After the ascitic tap, the respiratory distress decreased. HMC was drained. The baby was successfully weaned off the ventilator. A two-dimensional echocardiogram revealed a 4 mm ostium secundum atrial septum defect with the left to right shunting of blood. A toxoplasmosis, rubella, cytomegalovirus, and herpes screen done was negative in both mother and child. USG of the left eye was normal. Cystogenitoscopy revealed vaginal stenosis for which a vaginoplasty was done. The infant was discharged and is on regular follow-up for buphthalmos and also to assess weight gain (Fig. 3).

DISCUSSION

Persistent UGS is a cloacal anomaly and its incidence is approximately 0.6 in 10,000 female births [3]. The cloaca is separated by the urorectal septum to form the UGS and anal canal,

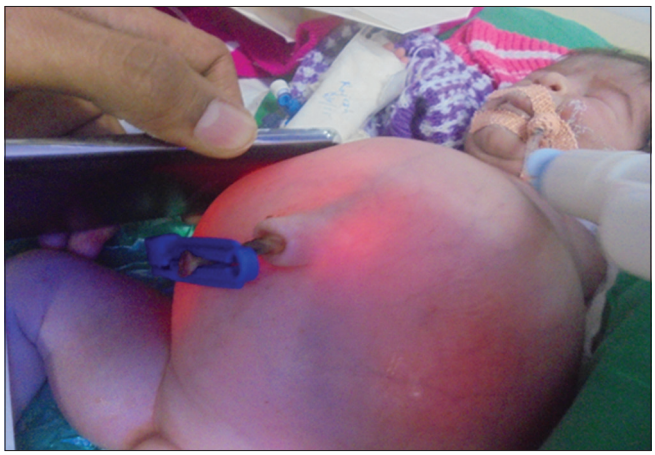

Figure 1: Gross abdominal distension due to ascites

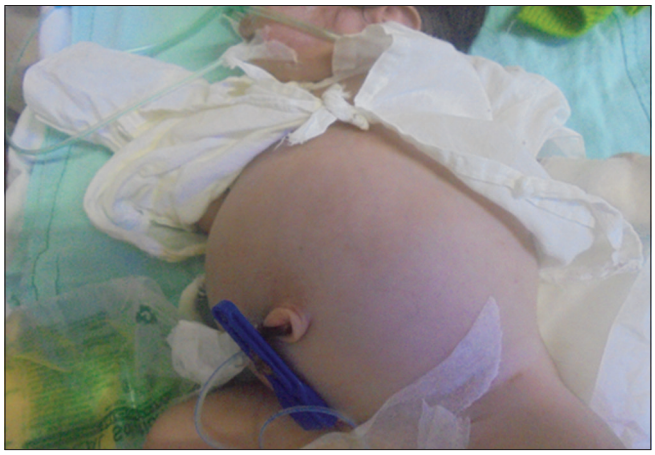

Figure 2: Transillumination test, demonstrating the ascitic fluid collection in the abdomen with pear shaped mass shadow, hydrometrocolpos, is seen in the hypochondriac region

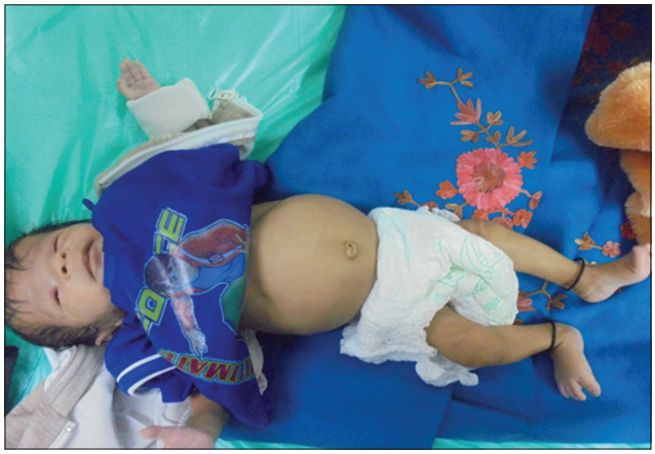

Figure 3: The child after drainage of ascites and the hydrometrocolpos

forms the urethra and lower one third of the vagina which appears at 8 weeks of gestation. Failure of the urethra-vaginal division results in a single passage draining the vagina and urethra and is referred to as a persistent UGS [3,6-9].

A persistent UGS usually presents with abdominal distension and frequently the patients have ambiguous genitalia. HMC (vagina and uterus are filled with fluid) could be due to a vaginal obstruction and reflux of urine from the urinary bladder into the vagina and uterus. This can also lead to ascites due to the retrograde flow of fluid from the uterus into the abdominal cavity. HMC in McKusick–Kaufman syndrome is due to cervical or vaginal atresia. Few females with HMC may present with syndromes such as cloacal dysgenesis sequence, Ellis-van Creveld, or Bardet–Biedl syndrome [5].

Pre-natal detection of the urogenital anomalies by USG is challenging due to its rarity, variation in manifestations, and poor quality of imaging. In fact, in our case, antenatal USG revealed an intrapelvic mass which was initially diagnosed as sacrococcygeal teratoma or neurogenic tumor. Although USG remains the first line of investigation, to further narrow down the differential diagnosis, fetal magnetic resonance imaging (MRI) is rapidly becoming an important modality of investigation as shown by Subramanian *et al*., Jackson *et al*., and Epelman *et al*. [10-12]. A fetal T1-weighted MRI could give better delineation of the anomalies and anatomical structures [10,11].

Postnatally, the lower abdominal mass could be due to HMC, seen in 15% of masses in that region in females. The other possibilities of abdominal-pelvic mass could be a distended bladder, bladder duplication, meconium pseudocyst, ovarian cyst, enteric duplication cyst, mesenteric cyst, rectal duplication, dilated bowel, or cystic neuroblastoma [6,13].

Loganathan *et al.* in a review of literature reported urinary ascites comprising of about 30% of all neonatal ascites cases [14]. Obstruction to the urinary tract is the most common cause of urinary ascites. A case series by Favre *et al.* in which 79 infants with ascites were analyzed, 15 cases had urinary ascites, of which 14 were due to posterior urethral valve [15]. UGS as a cause of urinary ascites is rare [14]. The blockage of the fallopian tubes due to chronic irritation of its mucosa could lead to HMC due to collection of secretions from the uterine and cervical glands [16]. The HMC could further lead to compression of the bladder, thereby obstructing urine output leading to oligohydramnios, reflux hydronephrosis, and urine diffusion into the peritoneal cavity causing fetal ascites. HMC in our case was due to obstruction to the outflow of secretions due to severe vaginal stenosis. Urinary ascites in this index case was confirmed by ascitic fluid biochemical examination of creatinine levels in the blood and ascitic fluid.

Management of HMC varies as per the underlying etiology. Persistent massive fetal ascites could cause respiratory embarrassment in immediate post-natal period as in our case was managed by mechanical ventilation and a therapeutic ascitic tap. Fetal abdominal paracentesis could help in decreasing respiratory compromise in the child, thus reducing the chance of ventilating a child [2,16].

The combination of HMC and cardiac anomalies in the absence of polydactyly makes the possibility of McKusick–Kaufman syndrome less likely in our neonate. In addition, the child also had vaginal stenosis which suggested a possibility of Bardet–Biedl syndrome (BBS); however, other features of BBS such as polydactyly, renal dysplasia, obesity, and retinal degeneration were not present in the child [17,18]. Screening of renal anomalies

in the presence of uterine anomalies is necessary because a third of uterine anomalies can be associated with renal abnormalities as in our case with the child presenting with unilateral hydronephrosis, which resolved spontaneously on follow-up [12].

Post-natal diagnosis was confirmed with USG of abdomen showing a persistent UGS. Features of ambiguous genitalia must be screened to rule out congenital adrenal hyperplasia [13,16]. Draining the ascitic fluid from peritoneal cavity and fluid from uterine cavity along with repair of the stenosis is a definitive treatment in such cases [17].

CONCLUSION

A high index of suspicion for persistent UGS must be kept as a differential diagnosis when a pelvic mass with fetal ascites is seen in antenatal scans, especially in neonates presenting with urinary ascites along with HMCs. Detection by pre-natal and immediate post-natal imaging will facilitate timely evaluation and early intervention including vaginal and urinary system decompression which would help to prevent complications.

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Funding: None; Conflict of Interest: None Stated.

How to cite this article: Ahmed M, Jayaswal SA, Malik S. Persistent urogenital sinus presenting as hydrometrocolpos and urinary ascites. Indian J Child Health. 2019; 6(10):578-580.

Doi: 10.32677/IJCH.2019.v06.i10.017