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A Case of Hydrometrocolpos and Polydactyly

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ABSTRACT: Neonatal hydrometrocolpos (HMC) is a rare Mullerian duct anomaly with an incidence of 0.006%. It occurs due to blockage of the vagina with accumulation of mucus secretions proximal to the obstacle. These secretions are secondary to intrauterine and postnatal stimulation of uterine and cervical glands by maternal estrogens. A triad of congenital HMC, polydactyly, and cardiac anomalies are the cardinal features of McKusick–Kaufman syndrome, which is also known as hydrometrocolpos-polydactyly syndrome. Bardet–Biedl syndrome is a well-known combination of hypogonadism, obesity, postaxial polydactyly, renal dysplasia, retinal degeneration, and mental impairment. In this case report, we describe a neonate with HMC, polydactyly, and hydronephrosis.

KEYWORDS: hydrometrocolpos, polydactyly, Bardet–Biedl syndrome, McKusick–Kaufman syndromes

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Introduction

Hydrometrocolpos (HMC) is a rare diagnosis and is even more rarely associated with polydactyly in neonates. It is a common presentation of Bardet–Biedl syndrome, McKusick–Kaufman syndrome (MKS), Ellis–van Creveld (EVC) syndrome, and Pallister–Hall syndrome. Neonatal HMC is caused by blockage of the vagina leading to accumulation of mucoid secretions proximal to the obstruction.[1] The mucous secretions are the result of both intrauterine and postnatal stimulation of uterine and cervical glands by maternal estrogens. A triad of congenital HMC, polydactyly, and cardiac anomalies are the cardinal feature of MKS, also addressed as "hydrometrocolpos-polydactyly syndrome".[2] This triad also occurs in Bardet–Biedl syndrome (BBS) along with hypogonadism, obesity, renal dysplasia, retinal degeneration, and mental impairment and also in EVC syndrome and Pallister–Hall syndrome.[3,4] We report a neonate with HMC, polydactyly, and hydronephrosis. The neonate was operated successfully and postoperative period was uneventful and is in regular follow-up today. The objective of this case report is to highlight the importance of antenatal detection and postnatal management of a diagnosed case of HMC.

Case Details

A late preterm, large for gestation, female infant was born to a G4P2A1L2 mother by caesarean section with a birth weight of 3.48 kg at 34 weeks of pregnancy. Baby cried immediately after birth and the Apgar score was 8 at 1 and 5 minutes of life. Maternal antenatal history was uneventful except for subclinical hypothyroidism detected during gestation. On a routine antenatal scan at 30 weeks of gestation, the fetus was diagnosed with right hand postaxial polydactyly (Fig. 1), midline cystic mass measuring 49 × 32 mm with low-level internal echoes behind urinary bladder, distension of Fallopian tube including the fimbrial end (Fig. 2), and minimal quantity of free fluid in the hip. A tentative diagnosis of HMC was made. Hydronephrosis of both kidneys with left pelvic dilatation more

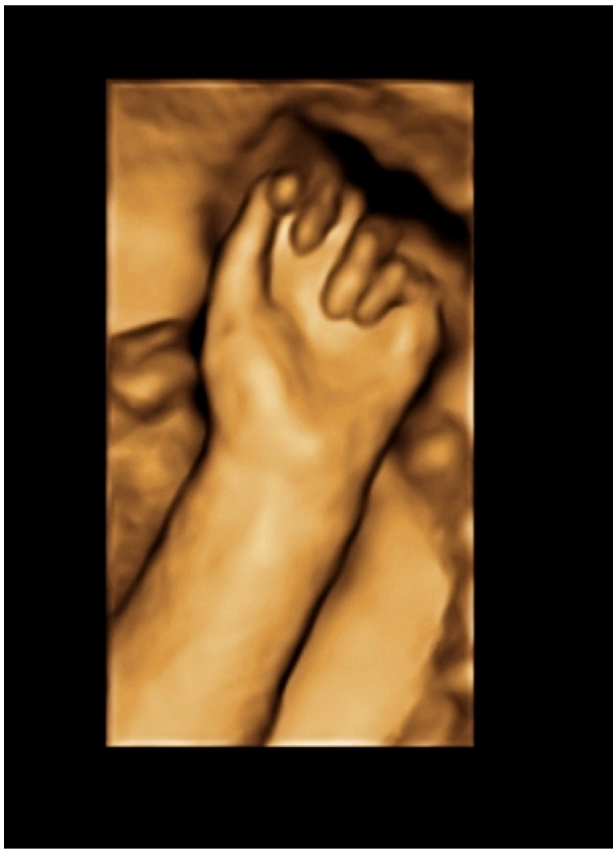

Figure 1. Antenatal scan showing polydactyly.

than right, echogenic left cortex, and normal right cortex were observed. Examination after birth revealed normal vital organs, no dysmorphism, postaxial polydactyly on right upper limb (Fig. 3), polydactyly on left lower limb (Fig. 4), distended abdomen, and palpable left kidney. No protruding mass from external genitalia. USG abdomen confirmed the diagnosis of bilateral hydroureteronephrosis with HMC. Abdominal CT scan performed before surgery

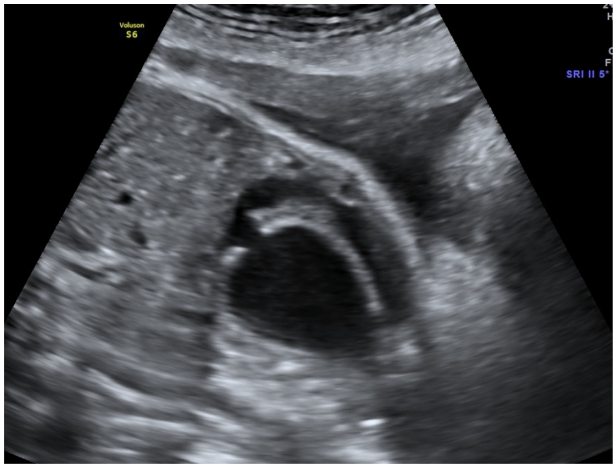

Figure 2. Antenatal scan showing dilatation of fallopian tubes. Also note that the uterine cavity is filled with the secretions.

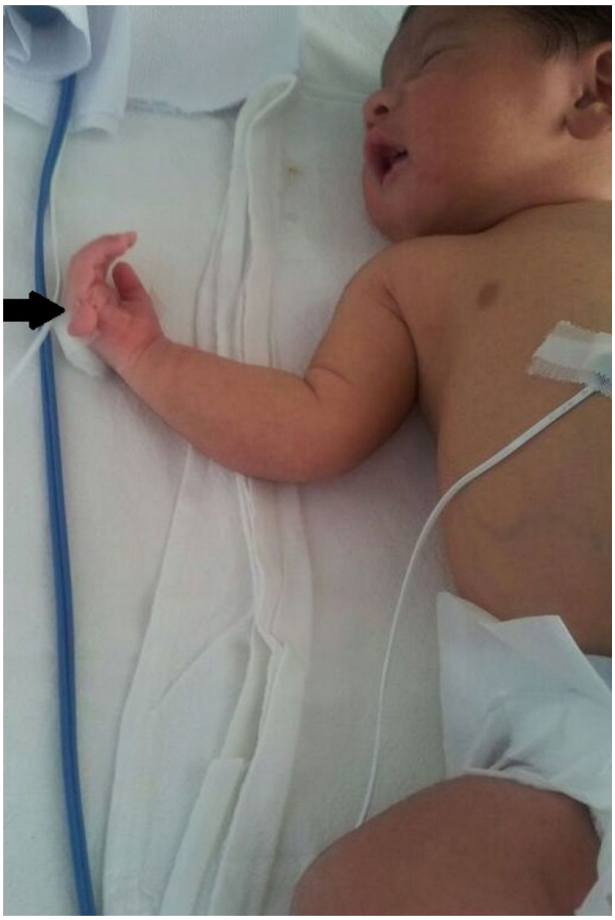

Figure 3. Postaxial polydactyly on the right upper limb (see arrow head).

Figure 4. Polydactyly on the left lower limb (see arrow head).

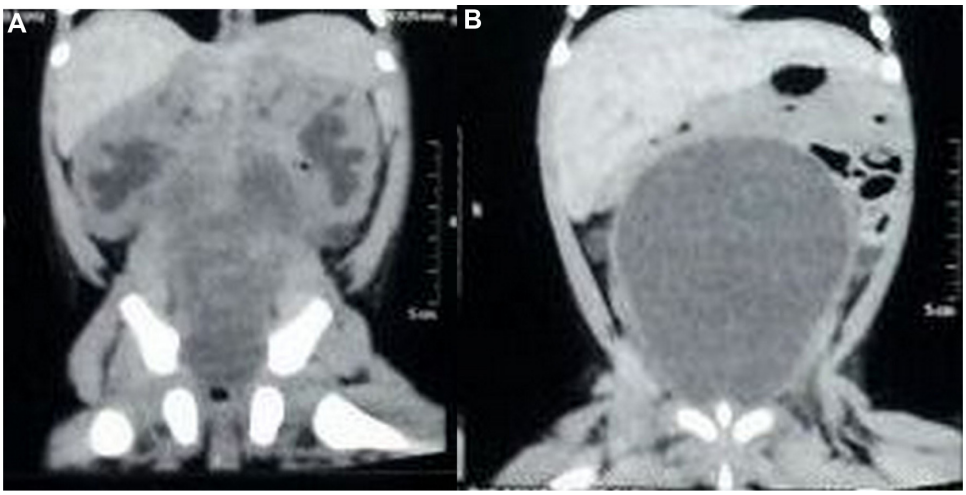

Figure 5. (**A** and **B**) CT scan showing large cystic area in pelvis and abdomen arising from pelvis posterior to urinary bladder measuring 11.7 × 6 cm with moderate hydroureteronephrosis of bilateral kidney.

revealed a large cystic area measuring 11.7 × 6 cm, arising from pelvis posterior to the urinary bladder (Figs. 5 and 6). Evaluation of other systems, including brain, heart, and skeletal survey, did not reveal any other malformations. The infant was operated on day 7 of life and the operative findings confirmed vaginal atresia with gross HMC with dilated vagina extending up to the epigastrium and uterus palpable in the right side of epigastrium. Thick viscid secretions were drained from the vagina and drains were placed with suprapubic tube and other across the membrane per vaginum. Infant was discharged at 15 days of age.

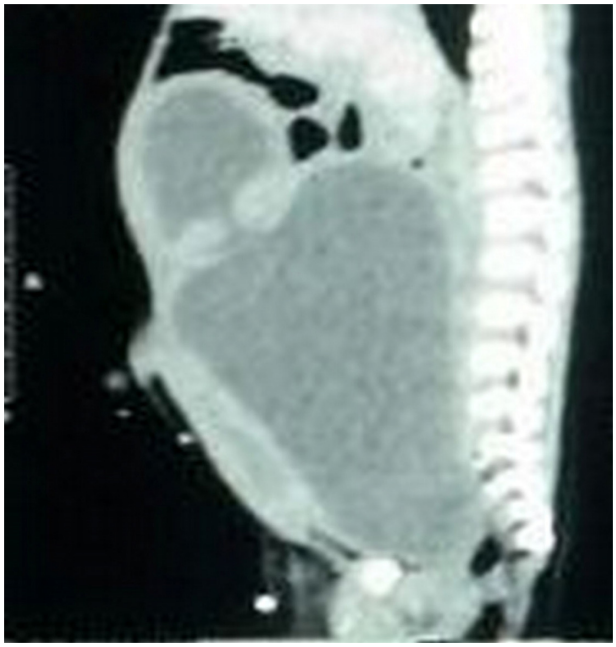

Figure 6. CT scan showing large cystic area in pelvis and abdomen arising from pelvis posterior to urinary bladder measuring 11.7 × 6 cm with moderate hydroureteronephrosis of bilateral kidney.

The diagnosis of MKS was considered as post axial polydactyly and HMC; however, there were no cardiac anomalies. In view of the absence of features such as retinal degeneration and renal anomalies, which are most common in BBS during neonatal period, it was not considered; also, obesity and mental retardation are usually seen in infancy.

Discussion

HMC is the distension of the cervix and uterus secondary to various causes. It is of two types.

1. Secretory type: It is secondary to accumulated mucus secreted by the uterine and cervical glands stimulated in utero by maternal estrogen.
2. Urinary type: It is secondary to the accumulation of urine in the presence of a vaginal obstruction. It is the result of urogenital or cloacal abnormalities.

The incidence of congenital HMC varies from 0.0014 to 0.1% in full-term newborn females with almost 90% having polydactyly. Neonatal HMC frequently results from anatomical malformations of the genital tract such as vaginal atresia, transverse vaginal septum, and imperforate hymen; which lead to blockage of drainage of the secretions, which in turn leads to HMC.[5,6]

Neonatal HMC is an obstructive Mullerian duct anomaly. The type of Mullerian duct anomaly is based on embryologic steps of lateral and vertical fusion during fetal life. During the process of lateral fusion, the Mullerian ducts develop at 5–6 weeks of gestational age from the coelomic epithelium simultaneously with Wolffian (mesonephric) ducts, which is placed lateral to coelomic epithelium. They usually fuse at about 7–9 weeks of gestational age in the midline to form the uterovaginal canal. During the process of vertical fusion at 8 weeks of gestation, the uterovaginal canal fuses with

urogenital sinus at the Mullerian tubercle; the urogenital sinus results from separation of the cloaca into the urogenital sinus and rectum. Simultaneously, the vaginal plate also develops distally. It initially undergoes proliferation and subsequently canalization. Hence, the vagina is formed by both the Mullerian ducts (upper two thirds) and the urogenital sinus (lower one third). Embryology reveals the different types of HMCs observed; secretory type occurs secondary to defect in upper two thirds and urinary type occurs as a result of defect in lower one third.[7]

Congenital HMC accounts for around 15% of intra-abdominal cystic masses in female infants. It has also been associated with urinary or intestinal tract abnormalities.[8] The clinical features are varied and usually seen as an abdominal mass that may compress the adjacent organs like bladder, ureters, bowel, or pelvic veins, resulting in urinary retention, constipation, ascites, lower limb edema, or rarely life threatening conditions.[9] Postnatally, it can be suspected when lower abdominal swelling persists in spite of urinary catheterization.[10]

Antenatally, ultrasonography remains the most widely used diagnostic imaging technique for identification of HMC and it is suspected when the scan shows the presence of large cystic abdominopelvic mass with a fluid-debris level.[11] MRI can also be used as a useful complementary tool for assessing fetal urogenital anomalies when ultrasonography is inconclusive.[12,13] Postnatally diagnosis of HMC is confirmed by ultrasound abdomen echography and CT scan.[14]

The definitive treatment involves drainage of the accumulated fluid in the uterine cavity and establishing communication between the vaginal epithelium and the vulva. In the cases of HMC secondary to imperforate hymen and low vaginal atresia, perineal approach is preferable. Treatment varies from the simple X-shaped hymenotomy for the isolated imperforate hymen to major surgery for complex urogenital abnormalities. Abdominoperineal approach is usually preferred in cases of high vaginal atresia.[15,16]

The MKS is induced by variations in the MKKS gene mapped onto chromosome 20p12 between D20S162 and D20S894 markers. The diagnosis of MKS in males is based on genital malformations (most commonly hypospadias, cryptorchidism, and chordae), postaxial polydactyly, and congenital heart disease (CHD). CHD includes atrioventricular canal, Ventricular septal defect (VSD), or hypoplastic left heart, which is present in nearly 10–20% of cases. Long-term cognition is normal.[11] Other associated findings include gastrointestinal abnormalities (28%): imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease and malrotation, and abnormalities (5%) of the eyes.[17]

HMC, polydactyly, and cardiac defects as well occur in BBS. The characteristic combination of findings in BBS are rod cone dystrophy in eyes (93–100%), polydactyly (58–69%), obesity (72–88%), learning disabilities (41–62%), hypogonadism in males (85–90%), and renal anomalies (25–100%). The secondary features include speech disorders or delays; eye abnormalities, such as complex female genito-urinary malformations, strabismus, cataract, and astigmatism; brachydactyly or syndactyly; developmental delays; ataxia; diabetes mellitus; craniofacial dysmorphism; nephrogenic diabetes insipidus; hepatic fibrosis; and congenital heart disease. The BBS phenotype is seen in individuals with mutations in 14 different genes.[18]

Other differential diagnosis includes EVC syndrome that is characterized by polydactyly, acromelic growth retardation, ectodermal dysplasia with dystrophy of nails, and cardiac anomalies (most common atrial septal defect), and Pallister–Hall syndrome that is characterized by facial anomalies, postaxial polydactyly, imperforate anus, and CNS anomalies like diencephalic hamartoblastoma.[19,20]

The limitation of our case report is that we were not able to perform the gene analysis of the infant to get the exact diagnosis of the syndrome. The suspicion of BBS or MKS is made based on the clinical features; and evaluation of an infant with MKS or BBS includes ultrasound study of pelvis, kidney and urinary bladder, skeletal radiographs, EKG, and echocardiogram. The BBS is confirmed by genetic analysis of BBS genes (BBS1–BBS11 genes) and the MKS is confirmed by genetic analysis of MKKS genes on chromosome 20p12 between D20S162 and D20S894 markers.[21] The EVC syndrome is confirmed by genetic analysis of EVC1 and EVC2 genes.[19] Pallister–Hall syndrome is confirmed by genetic analysis of GLI3 gene.[20]

Conclusion

HMC is a rare condition in the neonate and should be suspected when a prenatal ultrasound identifies a midline abdomino-pelvic mass. Prenatal diagnosis and early newborn imaging lead to early detection and treatment of these cases. Early treatment prevents complications secondary to compression and obstruction of surrounding structures.

Learning Points

- This is a rare malformation of female genital tract and can be diagnosed antenatally.
- Early and aggressive treatment is advocated to avoid complications secondary to obstruction and infections.
- Treatment consists of drainage of the accumulated secretions.

Author Contributions

Wrote the first draft of the manuscript: DS. Contributed to the writing of the manuscript: OTP, GMI. Agree with manuscript results and conclusions: DS, OTP, GMI, SM, GK. Made critical revisions and approved final version: SM, GK. All authors reviewed and approved of the final manuscript.

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