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 hydrenephrosis.

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

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 mucus 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 features 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 hydrenephrosis. 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. Hydrenephrosis of both kidneys with left pelvic dilatation more...
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.
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.

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

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**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.

**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.
Early and aggressive treatment is advocated to avoid 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 the 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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