IJN
Iranian Journal of Neonatology
http://ijn.mums.ac.ir

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Case Report

Urinary Type Hydrometrocolpos and Polydactyly in Two Newborns: Case Report

Ziba Mosayebi[1], Tahereh Esmaeilnia Shirvani[1], Vafa Ghorban Sabagh[2*], Maryam Ghavami-Adel[3], Bahareh Fasihpour[4], Hosein Dalili[2], Ali Fathi[4]

1. Family Health Institute, Maternal-Fetal and Neonatal Research Center, Tehran University of Medical Sciences, Tehran, Iran
2. Breastfeeding Research Center, Tehran University of Medical Sciences, Tehran, Iran
3. Department of Pediatric Surgery, Tehran University of Medical Sciences, Tehran, Iran
4. Breastfeeding Research Center, Tehran University of Medical Sciences, Tehran, Iran

ABSTRACT

Background: Abdominal masses secondary to urinary retention are rare among female neonates and approaching this pathologic condition, inevitably, poses a diagnostic challenge. Hydrometrocolpos is one example of this disease category which is responsible for only 15% of such cases whose association with polydactyly appears to be even less common. The present study aimed to report the accounts of two neonates with cystic abdominal masses diagnosed as urinary type hydrometrocolpos secondary to urogenital sinus anomaly. Autosomal recessive disorders are characterized by vaginal atresia with hydrometrocolpos, polydactyly, congenital heart defects, and non-immune mediated hydrops fetalis. The triad of hydrometrocolpos, polydactyly, and cardiac anomaly in the two patients presented herein is strongly suggestive of a case of McKusick-Kaufman syndrome.

Case report: This study reported two neonates with abdominal mass, polydactyly, and genitourinary tract malformation, with no family history. Relief of urinary obstruction by exploratory laparotomy, aspiration of fluid, and vaginal reconstruction gradually reversed the hydronephrosis and renal failure.

***Conclusion*:** It can be concluded that in hydrometrocolpos causing urinary obstruction, decompression of hydrometrocolpos can save the kidneys.

Keywords: Hydrometrocolpos, McKusick–kaufman syndrome, Polydactyly

Introduction

The presence of intra-abdominal cysts in the neonatal period is not common. The differential diagnosis could be hydronephrosis, anterior cystic hygroma, ovarian cysts, sacrococcygeal teratoma, anterior meningocele, and hydrometrocolpos (HMC) (1). Neonatal HMC accounts for 15% of intra-abdominal cystic masses. It is a rare Mullerian duct anomaly with an incidence rate of 1 in 16,000 live births (2). It occurs due to congenital blockage of the vagina with the accumulation of mucus secretions proximal to the obstruction. These secretions are whether secondary to intrauterine and postnatal stimulation of uterine and cervical glands by maternal estrogens or urinary in character.

Urinary HMC is found in patients with urogenital sinus or cloacal anomaly. The HMC may be isolated or associated with some genetic syndromes, like McKusick-Kaufman, Bardet-Biedl, Ellis-van Creveld, and Pallister-Hall syndromes. The triad of congenital HMC, polydactyly, and cardiac anomalies are the cardinal features of McKusick–Kaufman syndrome, which is also known as HMC-polydactyly syndrome (3). This case report describes two neonates with HMC, polydactyly, and hydronephrosis.

Case report

Case One

An early-term, appropriate for gestational age, female infant was born to a G3P2A1L1 mother by normal vaginal delivery at 37 weeks of pregnancy.

* *Corresponding author*: Vafa Ghorbansabagh, Breastfeeding Research Center, Tehran University of Medical Sciences, Tehran, Iran. Tel: 098 2161192407; Fax: 098 2166591315; Email: dr.sabagh@yahoo.com

Please cite this paper as:
Mosayebi Z, Esmaeilnia Shirvani T, Ghorban Sabagh V, Ghavami-Adel M, Fasihpour B, Dalili H, Fathi A. Urinary Type Hydrometrocolpos and Polydactyly in Two Newborns: Case Report . Iranian Journal of Neonatology. 2021 Jul: 12(3). DOI: 10.22038/ijn.2021.49155.1885

The Apgar score was nine at one and five min of life. Maternal antenatal history was unremarkable except for a history of prolonged rupture of membranes, reception of intravenous ampicillin intrapartum, and negative consanguinity. Results of laboratory tests, including blood urea nitrogen/creatinine ratio, were found to be within the normal range.

First, a routine ultrasound scan at 18 weeks of gestation was significant for slight echogenicity of both kidneys and the presence of polydactyly. Further prenatal ultrasound imaging at 20, 29, and 35 weeks of pregnancy remarked a suspected small subaortic ventricular septal defects (VSD), severe bilateral hydronephrosis, a cystic 44 mm pelvic lesion behind the urinary bladder, and mild ascites.

The neonate presented with mild respiratory distress during the first few hours of life which ceased after being treated with oxy-hood. Intravenous antibiotic therapy was initiated due to the positive history of premature rupture of membranes, and necessary laboratory tests and cultures were sent out. The clinical examination revealed abdominal distension, a huge suprapubic mass, and two palpable kidneys. Moreover, the anal opening was normal, marked by a mild anterior dislocation. Examination of the genitalia revealed a single urogenital opening and swollen labia majora. In the left hand and foot post-axial polydactyly and in the right foot polydactyl were observed (Figures 1 and 2).

The Plain Radiograph of the abdomen indicated a large soft tissue density in the central part of the abdomen with superior displacement of the bowel loops. Results of the ultrasound scan showed a huge suprapubic fluid-containing mass and mild ascites. It should be mentioned that other abdominal organs could not be assessed through ultrasound at the time.

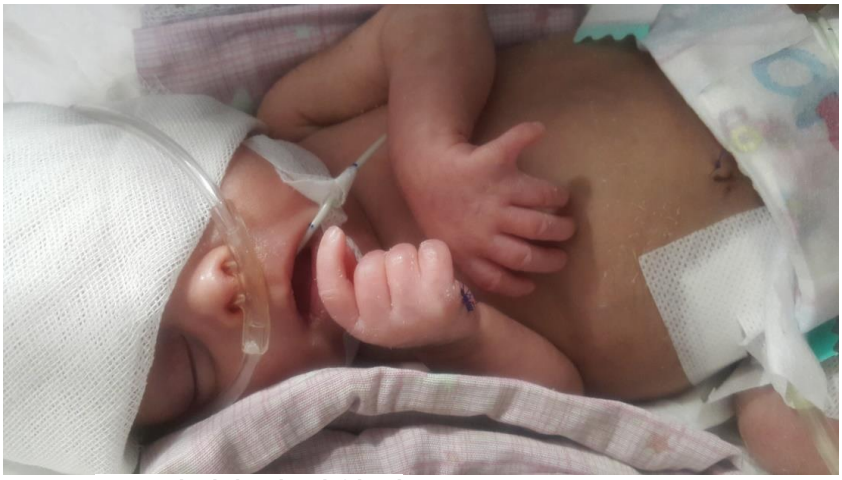

Figure 1. Post-axial polydactyly in left hand

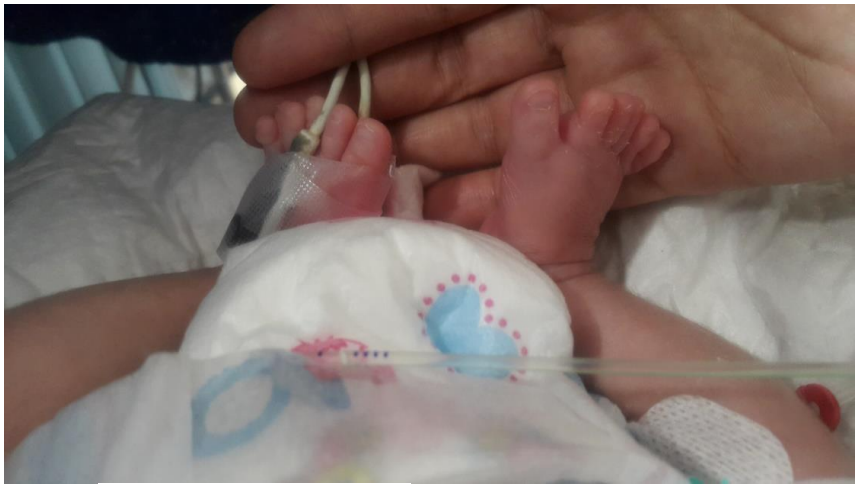

Figure 2. Post-axial polydactyly in right foot

Urinary catheter placement was initially tried with the purpose of ruling out urinary retention. However, the trial was unsuccessful and the catheter did not reach the bladder through the common urogenital opening, which greatly enhanced the probability of the existence of an anatomic abnormality. Therefore, suprapubic sampling was performed with the evacuation of 250 cc of clear liquid which was subsequently analyzed and found to have urine characteristics.

On repeated ultrasound scans, hydronephrosis of both kidneys was strikingly notable (right anterior-posterior [AP] diameter=13 mm and left AP Diameter=12 mm). The uterus was not conspicuously seen in the pelvis and the vagina appeared dilated, measuring at 44×33 mm, containing secretions and debris, imposing pressure on both ureters. Uterus was detected in the abdomen measuring at 24×30 mm with an endometrium thickness of 2.3 mm. It should be mentioned that other abdominal organs appeared to be normal.

Given high suspicion of HMC on the imaging test, a pediatric surgeon was consulted and the patient underwent exploratory laparotomy on the fifth postnatal day. Laparotomy revealed a severe inflammatory process accompanied by a 2×3 cm collection in the uterus. The collection was aspirated and the debris was sent for microbial culture. The vaginal investigation confirmed that the vagina and urethra opened to a common channel, suggestive of a low confluence urogenital sinus anomaly. A catheter was placed in the uterus of the patient in order to maintain proper drainage.

Post-surgical contrast study of the uterus was performed in order to rule out the presence of utero-vesical fistulas since the catheter was draining both secretions and urine. The contrast study did not indicate any filling defect or endometrial lesion in progress; however, a suspected narrow utero-vesical fistula was indicated (Figure 3). It should be mentioned that there were no post-surgical complications. Follow-up ultrasound scans indicated the gradual resolution of hydronephrosis while the diuresis of the patient was restored and renal function test results appeared normal. The specimen culture collected in the operating room was found positive for Klebsiella pneumoniae which prompted treatment with appropriate intravenous antibiotics. The postnatal echocardiogram confirmed the presence of a small VSD. The patient was discharged on the 21st day of life with a draining catheter after completion of the diagnostic and therapeutic measures, including intravenous antibiotic therapy. Follow-up cardiology visits and surgical vaginal repair were planned for the patient before discharge and integral counseling was provided for the parents.

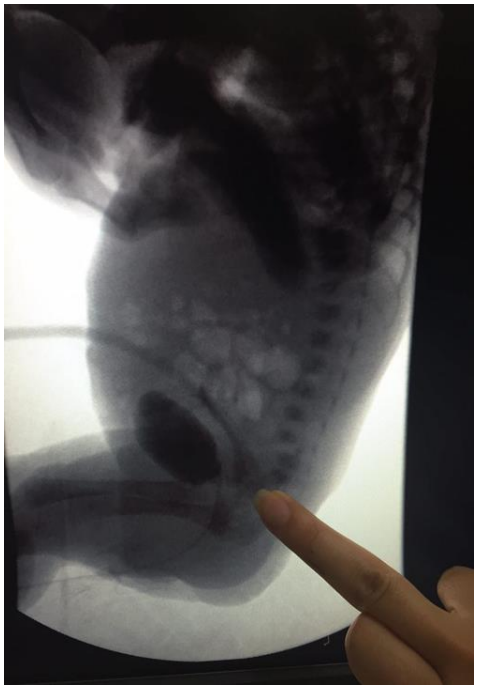

Figure 3. Narrow utero-vesical fistula

Case Two

A late preterm female infant was born to a G5P4A1L4 mother by normal vaginal delivery at 35 weeks and 6 days of pregnancy. The Apgar score was four at one min of life, reaching seven at five min by the aid of resuscitation. An unremarkable maternal antenatal history and negative consanguinity were reported. The ultrasound study of the fetus at 33 weeks of gestation showed severe ascites. The bladder was larger than normal with an adjacent cystic lesion of 35×33 mm in size. It should be mentioned that polydactyly of four limbs was also noted.

The baby was transferred to the neonatal intensive care unit after intubation, abdominal paracentesis, and primary stabilization at the delivery room. Moreover, 250 cc of clear liquid was evacuated through paracentesis and the fluid was confirmed to be urine after further analysis (creatinine of the sample was 10 mg/dL). Intravenous antibiotic therapy was initiated and followed by pertinent laboratory tests. On clinical examination, a large abdominal mass was palpated, the vagina was found to be atretic, and a single urogenital opening was noted. The anal

opening seemed to be normal, and the neonate had post-axial polydactyly in both hands and feet.

Further ultrasound scans revealed a huge cystic mass measuring at 35×40 mm in the abdomen and the uterus was not separately observable; a finding highly suggestive of HMC. Both kidneys were of normal size; however, mild bilateral hydronephrosis was reported with a pelvic AP diameter of 12 mm on both sides. The postnatal echocardiogram revealed mild mitral regurgitation, severe tricuspid regurgitation, and patent ductus arteriosus, for which proper cardiac management was performed. Results of lab tests, including kidney function tests, were found to be normal.

Vaginotomy and surgical catheter placement in the uterus were performed with the purpose of draining the secretions and reducing the intra-abdominal pressure. Voiding cystourethrogram was pursued after two weeks which was indicative of utero-vesical fistula. Gradual resolution of hydronephrosis on follow-up ultrasound studies was discerned along with the establishment of adequate diuresis and consequent normal renal function test results.

The patient was discharged on day 33 of life with a draining catheter after receiving appropriate treatment. Follow-up cardiology visits and surgical vaginal repair were also planned for the patient before discharge, similar to the previous case. Follow-up visits did not reveal any post-surgical complications. The probable diagnosis of McKusick-Kaufman syndrome (MKKS) was considered in both cases due to HMC, post-axial polydactyly, and cardiac anomalies. Due to financial restrictions, molecular genetic testing for MKKS was not performed in the two cases that were presented.

Discussion

Abdominal swelling of great size is a relatively uncommon presentation for infants. The most intuitive differential diagnoses in such neonates include neuroblastoma, intra-peritoneal fluid collection following organ failure, ovarian cysts, intra-abdominal sacrococcygeal teratoma, mesoblastic nephroma, bowel duplication, genitourinary anomalies, and anterior sacral meningocele (1). Congenital HMC as a cause of abdominopelvic mass in neonates has also been addressed in a few case reports in the last two decades (2). In the two cases presented in this report, abdominal distension was visibly evident and suprapubic masses were palpated in both kidneys. The HMC is a condition in which the uterus and vagina are distended by non-bloody retained fluid in the presence of distal vaginal outlet obstruction. It is considered a very rare condition with an incidence rate of 1 in 16,000 live births while the prevalence rate of imperforate hymen with hematocolpos has been reported to be 0.14% (3) (4).

It is important to highlight that the HMC is, by and large, of two types. In the secretory type, elevated maternal estrogen level leads to increased secretion from reproductive glands which, in turn, cause the accumulation of secretions due to vaginal blockage and distension of the vagina and uterus. Urinary type of HMC is secondary to the collection of urine in the presence of vaginal obstruction on account of urogenital sinus or cloacal anomalies (5).

Neonatal HMC is an obstructive Mullerian duct anomaly whose type is determined by the embryologic stages of lateral and vertical fusion during fetal life. During the lateral fusion, at 5-6 weeks of gestation, the Mullerian ducts develop from the coelomic epithelium simultaneously with and lateral to the Wolffian (mesonephric) ducts. At seven to nine weeks of gestation, they fuse together in the midline to form the uterovaginal canal. During the vertical fusion at eight weeks, the uterovaginal canal fuses with the urogenital sinus at the Mullerian tubercle, and the urogenital sinus results from the separation of the cloaca into the urogenital sinus and rectum. During the same period, the vaginal plate develops distally. First, it undergoes proliferation and then canalization followed by the formation of the vagina by both the Mullerian ducts (upper two-thirds) and the urogenital sinus (lower one-third). Embryology explains the manifoldness of HMCs, in that the secretory type develops secondary to a defect in the upper two-thirds while the urinary type is incurred by a faulty lower one-third (5).

The MKKS is an autosomal recessive disorder, first described by McKusick et al. in 1964 in two Amish siblings. Their rapidly-confirmed MKKS was induced due to mutation in the MKKS gene located at 20p12 between D20S162 and D20S894 markers (6). Mutation of this gene results in the formation of defective protein which leads to abnormalities in limbs, heart, and reproductive system. The HMC, polydactyly, and congenital heart diseases are the cardinal features of MKKS. Researchers have explained that HMC is usually diagnosed prenatally as the cause of abdominal cystic mass (7); however, in the present case, prenatal evaluations were equivocal. It should be mentioned that post-axial polydactyly is present

in 90% of cases who might also have syndactyly. Regarding the cases presented in this study, post-axial polydactyly existed in both neonates and the first case had syndactyly as well.

Congenital heart diseases concurrently found with MKS are atrioventricular canal, VSD, hypoplastic left heart, atrioventricular canal, tetralogy of Fallot, and left-sided superior vena cava which are present in nearly 10–20% of cases (8)(9). One of the cases presented in this study was confirmed to have a small VSD postnatally and the echocardiogram results of the other case indicated mild mitral regurgitation, severe tricuspid regurgitation, and patent ductus arteriosus.

Slavotinek et al. evaluated the most common features associated with MKK in 49 patients with MKK phenotype, 75% and 98% of whom were diagnosed at the time of birth and 6 months after birth, respectively. According to them, the most common manifestations were HMC (95%), hydronephrosis (63%), vaginal agenesis (59%), and infliction of the hands (29%) (3). Other associated findings are less common, including gastrointestinal abnormalities (28%), such as imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease, and malrotation.

Voiding cystourethrogram was performed on the second case and was indicative of a utero-vesical fistula. Given MKKS is a rare gene in males, its diagnosis in males is based on genital malformations, (most commonly hypospadias, cryptorchidism, and chordae), postaxial polydactyly, and congenital heart disease (8).

Cardinal manifestations of the MKKS gene in females are HMC and polydactyly, and this collection is often reported as the "HMC–polydactyly syndrome" in association with cardiac anomalies (10). Causes of HMC in most females are vaginal/cervical atresia or imperforate hymen (11). The HMC presents as a large cystic abdominopelvic mass causing compression of the surrounding structures and secondary hydronephrosis (9). Urogenital sinus is sometimes associated with HMC (12); other associated findings include gastrointestinal abnormalities (28%), such as imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease, and malrotation (13).

Some of these symptoms also exist in Bardet-Biedl Syndrome (BBS). It is of note to prognostically distinguish the two syndromes since MKKS is of a good prognosis while BBS is considered to have guarded prognosis. Therefore, these patients should be followed up for years as many features of BBS are not manifested till the age of 10-20 years old (10).

Early and aggressive intervention is advocated to prevent complications due to obstruction and concurrent infections. Treatment consists of surgical repair of the obstruction and drainage of the accumulated secretions. Transabdominal vaginostomy tube is a drainage technique to defuse the crisis till a definitive treatment is planned.

Conclusion

Overall, HMC is considered a rare condition in neonates whose prompt identification should not be overlooked but rather sought for when a prenatal ultrasound indicates a midline abdominopelvic mass in a female neonate. It may be isolated or associated with some genetic syndromes. Prenatal diagnosis and early newborn imaging make key contributions to the early detection and effective management of these cases. Early treatment prevents complications secondary to compression and obstruction of the surrounding structures.

Acknowledgments

The authors would like to appreciate all the colleagues involved in this case report and the parents who let them use the information of their neonates.

Conflicts of Interest

There was no conflict of interest in this study.

Ethical Considerations

This study is a case report and the identities of the patients were not used.

Authors' Contributions

This study was written by Vafa Ghorban Sabagh. Ziba Mosayebi, Tahereh Esmaeilnia Shirvani, Maryam Ghavami-Adel, Bahareh Fasihpour, Hosein Dalili, and Ali Fathi were involved either in the management of the patient or collection of the references.

Financial Disclosure

There was no financial benefit in this study.

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