

## Case report

### Approach to a newborn with urogenital sinus and hydrometrocolpos

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#### Abstract

Hydrometrocolpos is an accumulation of secretions in the vagina and uterus that can be a manifestation of syndromic and non-syndromic anomalies which can have many causes. Persistent urogenital sinus is one of the causes, in which there is a common channel of the urethra and vagina.

We report a case of a female preterm baby with a prenatal fetal ultrasound with hydramnios, moderate ascites and a retrovesical saccular image with thickened walls and residual content. She was admitted to NICU. No postaxial polydactyly (PAP) was noted, and echocardiogram showed a patent foramen ovale with a left to right shunt. Perineal examination revealed a well-formed vulva, with two perineal orifices, with a normally positioned anus. Abdominal sonography suggested a hydrometrocolpos, that latter was confirmed by MRI and raised the hypotheses of atresia of the lower third of the vagina or urogenital sinus. Bilateral ureterohydronephrosis was revealed. Cystography identified a persistence of the urogenital sinus. She underwent several catheterizations and aspirations so a vaginostomy to the abdominal wall was performed. Ophthalmology reported no abnormal findings. She was discharged home at 56 days of life with multidisciplinary follow-up. During the first year of life, she had several urinary tract infections. A genetic variant in probable homozygosity in the MKKS gene was found, with uncertain meaning. At thirteen months of life, she was submitted to a total urogenital mobilization and vaginostomy closure.

This newborn presented with HCM associated with PUGS but had no other clinical or genetic features of any syndromes. She does however have a genetic variant in the MKKS gene, not described before, which to date has uncertain meaning.

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**Keywords:** Hydrometrocolpos; McKusick-Kaufman syndrome; MKKS gene; Neonatology; Persistent urogenital sinus; Vaginostomy,

#### Introduction

Hydrometrocolpos (HCM) is a rare condition (reported incidence of 0.006%), where there is an accumulation of secretions in the vagina and uterus. The presence of secretions is caused by excessive stimulation by maternal oestrogen [1,2]. HCM is a manifestation of many different non-syndromic and syndromic anomalies. Imperforate hymen, vaginal atresia, persistent urogenital sinus (PUGS) and cloacal malformation are the main etiological factors [3].

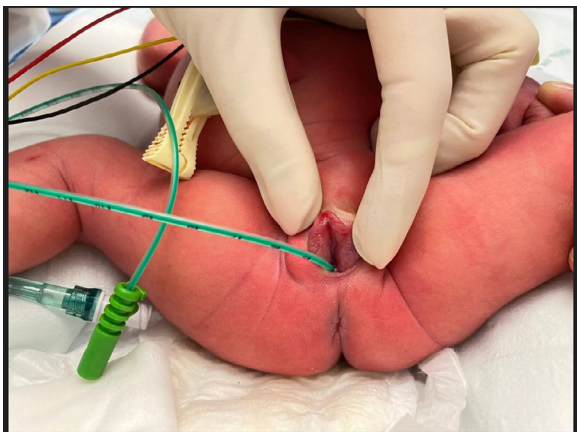
PUGS is a congenital condition where there is a common channel of the urethra and vagina, with an estimated incidence of 0.6/10,000 female births; it can be an isolated malformation or part of a syndrome. [4] There is a failure of urethrovaginal septation at six weeks' gestation that leads to drainage of the bladder and the vagina into a common channel. The perineum exhibits only two orifices, a ventral urogenital sinus and a dorsal anus. Sometimes HCM may be associated with syndromes, such as McKusick-Kaufman, Ellisvan Creveld or Bardet Biedl syndromes. [5]

McKusick-Kaufman syndrome (MKS) is a rare autosomal recessive syndrome, more common in females. Molecular genetic testing identified pathogenic variants in MKK gene [NDS1] 20p12, between D20S162 and D20S894. It's hallmarks, present in 94% of patients are: postaxial polydactyly (PAP), congenital heart defects and hydrometrocolpos (HMC) in females. Although these anomalies can be identified in prenatal ultrasounds, prenatal diagnosis is difficult. Bardet-Biedl Syndrome (BBS) is a generic name for a heterogeneous group of autosomal recessive disorders, characterized by retinal dystrophy or retinitis pigmentosa, PAP, obesity, nephropathy, and mental disturbances, or mental retardation and hydrometrocolpos. [6] The authors report a newborn case of hydrometrocolpos approaching prenatal and postnatal diagnosis evaluation, management, and treatment.

#### Case Description

Female preterm baby, born at 31 weeks, to a 24-year-old gravida 3 para 2. Pregnancy was uneventful with normal ultrasounds, until 30 weeks and 6 days when her mother was admitted with a threatened preterm labour. She underwent tocolysis and fetal lung maturation. At admission fetal ultrasound revealed hydramnios, moderate ascites and a retrovesical saccular image with thickened walls and a residual content. An urgent cesarean section was done at 31 weeks and 2 days due to a non-reassuring cardiotocography. Apgar score was 7/8/9. Anthropometry at birth was weight: 2299 g (>P97), length 45,5 cm (P90-97) and head circumference 30 cm (P50-90). She was admitted to NICU on non-invasive ventilation with respiratory and cardiovascular stability. No PAP was noted, and echocardiogram showed a patent foramen ovale with a left to right shunt. Perineal examination (figure 1) revealed a well-formed vulva, with absence of

vaginal opening a slightly inferior position of the urethra and a normally positioned anus. Abdominal sonography showed a cystic mass between the bladder and rectum measuring 57x24x36mm, suggestive of hydrometrocolpos.

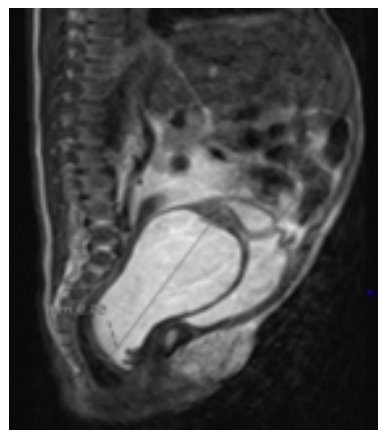


**Figure 1:** Newborn's perineal examination, with no vaginal opening and a inferior position of the urethra. Normal position of the anus.

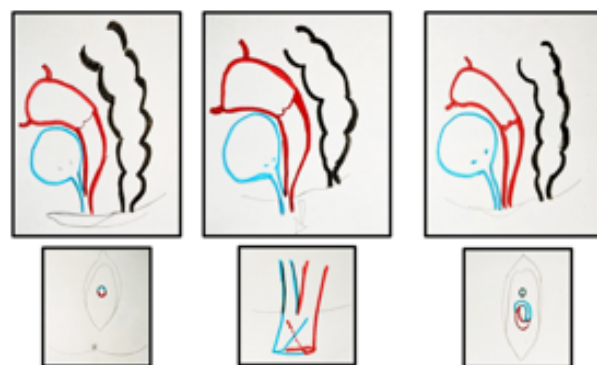
Abdominopelvic MRI (figure 2) confirmed the diagnosis of hydrometrocolpos (larger axis of 57 mm in the craniocaudal plane and 31mm in the anteroposterior plane) and raised the hypotheses of atresia of the lower third of the vagina or urogenital sinus (vaginal cavity with apparent termination in "pencil beak"). Bilateral uretero-hydronephrosis was revealed, more expressive on the left with pelvic and caliceal dilation, secondary to compression of the hydrometrocolpos. There was no renal dysfunction. Prophylactic antibiotics were started, and bladder catheterization was performed in order to relieve the compression effect of HMC. Cystography identified a common communication between the bladder and vaginal cavity, raising the diagnosis of persistence of the urogenital sinus. Hydrometrocolpos was drained but serial abdominal ultrasounds revealed recurrence of the hydrometrocolpos associated with feeding intolerance. Ultrasound-guided aspiration of HMC was repeated several times. At three weeks of life, she had a nosocomial urosepsis due to *Klebsiella pneumoniae* and completed 14 days of cefotaxime. At one month of age, bladder and vaginal catheterization were removed with recurrence of HMC, so a urethro-cysto-colposcopy was performed and a urogenital sinus was confirmed, with a common channel with approximately 2 cm and a vaginostomy to the abdominal wall was done without complications. Prophylaxis with trimethoprim was started. Throughout the hospitalization she remained stable. Ophthalmology reported no abnormal findings.

As the newborn didn't have the clinical diagnosis of McKusick-Kaufman syndrome, a multigene panel, that included the *MKKS* gene and other genes of interest for differential diagnosis, was performed. She was discharged home at 56 days of life with multidisciplinary follow-up (figure 3).

During the first year of life, she had several urinary tract infections (UTI) that required hospitalization due to *Klebsiella pneumoniae ssp pneumoniae ESBL+* and *Pseudomonas aeruginosa*. Prophylaxis was switched to nitrofurantoin. At thirteen months of life, she was submitted to total urogenital mobilization and vaginostomy closure (common channel pull-through) with unilateral circular W flap shape. Bladder and vaginal catheterization was maintained for 6 days. No complications were reported.



**Figure 2:** HCM (57 mm x 31mm). Vaginal cavity with apparent termination in "pencil beak" - atresia of the lower third of the vagina or urogenital sinus?



**Figure 3:** Visual schema of total urogenital mobilization and vaginostomy closure.

At eleven months it was found a genetic variant in probable homozygosity in the *MKKS* gene (with uncertain meaning). After surgery, she had a UTI due to *Klebsiella pneumoniae ESBL+* with a favorable course. Her last ultrasound showed normal kidney size and shape, but with hyperechogenicity of the pyramids and attenuation of corticomedullary differentiation. Hydrocolpos measured 17 mm. Prophylaxis was suspended at 15 months. She awaits cystography to exclude vesicourethral reflux.

## Discussion

HCM is a rare condition in newborns. We report a case of a female preterm with HCM associated with PUGS and enhance the importance of prenatal diagnosis and a timely postnatal treatment. The rapid diagnosis and surgical drainage reduces the comorbidities and seems to preserve future fertility and prevent endometriosis.

As in this case, the presentation can be an intraabdominal mass in utero and mass-related compression effects such as hydroureteronephrosis and bowel obstruction. [7] We enhance the difficulties of treatment approach in this case, with recurrence of HCM and its consequences such as vesical and bowel compression, so she had to go through several aspirations, catheterizations and a vaginostomy.

This newborn had PUGS-associated HCM without other clinical features of MKS or BBS. Genetic testing identified the probable homozygous variant: MKKS (NM\_170784.3):c.1382T>C (p.Leu461Ser), with no data available in the literature, analyzed by in-Silico predictors as moderately pathogenic and classified as of uncertain significance. No other clinically relevant variants were identified. Therefore, the genetic test was inconclusive.

Typically, McKusick-Kaufman syndrome is diagnosed in young children, whereas the diagnosis of Bardet-Biedl syndrome is delayed to the teenage years. [6] This case highlights the importance of a multidisciplinary team in the management and follow up of these newborns, such as obstetricians, geneticists, neonatologists, pediatric surgeons and ophthalmologists.

### Conflict of Interest Statement and Funding Acknowledgement

There was no conflict of interest to declare.

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