# McKusick-Kaufman Syndrome: Atretic Upper Vaginal Pouch; an Unusual Urogenital MR Finding

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ABSTRACT

McKusick-Kaufman syndrome is a rare autosomal recessive disease diagnosed by polydactyly, hydrometrocolpos, and congenital heart disease. We present an unusual laparotomy confirmed urogenital MRI finding (atretic vaginal pouch) in a 3-month-old girl with McKusick-Kaufman syndrome. Up to our knowledge, this MR finding has not been reported in the literature yet.



## CASE REPORT

### CASE REPORT

A 3-month-old female infant was referred due to urinary tract infection (UTI), multiple congenital anomalies, and renal failure. She was the first child of a consanguinity marriage, delivered by cesarean section at gestational age 36 weeks. The repeated prenatal sonography studies revealed large hyperechoic kidneys, dilated pyelocalyceal system, and oligohydramnios. The birth weight was 2710 gr, length 47 cm, and head circumference 33 cm (all indices on 50th percentile).

Clinical findings were pertinent for polysyndactyly of hands and feet (Figure 1) with normal nails, single urogenital orifice (Figure 2), anteriorly displaced anus, and abdominal distention with suprapubic mass. Ophthalmologic and ENT examinations were normal. The chromosomal study was compatible with 46, XX karyotype and female phenotype.

The radiographs of hands and feet demonstrated postaxial polysyndactyly (Figure 1).

Abdominopelvic sonography revealed a large cystic lesion with fluid-debris level posterior to the bladder in midline of abdominopelvic cavity, and moderate bilateral hydronephrosis (Figure 3).The kidneys were larger than normal, the long axis of the left kidney was 7.1cm and the right was 6.9cm. Serum creatinine was elevated (1.2mg/dl).

Cystography showed compressed bladder by posterior pressure effect with a fistula connecting the bladder neck to a small retrovesical blind pouch assumed to be atretic proximal vaginal canal. No vesicoureteral reflux was detected (Figure 4).

Echocardiography showed aortic root dilation. Chest X-ray, esophagography, brain sonography and CT scanning were normal.

MR imaging of the pelvis revealed a large inverted-pear shape, hyposignal T1W, hypersignal T2W, thin wall cystic structure in mid-pelvis pushing the bladder to the left side anteriorly with bilateral moderate hydronephrosis (Figure 5). The mid-pelvic cystic structure had a funnel-shape, closed, blunt, and inferiorly oriented neck.

MRI demonstrated a small hyposignal T1W, hypersignal T2W triangular fluid contained structure below the larger cystic structure, anterior to the rectum, and behind the bladder (Figure 6), corresponding to cystographic finding of atretic proximal vaginal canal.

During cystoscopy a single small orifice at the posteroinferior wall of the bladder was detected. Approach to the uterus via this orifice was not possible;thus, exploratory laparotomy was done and the collection of fluid was drained (Figurs 6,7). The laparotomy confirmed MRI and cystography findings of

hydrometrocolpos, enlarged bladder, and fistula between bladder neck and atretic proximal vaginal pouch.

## DISCUSSION

The presented case had rare associated anomalies including postaxial polydactyly, hydrometrocolpos, proximal vaginal atresia, dilation of aortic arch, vesicovaginal sinus, anterior displacement of anus, and renal failure. Syndromes consisting of polydactyly and hydrometrocolpos include McKusick-Kaufman syndrome, Bardet-Biedl syndrome, Pallister-Hall syndrome, Ellis-Van Creveld syndrome, orofaciodigital syndrome (type IV).

McKusick-Kaufman syndrome is an autosomal recessive disease. Hydrometrocolpos is present in 80-95% of females due to either vaginal atresia, imperforate hymen, or cervical atresia which leads to the development of an abdominopelvic mass with regional compression and secondary hydronephrosis (1). It is sometimes associated with urogenital sinus. In males, hypospadias and undescended testicles are the only gential abnormalities.

Postaxial polydactyly, mesoaxial polydactyly (rarely), or syndactyly is present in 90% of cases. Developmental dysplasia of the hips and lower extremity edema are other manifestations involving the limbs. Congenital heart disease (atrioventricular canal, ventricular septal defect, or hypoplastic left heart) is seen in 10-20% of cases (2). Additional less consistent findings are gastrointestinal abnormalities (28%) including imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease, and malrotation. Abnormalities of the eyes (5%) have also been described.

Bardet-Biedl syndrome is an autosomal recessive disorder characterized by retinal dystrophy or retinitis pigmentosa, postaxial polydactyly, obesity, nephropathy, and mental retardation.It is associated with mutation of BBS1-BBS10 gene and MKKS gene (2). Pallister-Hall syndrome is diagnosed by specific facial anomalies, postaxial polydactyly, imperforate anus, and brain anomalies including a diencephalic hamartoblastoma (3). There are reported cases of this syndrome with mutation of GLI3 gene that had overlapping features with MKKS (4). Our case had normal neurological development, ophthalmological examination, facial appearance, and nails, as well as unremarkable brain CT scanning.

In this case we found unique MRI appearance of atretic upper vaginal pouch as a small fluid contained space just below hydrometra and atretic cervix. Fluid signal in non-distended upper vaginal pouch without hydrocolpos could be an indirect sign of the presence of fistulous tract in any patient presenting with hydrometra and single external urogenital orifice.

Although ultrasonography remains the preferred modality for the initial study of cases suspected of having müllerian duct anomaly, MR imaging is suggested for patients with a technically inconclusive ultrasound examination. Console et al. compared MRI and ultrasound findings in 22 adult patients and concluded that MRI was an accurate imaging tool in uterine evaluation for planning treatment strategy (5).

Because the information obtained with MRI in evaluation of vaginal atresia is superior to ultrasound and CT scanning, it is the imaging examination of choice prior to surgical correction (6). In another retrospective study, negligible discordance (6.8%) was detected where the clinical and MR imaging of 44 females with congenital vaginal anomalies were compared (7). Marten et al. reviewed four patients suspicious to müllerian duct anomaly, who had laparoscopy or laparotomy confirmed MRI findings. They resulted that MRI was helpful in detection of müllerian duct atresia subtypes (8). MRI has an ability to optimally delineate uterine, cervical, and vaginal components; therefore, it may replace laparoscopy in the study of these organ anomalies. Unicornuate uterus and uterus didelphys, concealed hemorrhage within non-communicating horns, müllerian or cervical agenesis, and vaginal septa are the other anomalies that well defined by MRI and more information are given preoperatively in order to avoid diagnostic exploratory (9, 10, 11).

A recently published review article showed that endovaginal 3DUS had a very high sensitivity and specificity (100%) in accurately configuration of müllerian duct anomaly, but MRI had a wide range of sensitivity (100 to 28.6 %) and low specificity (66%) in delineating specific types of this anomaly; however, these two modalities need to be compared with each other in further studies (12).

Antenatal sonography of this case had failed to detect hydrometrocolpos and urogenital sinus. The presence of hyperechoic kidneys, dilated pyelocaliceal system, and oligohydramnios were an indirect clue to postrenal obstruction secondary to extrinsic distal urinary tract compression. Albeit prenatal ultrasonography is routinely used to screen fetal anomalies; this modality has a burden on detecting of hydrometrocolpos, urogenital sinus, cloacal, and other mulerian duct anomalies (13). Nevertheless, MRI is a useful modality to elucidate the cloacal anomalies where ultrasound alone is inconclusive (14, 15).

Although MRI is able to delineate atretic vaginal pouch, we should be alert to investigate the possibility of coexistent vesicovaginal fistula which is not detectable by routine MRI study.

## TEACHING POINT

Whenever a patient presents with MRI appearance of atretic vaginal pouch, cystographic evaluation for presence of vesicovaginal fistula is recommended. Fluid signal in non-distended upper vaginal pouch without hydrocolpos could be an indirect sign of the presence of fistulous tract in any patient presenting with hydrometra and single external urogenital orifice.

## ABBREVIATIONS

T1W= T1-Weighted
T2W=T2-Weighted
MKKS= McKusick-Kaufman syndrome
3DUS= Three dimensional ultrasonography
GRE=Gradient Echo

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## FIGURES

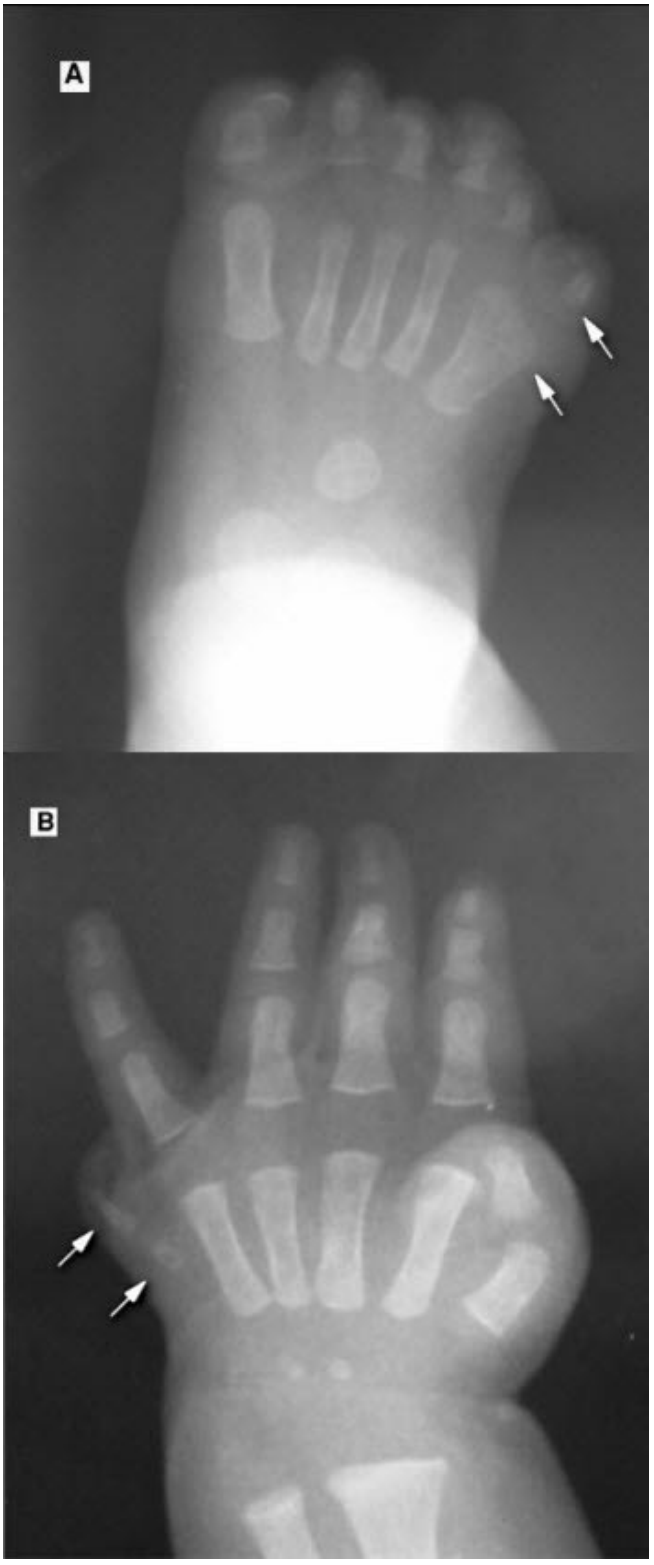


**Figure 1:** A 3-month-old girl with McKusick-Kaufman syndrome. Radiographs of foot (A) and hand (B) show postaxial polysyndactyly (arrows).

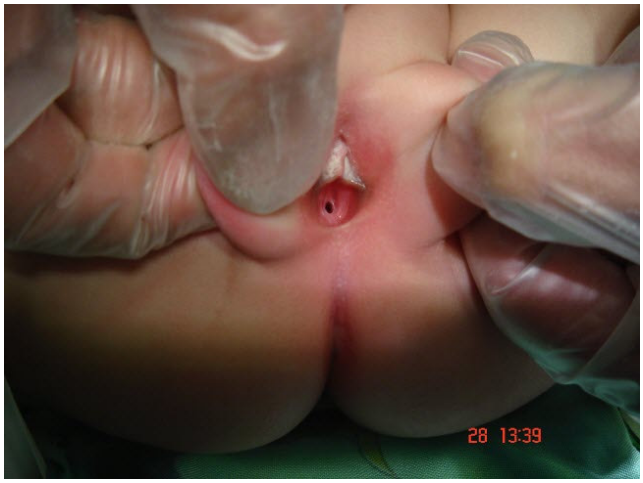


**Figure 2:** A 3-month-old girl with McKusick-Kaufman syndrome. Clinical image shows a single urogenital orifice.

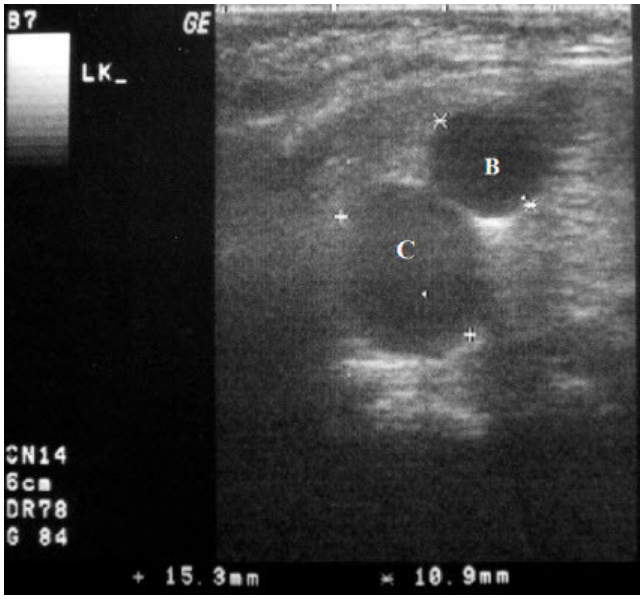


**Figure 3:** A 3-month-old girl with McKusick-Kaufman syndrome. The mid-sagittal sonographic image shows a hypoechoic cystic structure measuring 15.3 mm (C) posterior to the bladder (B) with fluid-debris level in dependent part.

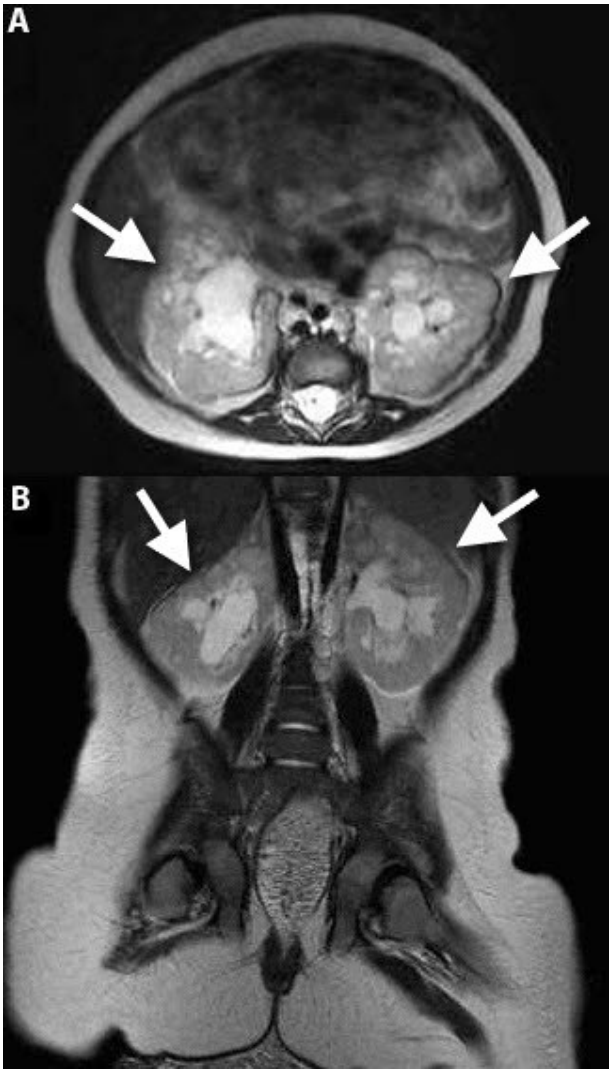


**Figure 5:** A 3-month-old girl with McKusick-Kaufman syndrome. Axial (A), Coronal (B) T2W MR images of kidneys shows bilateral hydronephrosis and dysplastic parenchyma (arrows).

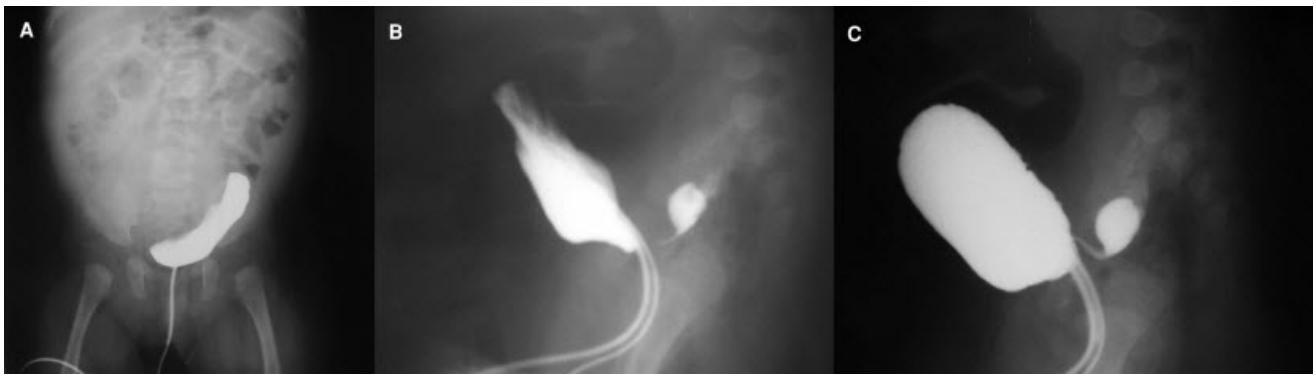


**Figure 4:** A 3-month-old girl with McKusick-Kaufman syndrome. Cystographic images: AP view (A), lateral view (B), and magnified view (C) reveal compressed bladder left anterolaterally by a posterior soft tissue density and a fistulous tract into a small posterior blind pouch.

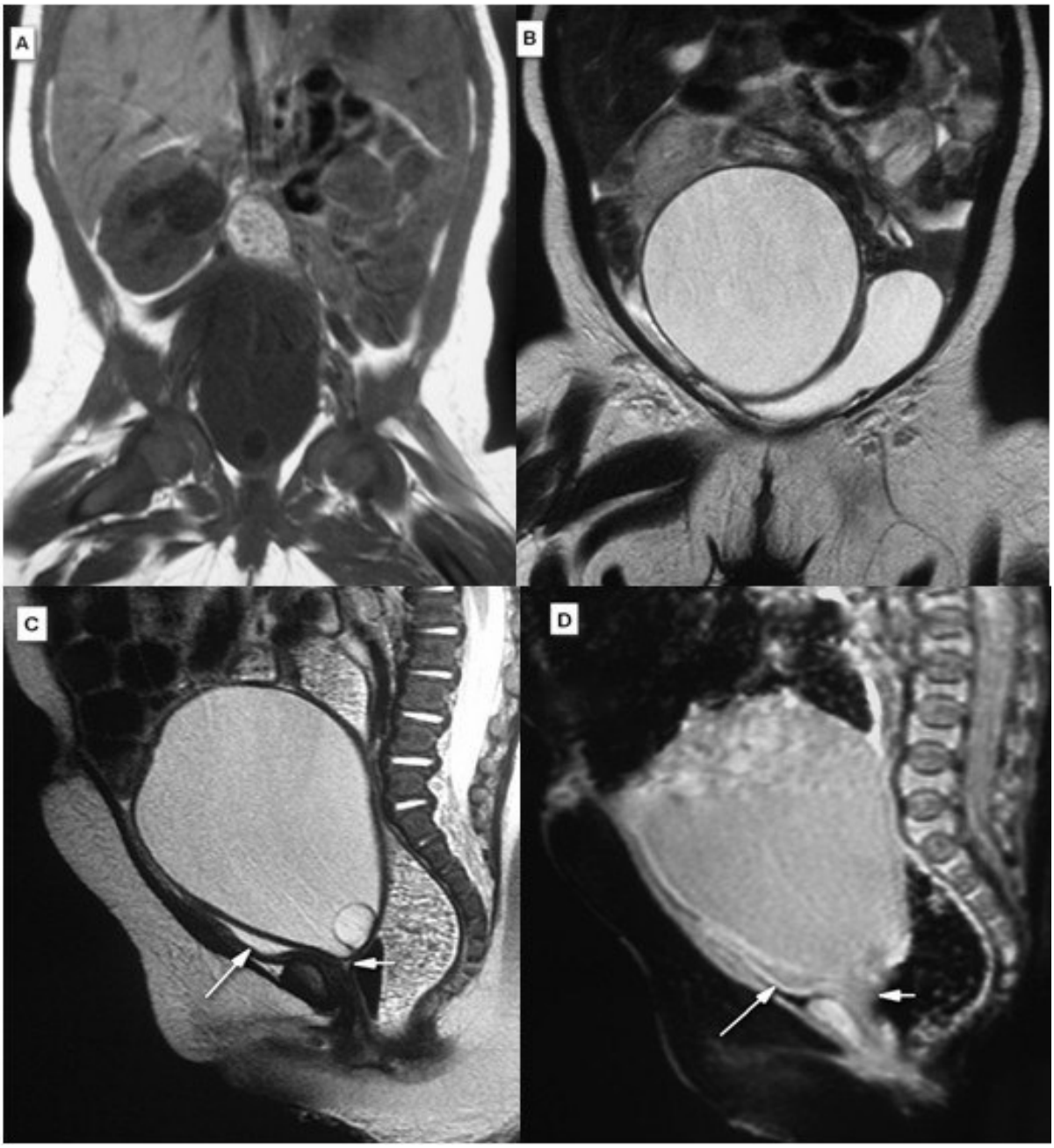


**Figure 6:** A 3-month-old girl with McKusick-Kaufman syndrome. MRI of abdominopelvic cavity : Coronal T1W (A), Coronal T2W (B), sagittal T2W (C), and sagittal GRE (D) reveal hydrometra, atretic cervix, compressed bladder (long arrow), and atretic proximal vaginal pouch (short arrow).

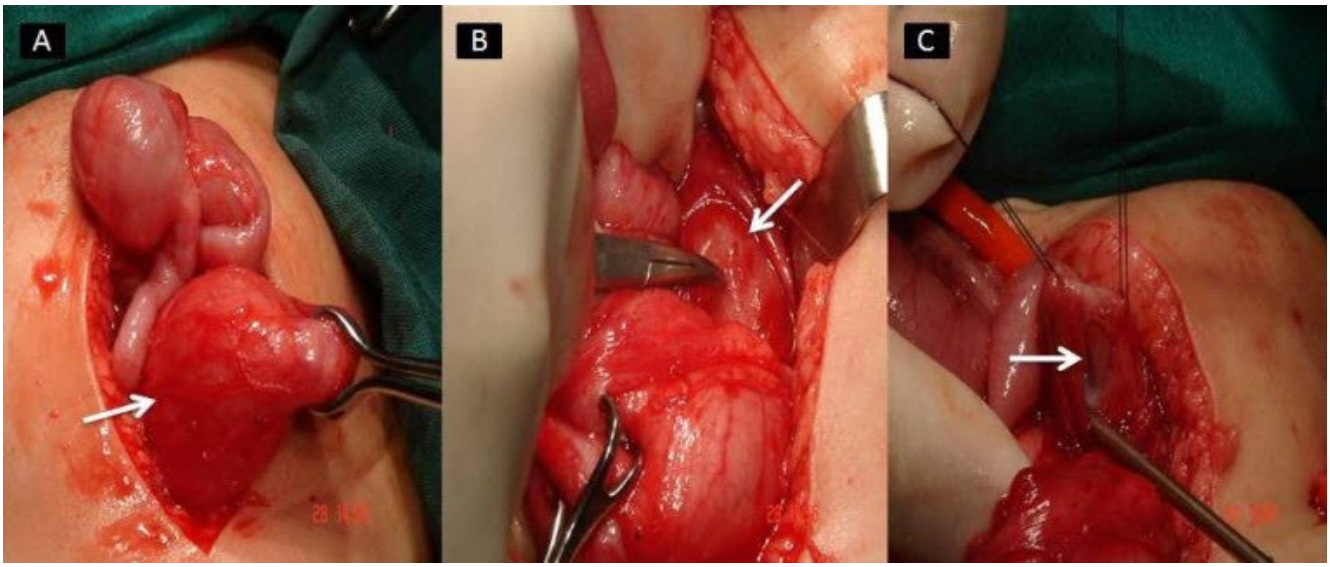


**Figure 7:** A 3-month-old girl with McKusick-Kaufman syndrome. Clinical photograph demonstrate hydrometrocolpos (A), bladder (B), and the fluid drained from uterus(C).

KEYWORDS

MRI, hydrometrocolpos, hydrometra, McKusick-Kaufman, polydactyly, vaginal atresia