

## Mckusick Kaufman Syndrome – Delayed Presentation in an Infant: A Case Report

Dr. Prashant S Patil, Dr. Kothari Paras L, Dr. Gupta Abhaya, Dr. Kekre Geeta, Dr. Dikshit Vishesh K, Dr. Kamble Ravi

Department of Paediatric surgery, L. T. M. G. Hospital, Sion, Mumbai, Maharashtra, India

### \*Corresponding author

Dr. Prashant Sadashiv Patil

Email: [docprashant2010@gmail.com](mailto:docprashant2010@gmail.com)

---

**Abstract:** A 7 1/2 month female child came with features of abdominal distension along with fever and frequency of micturition. Child was operated outside for similar complaints two months back. Examination revealed single opening in the introitus with postaxial polydactyly with syndactyly. Investigations including USG and CT abdomen showed cystic lesion behind the bladder. Abdominal exploration after genitoscopy revealed pyometrocolpos with high vaginal atresia. A temporary vaginostomy was done. We present a delayed presentation of McKusick Kaufman syndrome.

**Keywords:** Lump abdomen, Hydrometrocolpos, postaxial polydactyly, McKusick Kaufman syndrome, ultrasonography, Computed tomography

---

### INTRODUCTION

McKusick–Kaufman syndrome (MKKS) is a rare autosomal recessive syndrome. Cardinal features of neonatal MKKS include hydrocolpos, hydrometrocolpos, polydactyly, and congenital heart diseases [1-3]. Hydrocolpos or hydrometrocolpos caused by obstruction of the vagina accounts for 14-17 % of abdominal masses in newborn girls. Obstruction is secondary to an imperforate hymen, or a stenotic or atretic vagina.

### CASE REPORT

A 7 ½ month female child was brought by parents with complaints of abdominal distension, fever, frequent urination since 10 -15 days along with history of straining while passing stools. Baby was first issue of third degree consanguineous marriage, delivered normally at full term. Antenatal USG was normal. APGAR score was 8 and 9 at 1 min and 5 mins. Baby had normal developmental milestones. Family history was not significant.

The present complaints started with fever and loose motions 2 ½ months back for which the child was attended by a paediatrician. USG abdomen at that time was s/o fluid filled cyst. Child was operated by a general surgeon and the operative notes described it to be a cyst arising from vagina, contents of cyst were pus and serous fluid. Child was treated with antibiotics and IV fluids and discharged after 12 days. After a period of one month, the symptoms recurred, followed by gradual abdominal distension.

On general examination child was stable and active. Postaxial polydactyly in both hands and postaxial polydactyly with syndactyly were present in both feet (Fig. 1 & 2).



Fig. 1: Postaxial polydactyly in both hands



Fig. 2: Postaxial polydactyly and syndactyly in both feet

Abdominal examination revealed a tender large globular mass in the lower abdomen arising from pelvis. Superiorly it was palpable 3 cm above the umbilicus and laterally it occupied both the flanks. Liver and spleen were not palpable. Scar of previous operation was present in the right infraumbilical region. Hernial orifices were normal.

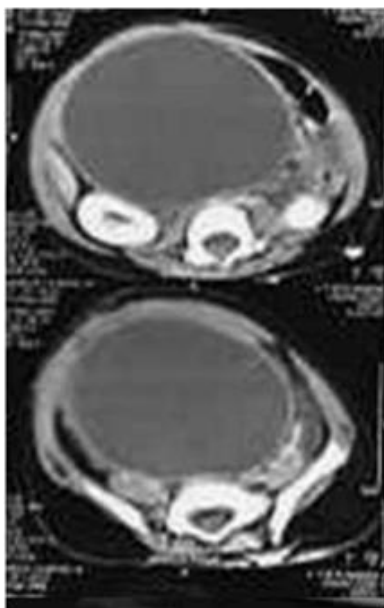
Genital examination revealed a single opening in the vulva through which the child is passing clear urine. Vaginal opening is not seen. There was no bulge in the vulva posterior to the previous opening (Fig. 3). Anal opening was normal.



**Fig. 3: Single opening in intoitus**

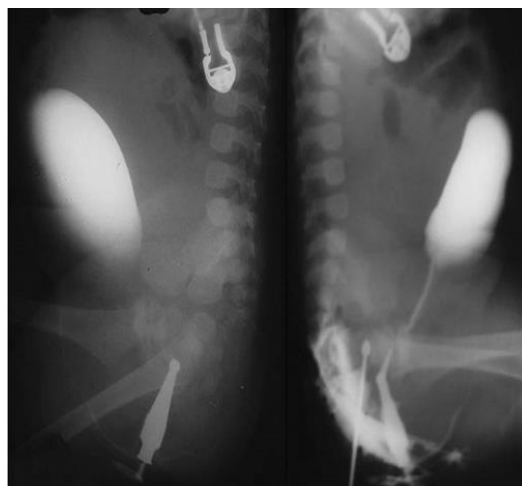
Per rectal examination revealed soft indentable swelling felt through anterior wall of rectum about 5 cm from anal verge and upper limit could not be reached. Cervical impression was not felt.

Child underwent USG and CT (Contrast) abdomen and pelvis (Fig. 4) which revealed a large cystic lesion posterior to collapsed bladder, extending superiorly. Uterus and adnexa could not be identified. Mild bilateral hydroureteronephrosis was present.



**Fig. 4: CT abdomen showing cystic swelling behind bladder**

Child was planned for genitoscopy with genitogram and exploratory laparotomy. Genitoscopy revealed single channel which was going anterosuperiorly and ending in the bladder. Bladder could not be distended properly and ureteric orifices were not visualized. Dye study was done which showed single channel leading to bladder with pressure effect from soft tissue shadow posteriorly. There was no evidence of reflux (Fig. 5).



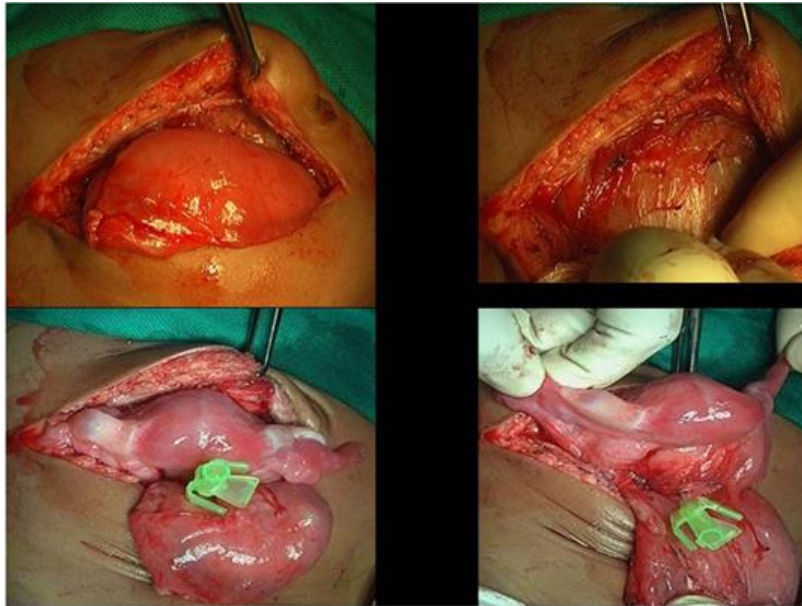
**Fig. 5: Intra-op contrast study showing normal bladder**

Child was explored through the previously operated scar. Intraoperative findings showed compressed bladder anteriorly, with markedly distended Vagina and uterus posteriorly extending into the epigastric region. As the swelling could not be delivered out of the wound, aspiration was done which revealed thin watery fluid followed by frank pus. About 480 cc fluid with pus was drained leading to collapse of the swelling (Fig. 6). Uterus and adnexa were delivered out and thoroughly inspected. Vaginotomy was done in the anterior wall. Telescopy confirmed blind ending vagina. In view of infected hydrometrocolpos simple vaginostomy was done in right iliac fossa, with plan to reconstruct vagina at a later date.

The child was retrogradely investigated with 2D Echo and fundus examination which were normal.

## DISCUSSION

McKusick-Kaufman was first described by McKusick *et al* [1] in 1964 in two Amish sibships and rapidly confirmed [2, 3]. This disease occurs due to mutation in the MKKS gene that is present at 20p12 location. Mutations in the MKKS gene result in formation of defective protein which is similar to the members of chaperonin family, leading to anomalous limbs, heart, and reproductive system [4].



**Fig. 6: Intra-op photo of cystic swelling before and after aspiration**

Cardinal features of MKKS in females are hydrometrocolpos and polydactyly, and it is often reported as the “hydrometrocolpos–polydactyly syndrome” in association with cardiac anomalies. Causes of hydrometrocolpos in most of the females are vaginal/cervical atresia or imperforate hymen. Hydrometrocolpos presents as large cystic abdominopelvic mass causing compression of the surrounding structures and secondary hydronephrosis [5, 6]. Urogenital sinus is sometimes associated with hydrometrocolpos. Though MKKS is rare in males, if present, it is sometimes associated with hypospadias, chordee, and cryptorchidism. Polydactyly is present in 90% of cases. It is predominantly post-axial and rarely mesoaxial. Syndactyly may also be encountered. MKKS shows association with congenital heart defects such as atrioventricular canal defects, ventricular septal defect, and hypoplastic left heart from 10% to 20% of cases. Other less commonly associated findings are gastrointestinal abnormalities (28%) that consist of imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease, and malrotation. Abnormalities of the eyes (5%) are also mentioned in literature [5]. Neonatal hydrometrocolpos is an obstructive Mullerian duct anomaly. Mullerian duct anomalies are basically of two types. They can be due to Mullerian agenesis or obstruction of the Mullerian duct. Third trimester antenatal USG or USG in the neonate can diagnose MKKS by the presence of large cystic abdominopelvic mass with a fluid-debris level. Adolescents usually present with amenorrhea and cyclic abdominal pain. Approximately, 45% of vaginal septa occur in the upper vagina, 40% in the middle vagina, and 15% in the lower vagina. In our case, stenosis in the lower third of vagina was found.

Bardet–Biedl syndrome (BBS) also presents with post-axial polydactyly and hydrometrocolpos. It is

an autosomal recessive disorder characterized by retinal dystrophy or retinitis pigmentosa, postaxial polydactyly, obesity, nephropathy, and mental retardation. The diagnosis can only be made if four of the five major manifestations are present in a person and remains a difficult diagnosis in infancy, as the appearance of several key features is delayed. Typically, MKKS is diagnosed in very young children, whereas the diagnosis of BBS is often delayed to the teenage years [4].

Other syndromes like Ellis–van Creveld syndrome characterized by polydactyly, acromelia, and cardiac anomalies and Pallister–Hall syndrome characterized by facial anomalies, postaxial polydactyly, imperforate anus, and CNS anomalies like diencephalic hamartoblastoma may also be considered among the differential diagnoses [7–9]. To conclude, MKKS should be given a thought in cases of neonatal abdominal distension, especially in females. All cases of diagnosed MKKS in infancy should be reevaluated for retinitis pigmentosa and other signs of BBS, as some of these children may be affected by BBS.

#### REFERENCES

1. Mckusick VA, Bauer RL, Koop CE, Scott RB; Hydrometrocolpos as a simply inherited malformation. *JAMA*, 1964; 189(11):813–6.
2. Dungy CI, Aptekar RG, Cann HM; Hereditary hydrometrocolpos with polydactyly in infancy. *Pediatrics*. 1971; 47(1):138–41.
3. Kaufman RL, Hartmann HF, McAlister WH; Family studies of congenital heart disease II: A syndrome of hydrometrocolpos, postaxial polydactyly and congenital heart disease. *Birth Defects*, 1972; 8(5):85–7.
4. Stone DL, Slavotinek A, Bouffard GG, Banerjee-

- 
- Basu S, Baxevanis AD, Barr M, Biesecker LG; Mutation of a gene encoding a putative chaperonin causes McKusick-Kaufman syndrome. *Nature genetics*, 2000; 25(1):79-82.
5. David A, Bitoun P, Lacombe D, Lambert JC, Nivelon A, Vigneron J, Verloes A; Hydrometrocolpos and polydactyly: a common neonatal presentation of Bardet-Biedl and McKusick-Kaufman syndromes. *Journal of medical genetics*, 1999; 36(8):599-603.
  6. Rosenberg HK, Chaudhry H; Pediatric pelvic sonography. In: Rumack CM, editor. *Diagnostic Ultrasound*. 4th ed. Philadelphia: Elsevier Mosby Publishers, 2011; 1936–8.
  7. Ruiz-Perez VL, Ide SE, Strom TM, Lorenz B, Wilson D, Woods K, King L, Francomano C, Freisinger P, Spranger S, Marino B; Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodistal dysostosis. *Nature genetics*, 2000; 24(3):283-6.
  8. Al-Khenaizan S, Al-Sanna N, Teebi AS; What syndrome is this? Chondroectodermal dysplasia: The Ellis-van Creveld syndrome. *Pediatr Dermatol.*, 2001; 18(1):68–70.
  9. Unsinn KM, Neu N, Krejci A, Posch A, Menardi G, Gassner I; Pallister-Hall syndrome and McKusick-Kaufmann syndrome: One entity? *J Med Genet.*, 1995; 32(2):125–8.