Clinics of Surgery®

ISSN 2638-1451 | Volume 11

Case Report Open Access

McKusick-Kaufman Syndrome: A Report of Four Cases

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Received: 08 Apr 2026
Accepted: 28 Apr 2026
Published: 30 Apr 2026
J Short Name: COS

Keywords:

McKusick-Kaufman Syndrome; Hydrometrocolpos; Postaxial Polydactyly; Congenital Heart Disease; Bardet-Biedl Syndrome

Citation:

Ahmed H AlSalem, McKusick-Kaufman Syndrome: A Report of Four Cases. Clinics of Surgery® 2026; V11(1): 1-8

1. Abstract

McKusick-Kaufman syndrome is a rare condition characterized by the combination of postaxial polydactyly, congenital heart disease, and hydrometrocolpos. Hydrometrocolpos in infants usually presents as a large cystic abdominal mass arising from the pelvis. This is caused by dilatation of the vagina and uterus because of the accumulation of cervical secretions from maternal estrogen stimulation. Hydrometrocolpos in these patients is caused by vaginal atresia, a transverse vaginal septum, or an imperforate hymen. Postaxial polydactyly is the presence of additional digits on the hand and the foot. A variety of congenital heart defects have been reported in patients with McKusick-Kaufman syndrome including atrioventricular canal, atrial septal defect, ventricular septal defect, single atrium or a complex congenital heart malformation. We report 4 newborns with McKusick-Kaufman syndrome outlining aspects of clinical features, diagnosis and management.

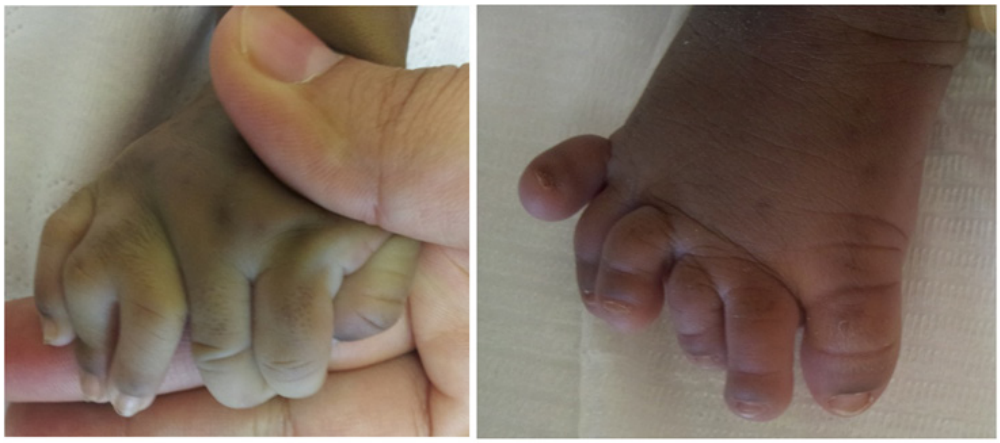
2. Introduction

McKusick-Kaufman syndrome is a rare autosomal recessive genetic disorder characterized by a triad of postaxial polydactyly, congenital heart disease, and hydrometrocolpos in females and genital malformations in males [1-5]. In males the most encountered genital malformations include hypospadias, cryptorchidism, and chordee [6]. It is caused by mutations in the MKKS gene on chromosome 20, leading to improper chaperonin protein function [7, 8]. This condition was first described in the Old Order Amish population, where it was estimated to affect 1 in 10,000 people. In the Amish population, the presentation of McKusick-Kaufman syndrome is variable as 70% of affected females have hydrometrocolpos, 60% have postaxial polydactyly, and 15% have congenital heart disease [9]. The diagnosis of McKusick-Kaufman syndrome is commonly established clinically based on the clinical diagnostic criteria which include hydrometrocolpos and postaxial polydactyly in the absence of clinical findings suggestive of an alternative diagnosis [9-12]. The diagnosis of McKusick-Kaufman syndrome can be confirmed by molecular genetic testing. The findings of biallelic pathogenic variants in MKKS identified by molecular genetic testing will confirm the diagnosis. It is also important to exclude Bardet-Biedl syndrome which is an allelic condition with considerable clinical overlap and age-dependent clinical features including retinal dystrophy, obesity, and intellectual disability [13-15[. We report 4 patients with McKusick-Kaufman syndrome outlining aspects of clinical features, diagnosis and management.

3. Case Reports

3.1. Case No. 1

A female newborn, a product of full term was born by caesarian section delivery. Her birth weight was 3.7 kg. She was referred to our hospital with abdominal distension. Her antenatal ultrasound showed congenital heart disease and a cystic pelvic mass. Clinically, she had abdominal distension and a palpable lower abdominal mass arising from the pelvis. She was also found to have polydactyly and syndactyly of the right hand and polydactyl of the right foot (Figures 1A and 1B). There was also an anterior ectopic anus (Figure 2). Her cardiovascular evaluation revealed common atrio-ventricular (A-V) canal with single atrium, moderate patent ductus arteriosus and severe A-V regurgitation. Abdominal ultrasound and CT-scan showed hydrometrocolpos and bilateral hydronephrosis and hydroureters and left perinephric fluid collection (Figures 3A and 3B). She was operated on and surgery revealed hydrometrocolpos secondary to a low vaginal atresia. An abdomino-perineal vaginal pull through was performed. The post-operative period was uneventful, and she was discharged home on the tenth postoperative day. She is now 12 years old and doing well with no other complaints.



Figures 1A and 1B: Clinical Photograph Showing Right Hand a d Right Foot Polydactyly. Note Also The Associated Syndactyly of The Right Hand.

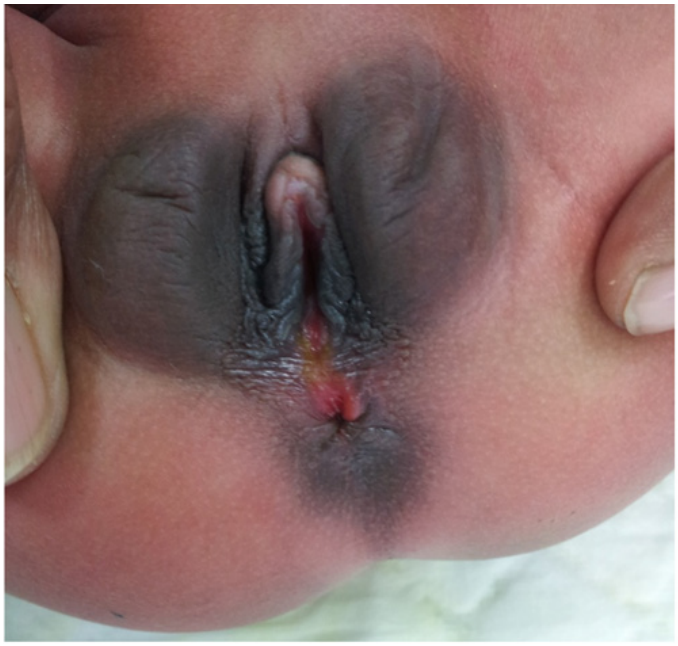
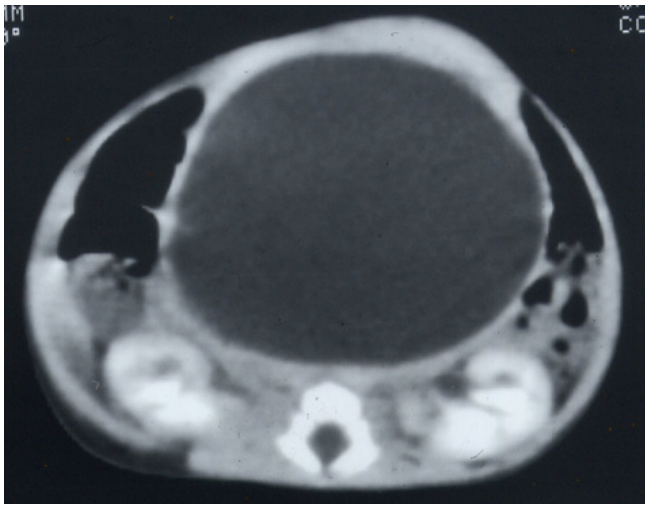


Figure 2: A clinical photograph showing an associated anterior ectopic anus in a newborn with McKusick-Kaufman Syndrome.



Figures 3A and 3B: CT-scan of the abdomen and pelvis showing hydrometrocolpos in a newborn with McKusick-Kaufman Syndrome. Note the large cystic mass in the first photograph representing the markedly distended vagina.

3.2. Case No. 2

A 3-days old female was referred to our hospital with abdominal distension and respiratory distress. Clinically, she was found to have abdominal distension with a palpable large abdominal mass arising from the pelvis (Figure 4). She was having cyanosis and mild respiratory distress. She was found to have polydactyly involving both upper and lower limbs (Figures 5A and 5B). Perineal examination revealed an anteriorly displaced anus and a single urethral opening but no apparent vaginal opening. Abdominal and pelvic ultrasound and CT- scan revealed a large 10x14 cm cystic mass arising from the pelvis and extending into the abdomen. There was also bilateral hydronephrosis and hydroureters (Figures 6A and 6B). The urinary bladder was also distended. Her cardiovascular evaluation revealed a single atrium. She was diagnosed to have vaginal atresia with anterior displaced anal opening. She underwent an abdomino-perineal vaginal pull through and postoperatively she did well and was discharged home three weeks in good general condition. She was seen last when she was 13 years old, doing well and no other complaints.

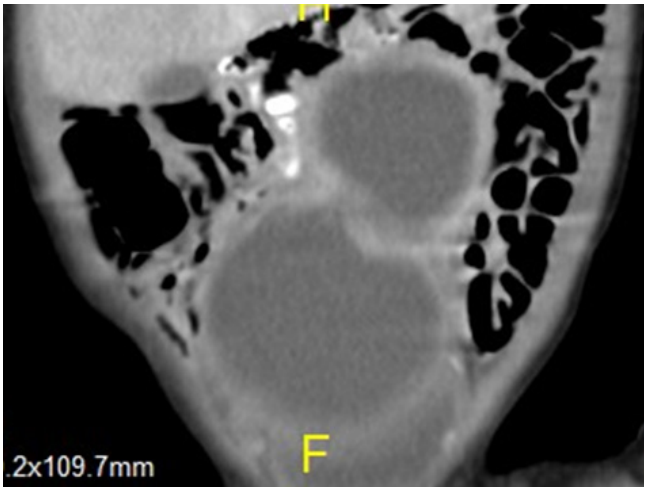
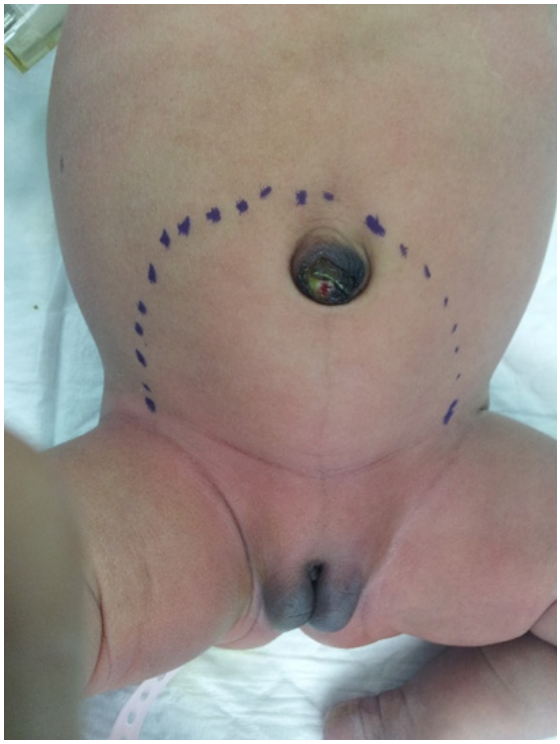
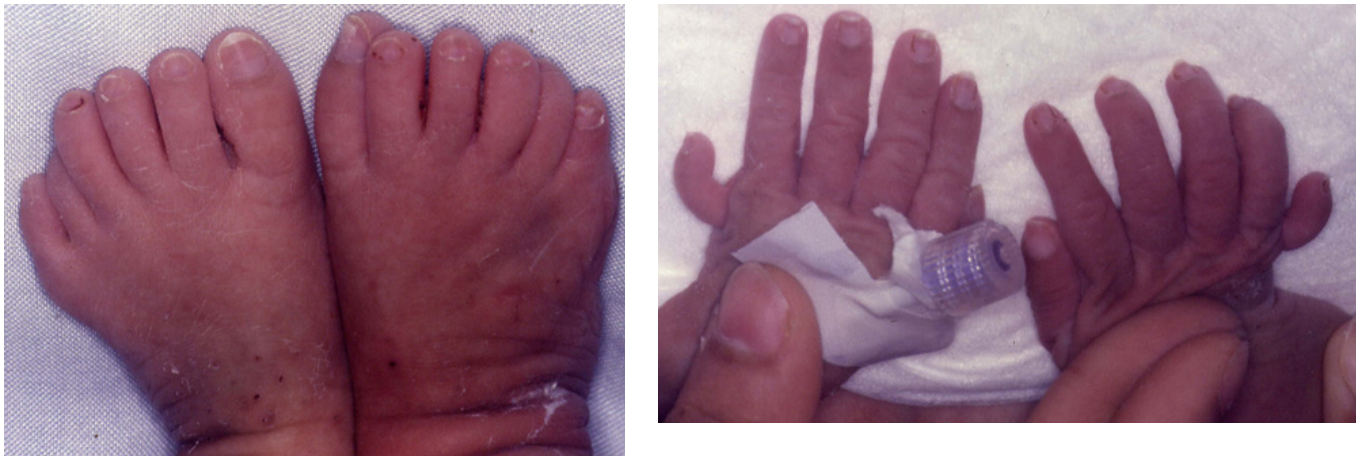


Figure 4: A clinical photograph showing a large abdominal mass arising from the pelvis and extending into the abdominal cavity.



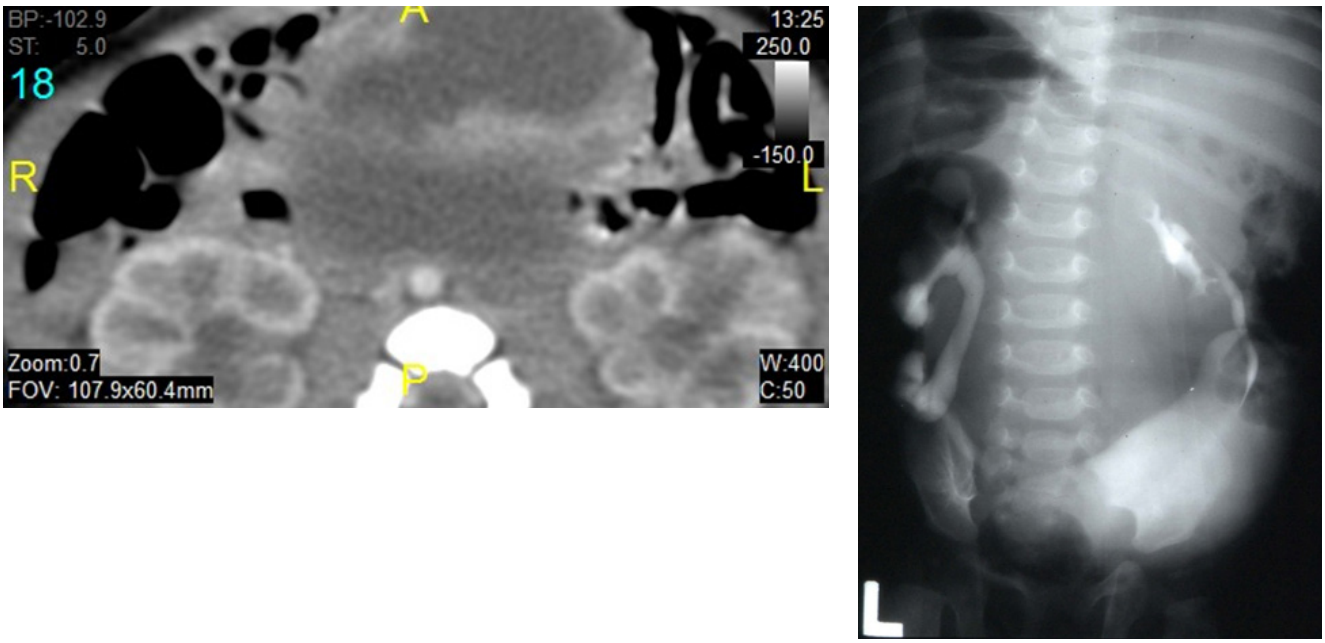
Figures 5A and 5B: Clinical photoraphs showing bilateral upper and lower limbs polydactyly in a newborn with McKusick-Kaufman Syndrome.



Figures 6A and 6B: CT-scan and intravenous urography showing bilateral hydroureters and hydronephrosis in a newborn with McKusick-Kaufman Syndrome. The hydroureteronephrosis was secondary to pressure effect from the markedly distended vagina and uterus (Hydrometrocolpos).

3.3. Case No. 3

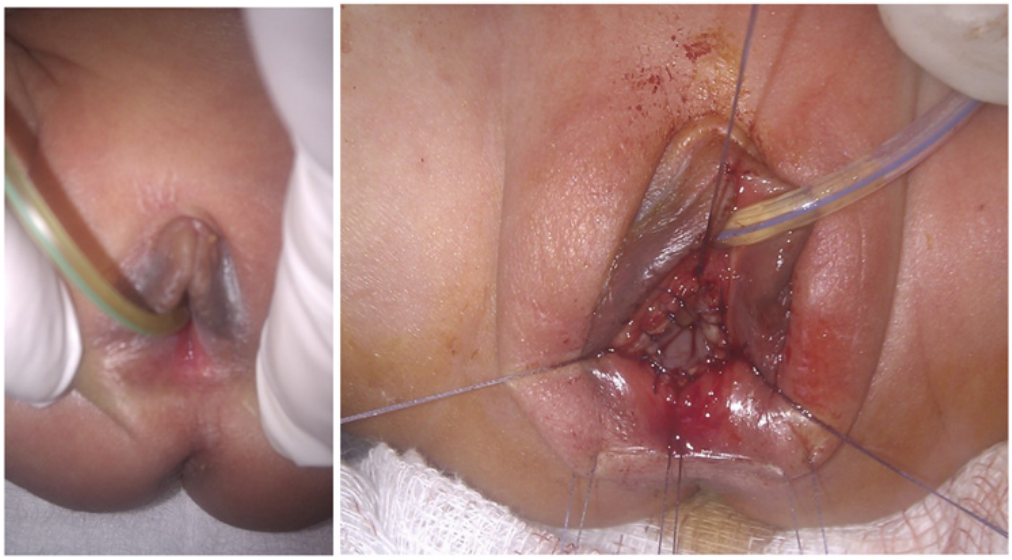
A one-week-old female was referred to our hospital with a recurrent abdominal distension and abdominal mass. She was diagnosed to have vaginal atresia and hydrometrocolpos. She had surgery in another hospital, the details of it were not clear. Clinically, she was well with abdominal distension and a palpable abdominal mass arising from the pelvis. She was also having a Foleys catheter which was draining a clear urine. She was found to have polydactyly involving both upper and lower limbs. Her cardiovascular evaluation was reported as normal. Abdominal and pelvic ultrasound and CT- scan revealed a large cystic mass arising from the pelvis and measuring 8x9 cm representing a distended uterus and vagina. There was an associated bilateral hydroureteronephrosis. She was diagnosed to have vaginal atresia with hydrometrocolpos. She was operated on and underwent an abdomino-perineal vaginal pull through (Figures 7A and 7B). Postoperatively, she did well and was discharged home two weeks postoperatively. On follow-up, she was last seen in the clinic when she was 14 years old, doing well with no other complaints.



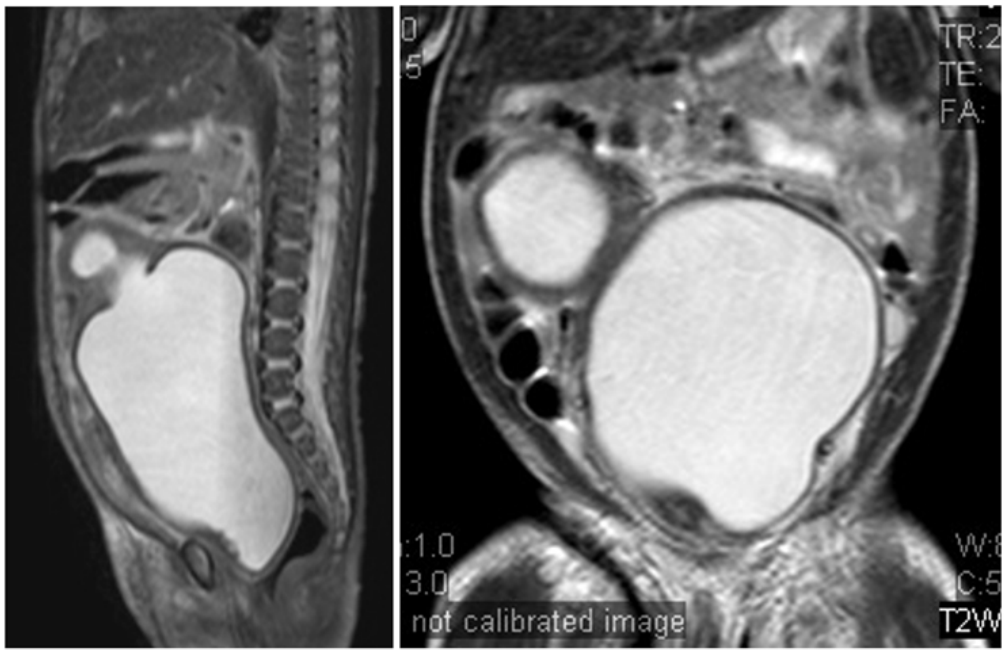
Figures 7A and 7B: Clinical intraoperative photographs showing vaginal atresia and postoperative abdominaopeineal vaginal pullthrough. Note the singel perineal opening with a Foley's catheter inseted into the unrinary bladder and no apparent vaginal opening. Note also the newly created vaginal opening in the second photograph following an abdominopeineal vaginal pullthrough.

3.4. Case No. 4

A one-day old female, a product of 37 weeks' gestation and normal vaginal delivery. Her birth weight was 3.1 kg. Her antenatal ultrasound revealed hydrometrocolpos associated with obstructive uropathy. Clinically, she was found to have abdominal distension with a large palpable abdominal mass arising from the pelvis. She was also found to have postaxial polydactyly involving all four limbs. Her investigations revealed normal renal function and normal electrolytes. Her abdominal ultrasound and MRI showed marked hydrometrocolpos, severe bilateral hydroureteronephrosis and distended urinary bladder. There was a large midline cystic mass behind the urinary bladder filled with hypoechoic fluid with homogenous low-level internal echoes. The mass measured 6.9x8.2x11.2 cm (Figures 8A and 8B). This was associated with hydronephrosis and hydroureters. She underwent cystoscopy with Foley's catheter insertion and percutaneous pigtail catheter drainage of the hydrometrocolpos. A contrast study on the 7th day showed reduced vaginal distension. A repeat ultrasound showed reduction in the degree of hydroureteronephrosis. The catheter was removed. The patient was observed but there was reaccumulation of fluid within the vagina and she underwent an abdomino-perineal vaginal pull through. Postoperatively, she did well and was discharged home on the 10th postoperative day. She is now 4.5 years postoperatively, doing well and has no other complaints.



Figures 8A and 8B: MRI of the abdomen and pelvis showing a markedly distend vagina arising from the pelvis and extending into the peritoneal cavity. This was secondary to vaginal atresia. Note also the distended uterus.



4. Discussion

McKusick-Kaufman syndrome was first described by McKusick in 1978 in the Amish population as a triad of hydrometrocolpos, postaxial polydactyly, and congenital heart disease. It was named after Dr. Robert L. Kaufman and Victor McKusick, who studied the condition and outlined the clinical features]1-5].

McKusick-Kaufman syndrome is caused by biallelic pathogenic variants in MKKS (20p12.2). The MKKS gene is also called the BBS6 gene. This gene provides instructions for making a protein that plays an important role in early development, specifically in the formation of the limbs, heart, and reproductive system. The protein's structure suggests that it may belong to a family of proteins called chaperonins. McKusick-Kaufman syndrome is a rare inherited genetic disorder. It is caused by mutations in the MKKS gene which is present on chromosome 20p12.2-p12.1 and it is inherited as an autosomal recessive pattern [2,5,16]. This means that both parents of an affected patient must be heterozygous carriers of the mutation gene. The MKKS gene is also called the BBS6 gene. This gene is valuable for the production of a protein that plays an important role in early development and formation of the limbs, heart, and reproductive system. McKusick-Kaufman syndrome is inherited in an autosomal recessive manner and if both parents of an individual with McKusick-Kaufman syndrome are known to be heterozygous for an MKKS pathogenic variant, each sibling has at conception a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier. Another important syndrome which must be considered when diagnosing McKusick-Kaufman syndrome is Bardet-Biedl syndrome because of the clinical overlap of both syndromes [13,14,15]. Bardet-Biedl syndrome is a generic name for a heterogeneous group of autosomal recessive disorders with at least four loci in 16q13-q22, 11q13, 3p11-p13, and 15q22 [17-20]. It is characterized by retinal dystrophy or retinitis pigmentosa, postaxial polydactyly, obesity, nephropathy, and mental disturbances, or mental retardation. It is also associated with hydrometrocolpos, usually secondary to vaginal atresia or transverse vaginal septum [18,19]. Typically, McKusick-Kaufman syndrome is diagnosed in newborns or young children, whereas the diagnosis of Bardet-Biedl syndrome is delayed to the teenage years. The retinal dystrophy or retinitis pigmentosa usually appear between 10 and 20 years of age. The overlap of both syndromes poses a diagnostic challenge. This is also important with regards to genetic counseling [20,21]. The possibility of Bardet-Biedl syndrome must be kept in mind and premature diagnosis of McKusick-Kaufman syndrome should be avoided.

McKusick-Kaufman syndrome is a rare autosomal recessive genetic disorder characterized by the triad of hydrometrocolpos, postaxial polydactyly, and congenital heart defects. In the Amish population, McKusick-Kaufman syndrome showed clinical variations as 70% of affected females have hydrometrocolpos, 60% of affected patients of both sexes have postaxial polydactyly, and 15% of affected individuals of both sexes have congenital heart disease [9]. It is important to note, many patients with hydrometrocolpos and postaxial polydactyly diagnosed as having McKusick-Kaufman syndrome have been reported at an age too young to manifest the age-dependent clinical features of Bardet-Biedl syndrome [11]. Hydrometrocolpos and polydactyly are typically present in the newborn period and because of this, the diagnosis of McKusick-Kaufman syndrome is usually made at birth. There are however clinical features of Bardet-Biedl syndrome which are not apparent in the neonatal period and are age dependent features including retinal dystrophy, obesity, and intellectual disability. This makes it difficult to differentiate between the two syndromes in the neonatal period [9]. Many authors stress the fact that the clinical diagnosis of McKusick-Kaufman syndrome should not be confirmed until the patient has reached age five years without showing the clinical features for Bardet-Biedl syndrome. Add to this the fact that, McKusick-Kaufman syndrome is relatively rare compared to Bardet-Biedl syndrome outside the Amish population.

The clinical features of McKusick-Kaufman syndrome are variable. These include hydrometrocolpos which is found in 95% of affected females, postaxial polydactyly in 95% of affected individuals and congenital heart defects in 14% of affected individuals. Other genitourinary malformations reported in affected males include cryptorchidism, chordee and hypospadias. Other digital anomalies reported include syndactyly, metacarpal and tarsal anomalies. There are other less common associated anomalies including hydronephrosis, Hirschsprung disease and anterior ectopic anus. Two of our patients had anterior ectopic anus and one of them had polydactyly and syndactyly. One of the common clinical features of McKusick-Kaufman syndrome is hydrometrocolpos. This usually presents as a large cystic abdominal mass arising from the pelvis and extending upwards. It develops secondary to dilatation of the vagina and uterus because of the accumulation of cervical secretions from maternal estrogen stimulation. These fluids accumulate because of distal vaginal obstruction secondary to vaginal atresia, a transverse vaginal membrane, or an imperforate hymen [6,10,23]. Post-axial polydactyly is characterized by the presence of additional digits on the ulnar side of the hand and the fibular side of the foot. This may be associated with syndactyly also as in one of our patients. A variety of congenital heart defects have been reported in McKusick-Kaufman syndrome including atrioventricular canal, atrial septal defect, a single atrium, ventricular septal defect, or a complex congenital heart malformation.

The diagnosis of McKusick-Kaufman syndrome is commonly made clinically. This is based on the presence of hydrometrocolpos and postaxial polydactyly with or without congenital heart disease. This is in the absence of clinical or molecular genetic findings suggestive of an alternative diagnosis. The diagnosis can be confirmed in proband by the findings of biallelic pathogenic variants in MKKS identified by molecular genetic testing. It is important to ensure that the proband does not have Bardet-Biedl syndrome, an allelic condition with considerable clinical overlap

with McKusick-Kaufman syndrome and age-dependent features including retinal dystrophy, obesity, and intellectual disability. This is the reason why others recommend waiting before confirming the diagnosis of McKusick-Kaufman syndrome until the patient has reached five years of age without fulfilling the diagnostic criteria for Bardet-Biedl syndrome, or manifesting additional findings of an alternative diagnosis. It is also important as mentioned above to exclude Bardet-Biedl syndrome which is an allelic condition with considerable clinical overlap with McKusick-Kaufman syndrome but there are age-dependent clinical features including retinal dystrophy, obesity, and intellectual disability which will manifest at an older age group [20,21].

The management of McKusick-Kaufman syndrome starts immediately after birth. These patients are born with hydrometrocolpos which can attain a large size leading to pressure effect on the urinary system, the diaphragm and gastrointestinal tract. Commonly these patients have associated hydroureters and hydronephrosis because of compression of the hydrometrocolpos and if this pressure is not relived, it will lead to damage to the kidneys. A large hydrometrocolpos may also compromise ventilation because of pressure on the diaphragm. Hydrometrocolpos if left untreated may be complicated and become secondary infected leading to Pyo-hydrometrocolpos and sepsis. This will require immediate intervention to decompress the hydrometrocolpos This can be temporarily drained percutaneously or if the patient's general condition is stable, an abdominoperineal vaginal pull through can be done as a definitive procedure [23,24]. Patients with congenital heart defects may require cardiac surgery to correct their abnormalities. The prognosis for patients with McKusick-Kaufman syndrome is variable depending on the severity of their symptoms and the presence of associated anomalies, specifically the congenital heart defects. Early diagnosis and treatment can improve the outcome. Recently, advances in gene therapies may prove valuable addressing the underlying genetic mutations that cause McKusick-Kaufman syndrome.

In conclusion, McKusick-Kaufman syndrome is a rare autosomal recessive condition characterized by postaxial polydactyly, genital malformations, and heart defects. The diagnosis is clinical and can be confirmed by genetic testing. Treatment of McKusick-Kaufman syndrome requires a multidisciplinary approach and immediate intervention to decompress the hydrometrocolpos. The prognosis varies depending on the severity of the associated congenital heart defects. Early diagnosis and intervention can improve outcomes for affected individuals.

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