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Etiology of Abdominal Masses: Mckusick Kaufman Syndrome in Two Neonates

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Abstract

McKusick-Kaufman syndrome (MKKS) is an autosomal recessive inherited genetic disorder characterized by postaxial polydactyl, congenital heart disease, and genital malformations. McKusick-Kaufman syndrome should be considered in the differential diagnosis of fetal abdominal masses. Detailed prenatal malformation screening is becoming important when a fetal abdominal cystic mass is been detected. Here, we report a case of MKKS in two female newborn presenting with abdominal masses.

Keywords: MKKS; Polydactyl; Urogenital Abnormality; Abdominal mass

1. Introduction

McKusick Kaufman syndrome (MKKS) is an autosomal recessive inherited disease, consisting of a triad of postaxial polydactyl, congenital heart disease, and hydrometrocolpos in females and genital malformations in males (most commonly hypospadias) [1] We report two cases of MKKS that considered McKusick-Kaufman syndrome in a preliminary diagnosis.

2. Case 1

A girl born vaginally at 3100 G at 40+2 gestational weeks from a mother with G1P1 admitted to a neonatal intensive care unit with a preliminary diagnosis of hydronephrosis and cystic mass in the abdominal cavity, found in the prenatal period. There was consanguineous marriage between mother and father. On physical examination, her abdomen distended, and polydactyl was present in both hands and feet. On the lower quadrant, a hard and smooth mass was palpated, which came into the hand about 6x7 cm in size. An ultrasound examination of the abdomen revealed bilateral kidneys were large and the pelvic system dilated. A cystic lesion, 8x6x8 cm in size, thick walled with dense content, with no trace of vascularity observed, extending from the left adnexal chamber to the upper left quadrant. The patient consulted in the pediatric surgery department with the current findings and underwent surgery with a preliminary diagnosis of ovarian cyst. During the operation, the patient found to have a single orifice in the genital area. Intraoperative cystourethroscopy, the urethra observed, but the vaginal opening was not been monitored. Hydrometrocholpos secondary to vaginal atresia considered and vaginostomy performed in the patient. Echocardiography in terms of abnormalities that may accompany it showed no features other than mild left ventricular hypertrophy and mild pulmonary hypertension (PHT). The eye examination showed no features, and voiding imaging resulted normally. The patient monitored by the departments of pediatric surgery, pediatric cardiology and pediatric nephrology with a preliminary diagnosis of McKusick Kaufman Syndrome. In the following period, urinary antibiotic prophylaxis started to the patient who scheduled to perform a surgical correction operation. The family were been given genetic counselling and the patient was discharged.

3. Case 2

The girl was born to a 35 year old G4P4 woman at 39 weeks' gestation by spontaneous labor; the infant's birth weight was 2850 g. The baby admitted to the neonatal intensive care unit with complaints of respiratory distress and feeding difficulty, which began immediately after birth. There was consanguineous marriage between mother and father. A physical examination of the child, who did not have a feature in his pedigree, showed a hard and smooth palpated mass of about 10x10 cm,

which caused tension in the abdomen and noticeable superficial veins, widespread distension in the lower quadrant of the abdomen. Besides, a systolic murmur of 2/6 intensity was found in the mesocardiac focus. Labium majus found to be swollen and edematous. There were six fingers on the bilateral hands and feet (Figure 1).

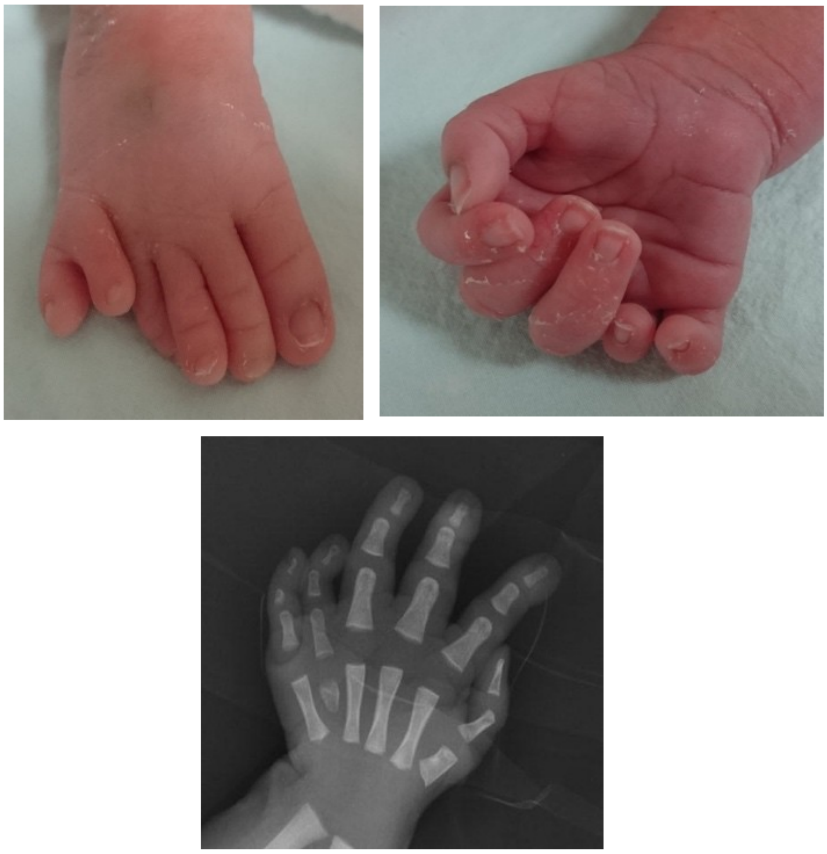

Figure 1: Postaxial polydactyly of the case

No other pathological findings found in other system examinations. A plain X-ray taken after birth showed that the diaphragm pushed up, and there was no gas shading in the lower area of the abdomen. Abdominal ultrasonography showed a minimal increase in sizes of both kidneys (right 54 mm, left 52 mm) and mild pelvic ectasia, an increase in bilateral parenchyma echogenesty at stage 2 level, as well as a properly walled, anechoic, cystic lesion of 92x97 mm in the pelvis. A CT scan of the abdomen identified a cystic lesion of 66x82 mm of possible uterine origin, extending from the lower quadrant of the abdomen to the upper quadrant (Figure 2).

In biochemical parameters, blood urea nitrogen and creatinin levels were in normal range. Her echocardiography identified the patent foramen ovale. Hy-

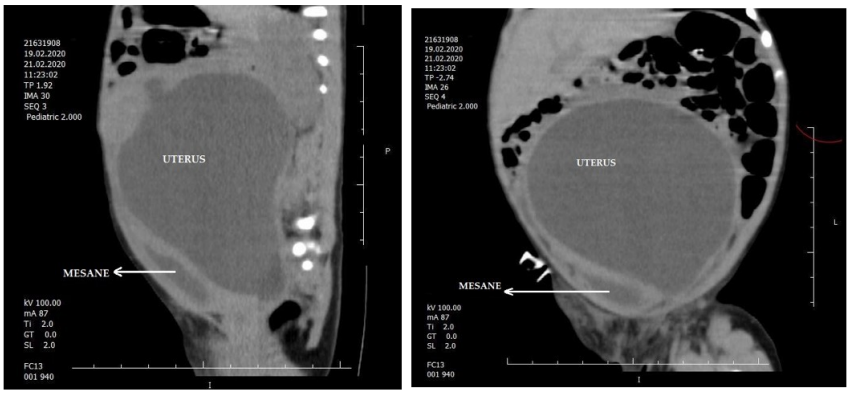

Figure 2: Radiological images of the case

drometrocholpos due to vaginal atresia observed in exploratory laparotomy performed by pediatric surgery. A transabdominal catheter inserted into the uterus for drainage purposes. The amount of fluid arriving in the drains gradually decreased. The patient, whose enteral nutrition was gradually increased, was been followed up with preliminary diagnoses of MKKS and Bardet-Biedl syndrome. Follow-up planned in terms of differential diagnosis and the patient who was scheduled to undergo surgical correction later in life was given a urinary tract prophylaxis. The family given genetic counseling and the patient discharged.

4. Discussion

McKusick-Kaufman Syndrome (MKKS) is an autosomal recessive disease characterized by postaxial polydactyly, congenital heart disease, and hydrometrocholposis in girls, and genital malformations (most commonly hypospadias and cryptorchidism) in boys [1, 2]. First described in the Amish community in 1964, the frequency of the syndrome in the Amish community is 1/10000; its frequency in other communities is unknown [3]. The gene that causes the disease, called BBS6, is located at 20p12 [1, 3]. Most patients have polydactyl (60%), congenital heart defects (15%) and urogenital abnormalities. The frequency of hydrometrocholpos has been determined as 70%. Other abnormalities such as hypoplastic lung, Hirschsprung's disease, imperforate anus, rectovaginal fistula, hypospadias, cryptorchidism, vaginal atresia, hydronephrosis, polycystic kidney may also be observed [1, 2]. Both of our cases had hydrometrocholpos and urogenital abnormalities due to vaginal atresia. In infants, hydrometrocholpos usually occurs as a large abdominal cystic mass caused by the dilation of the vagina and uterus by the accumulation of cervical secretions secondary to maternal estrogen secretion. Its etiology includes vaginal atresia, transverse membrane, or imperforate hymen, which develops due to abnormal mullerian sewage during embryogenesis. The mass can be large enough to cause intestinal obstruction, the development of hydroureteronephrosis, or diaphragm pressure [2]. In both cases, kidney expansion was present due to pressure, and respiratory distress due to diaphragm pressure was also observed in accordance with the literature. Postaxial polydactyl (PAP) is defined as the presence of additional fingers on the ulnar side of the hand and the fibular side of the foot, and can be seen in 60% of all patients [4]. In both cases, polydactyl was present in bilateral hands and feet.

Diagnosis in MKKS syndrome is based on clinical findings. Distal vaginal agenesis, hydrometrocolpos, and postaxial polydactyl in infants without a family history considered sufficient clinical evidence of this syndrome, and the diagnosis can usually made at birth [5]. Bardet Biedel, VACTERL, Ellis van Creveld and Pallister Hall Syndromes should also considered in the differential diagnosis [1]. Caution should focus on in the differential diagnosis, especially since McKusick-Kaufman Syndrome and Bardet-Biedel Syndrome show similar characteristics. MKKS syndrome is separated from Bardet Biedl Syndrome by retinitis pigmentosa, obesity, renal anomalies, hypogonadism, and lack of growth and development retardation ([5, 6]. It may be difficult to make a differential diagnosis of these two syndromes since most of these features are not visible in neonatal and early childhood. Both MKKS and BBS caused by a mutation in the MKKS or BBS6 gene contained in 2p12 [2]. No reliable phenotypic distinguishing features have found in terms of both syndromes. Despite this, abnormalities of the upper genital structure, such as the uterus, ovary and fallopian tubes, are more common in BBS [1, 7]. Since retinitis pigmentosa did not detect in our cases and other findings require time to determine the diagnosis of BBS did not consider. In the literature, a series of cases published in which he was been diagnosed with MKKS during childhood but later

understood to be BBS. Because the cardinal findings of BBS appear at a later age, researchers recommend that all patients diagnosed with McKusick-Kaufman syndrome during the neonatal period be reevaluated for Bardet Biedel Syndrome until the age of 5 [7, 8]. Cardiac findings include ventricular septal defect, atrial septal defect, vena cava settlement anomalies, atrioventricular canal defects, Arcus aorta development disorders, hypoplastic left heart, tetralogy of Fallot [8]. However, there are also cases diagnosed with MCCS whose echocardiogram is not reported any abnormality in the literature [7]. Echocardiographic imaging in our case showed mild pulmonary stenosis and pulmonary hypertension in one patient, and PFO in the other, and cardiologic follow up was recommended for both our patients. The treatment of current findings includes surgical repair of obstruction causing hydrometrocholposa, drainage of accumulated fluid, and corrective operations for polydactyl, congenital heart defects, and other abnormalities [9]. Vaginoplasty or urethroplasty should be considered for urogenital anomaly repair in later periods. In both of our patients, vaginal atresia, which creates obstruction, surgically drained, and in terms of other abnormalities, its operation planned in old age. Genetic counseling is important in MKKS, which are autosomal recessive transitive. If the mutation can identified in the index case, prenatal diagnosis or even preimplantation genetic diagnosis can made in subsequent pregnancies [4]. Although there is no data on the expected life expectancy of patients, there is no information in the literature that life expectancy reduced. Publications are available that growth, development, and fertility are normal in these patients [10]. In terms of appropriate treatment and follow-up approach in patients diagnosed with MKKS, it requires follow up and genetic counseling by the departments of pediatrics, pediatric surgery, eye, plastic and reconstructive surgery. Antibiotic prophylaxis in patients required to prevent recurrent urinary tract infections, monitoring of renal function and blood pressure, and monitoring of the development of other signs that may related to BBS are necessary. In this case, it desired to draw attention to the clinical presentation of MKKS, a rare syndrome,in the neonatal period and the approaches that should considered in its follow up.

Competing interests

The authors declare that they have no conflict of interests.

Consent for publication

All authors read and approved the manuscript.

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