Taiwanese Journal of Obstetrics & Gynecology 53 (2014) 241–244

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com

Case Report

Prenatal ultrasonography and postnatal follow-up of a case of McKusick-Kaufman syndrome

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ARTICLE INFO

Article history:
Accepted 20 July 2012

Keywords:
hydrometrocolpos
McKusick-Kaufman syndrome
polydactyly

ABSTRACT

Objective: McKusick-Kaufman syndrome (MKS) is a rare autosomal recessive syndrome characterized by hydrometrocolpos (HMC) and postaxial polydactyly (PAP). It is very difficult to diagnose MKS prenatally because of overlapping manifestations and associated anomalies with other syndromes. Herein, we present a case of MKS with prenatal ultrasound illustrating a fetal abdominal cystic mass.

Case Report: A 33-year-old woman, gravida 3 para 2, was referred to our obstetrics clinic at 34 weeks' gestation for fetal abdominal cyst detected by prenatal ultrasound. Our ultrasound illustrated a fetal abdominal cystic mass with two communicating components (suspected HMC) and polydactyly involving both hands and feet. At birth, the gross appearance revealed abdominal distention, vulva edema, and PAP. MKS was highly suspected. Abdominal computed tomography (CT) at 3 days of life showed HMC with a transverse vaginal septum. At 3 months of age, she received colpotomy and vaginal reconstruction to relieve the abdominal distension by HMC. Then she accepted corrections of PAP of both hands and feet at 8 months and 10 months. At 5 years of age, her body and mental development did not show any retardation. Pediatric ophthalmologic examination revealed no specific findings. Given the above evidences, the diagnosis of MKS was finally made at 5 years of age.

Conclusion: Rare syndromes like MKS may need early comprehensive evaluations and consultations. Although prenatal diagnosis might be impossible for MKS, prenatal awareness by fetal ultrasound is very helpful to assist early management and maternal transfer. The final diagnosis and appropriate management of MKS requires the collaboration of obstetricians, geneticists, pediatricians, and ophthalmologists as soon as abnormal signs are detected *in utero*.

Introduction

McKusick-Kaufman syndrome (MKS) is a very rare autosomal recessive syndrome characterized by hydrometrocolpos (HMC) and postaxial polydactyly (PAP). Associated anomalies of MKS include congenital heart defects (atrio-ventricular canal, atrium septum defect, ventricle septum defect, hypoplastic left heart, tetralogy of Fallot, and patent ductus arteriosus), imperforated anus, esophageal atresia, tracheoesophageal fistula, and congenital tracheal stenosis [1–8]. To date, fewer than 100 cases of MKS have been reported in the literature. MKS is more frequent in females and was first described in the Amish in 1964 [1–8]. In males, genital malformations including hypospadias, cryptorchidism, and chordee are most common. Furthermore, in females, failure of the distal third of the vagina to develop (vaginal agenesis), a transverse vaginal membrane, or an imperforate hymen may cause HMC. Therefore, HMC in MKS infants usually presents as a large cystic abdominal mass arising out of the pelvis, caused by dilatation of the vagina and uterus. As a result of the accumulation of cervical secretions from maternal estrogen stimulation, HMC develops with increasing age [1–8].

Although HMC, PAP, and congenital heart defects may be detected by prenatal ultrasound, the accuracy of prenatal ultrasound diagnosis for MKS is still unknown. Because the degree of PAP in MKS is variable and HMC may not be apparent until birth, prenatal diagnosis of MKS is rather difficult [1–8]. Herein, we present a case with a fetal abdominal cystic mass and polydactyly at 34 weeks' gestation by prenatal ultrasound. After birth, this female

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http://dx.doi.org/10.1016/j.tjog.2012.07.042

baby underwent complete examinations and surgical interventions. She received regular follow-ups at our pediatric clinic until the age of 5 years, at which time the final diagnosis of MKS was made.

Case presentation

A 33-year-old woman, gravida 3 para 2, was referred to our obstetrics clinic at 34 weeks' gestation for fetal abdominal cyst detected by prenatal ultrasound. Her obstetric history consisted of two normal pregnancies with term delivery. The parents were not consanguineous. The maternal four serum markers screening for chromosome abnormalities revealed low risk for aneuploidy. Her prenatal care was uneventful until 34 weeks' gestation when a fetal abdominal cyst was detected by ultrasound at her local clinic. She was immediately transferred to our hospital for further care. Our three-dimensional (3D) ultrasound depicted a fetal abdominal cystic mass with two communicating components and HMC was suspected (Figs. 1 and 2). In addition, polydactyly, involving both hands and feet, were illustrated. With the impressions of fetal Müllerian duct anomaly and polydactyly, several syndromes including MKS were made. After comprehensive consultations, she decided to continue the pregnancy and receive neonatal management after birth.

At 39 weeks' gestation, a female baby was born by smooth vaginal delivery, with birth weight 3448 g. Apgar scores were 7 and 10 at 1 minute and 5 minutes, respectively. Gross appearance revealed marked abdominal distention, vulva edema, and PAP. Her karyotyping was normal female (46,XX). Neonatal X-ray confirmed supernumerary metatarsal, metacarpal bones, and phalanges in both hands and feet (Figs. 3 and 4). Neonatal abdominal ultrasound demonstrated a huge cystic mass at the suprapubic area and mild hydronephrosis of bilateral kidneys. Neonatal abdominal computed tomography (CT) showed HMC with a transverse vaginal septum (Figs. 5 and 6). Initially, ultrasound-guided transabdominal aspiration of the HMC was undertaken at 6 days of life. Abdominal distention was relieved and feeding was smooth after treatment. In addition, urination was good and there was no renal dysfunction. Therefore, she was discharged from our hospital and had regular follow-ups in our outpatient pediatric clinic.

However, urinary tract infection, bilateral hydronephrosis and hydroureter, and progressive HMC were noted at 3 months of age. In order to relieve the compression effect of HMC, she underwent colpotomy and vaginal reconstruction. After the operation, the urinary tract infection and abdominal distention subsided. Follow-up abdominal ultrasound at 3 months of age showed decreased size of HMC and improved bilateral hydronephrosis and hydroureter. Regular check-ups by means of pediatric and gynecologic ultrasound examinations, together with urinalysis, blood urea nitrogen, and creatinine, were performed after discharge. Furthermore, she accepted corrections of PAP of both hands and feet at 8 and 10 months of age.

At 5 years of age, a pediatric ophthalmologist visited the girl and confirmed no abnormal findings. In addition, her body and mental

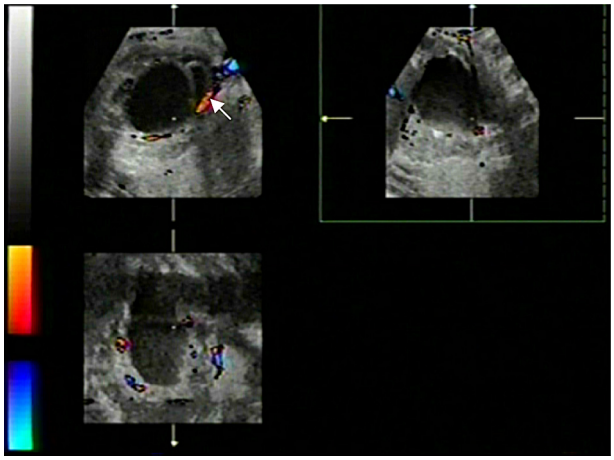

Fig. 1. Prenatal fetal three-dimensional (3D) ultrasound (multiplanar orthogonal view) showed a cystic mass behind the bladder in the fetal abdomen (arrow).

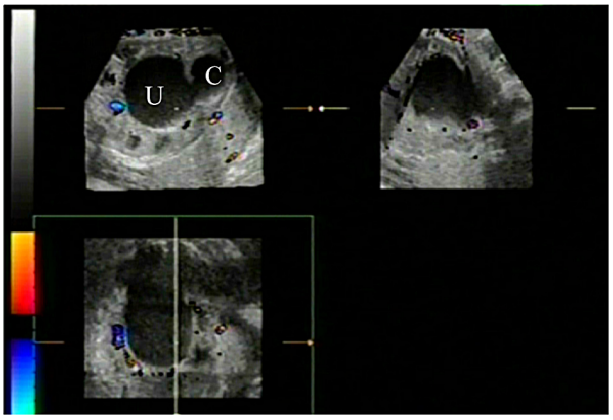

Fig. 2. Prenatal fetal three-dimensional (3D) ultrasound depicting a cystic mass with two communicating components, which was compatible with fetal hydrometrocolpos (HMC). C = hydrocolpos; U = uterus with hydrometra.

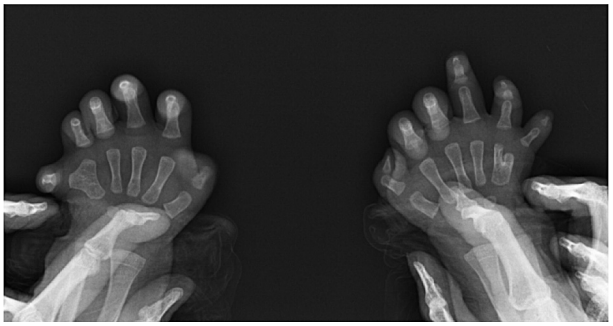

Fig. 3. Neonatal X-ray of bilateral hands with postaxial polydactyly (PAP).

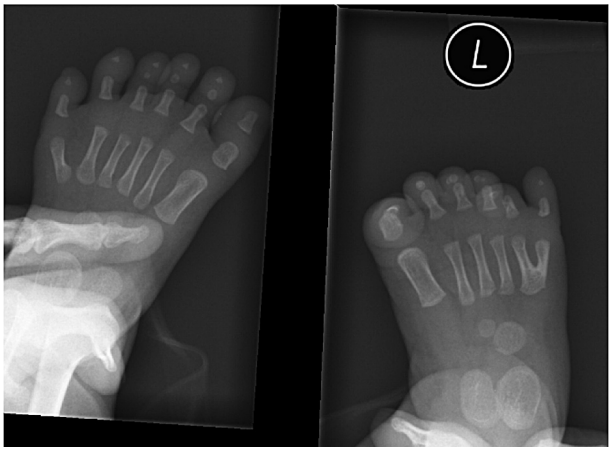

Fig. 4. Neonatal X-ray of bilateral feet with postaxial polydactyly (PAP).