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Case Report

Mckusick Kaufman Syndrome Accompanied by Congenital Laryngomalacia, Intestinal Malrotation and Hypothalamic Hamartom

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Abstract

McKusick-Kaufman syndrome is a rare syndrome inherited in an autosomal recessive pattern with a phenotypic triad comprising hydrometrocolpos, postaxial polydactyly and congenital cardiac disease. The syndrome is caused by mutations in the MKKS gene mapped onto chromosome 20p12. Diagnosis of this syndrome is based on clinical and imaging findings. In our paper, we have presented the first McKusick-Kaufman syndrome patient accompanied by congenital laryngomalacia, intestinal malrotation and hypothalamic hamartoma as well as postaxial polydactyly, hydrometrocolpos in the neonatal period.

INTRODUCTION

McKusick-Kaufman syndrome (MKS) was first described in 1964 as an autosomal recessive disorder with variable expression in an Amish population [1]. MKS has three characteristic fetures which are postaxial polydactyly, congenital heart disease and hydrometrocolpos (HMC) in females and genital malformations such as hypospadias and cryptorchidism in males [2,3]. HMC is associated with vaginal agenesis, imperforate hymen or transverse vaginal membrane. Accumulation of cervical secretions under maternal estrogen stimulus causes dilatation of the vagina and uterus and generally presents as a huge cystic mass extending to upper abdomen [3]. In the literature screening, we have recognized that our study is the first MKS case report for a patient with congenital laryngomalacia, intestinal malrotation and hypothalamic hamartoma.

CASE PRESENTATION

A female newborn was born at 39th week of gestation by caesarean section with birth-weight of 3100 gr from 36-year-old gravida 7, para 7 women. Apgar scores were 5 and 9 at 1 and 5 minutes. The parents are healthy first-degree cousins and have six healthy girls. Her prenatal history and sonograms were normal. After birth, the baby was referred to our hospital because of abdominal distension and multiple anomalies.

Physical examination revealed massive abdominal distention due to a large mass extending from the lower abdomen to the inferior margin of the liver. Basic respiration was regular. At times, however, there were episodes of inspiratory stridor. Postaxial polydactyly on both upper extremities, syndactyly of the right hand and central polydactyly-syndactyly of the 3th, 4th and 5th phalanges of the lower extremity was present. There was no vaginal opening. She was dysmorphic with depressed nasal bridge and low-set ears. There was no finding to suggest congenital heart disease. Ophthalmologic examination and echocardiography were normal.

The radiographs of hands and feet demonstrated postaxial polydactyly and syndactyly (Figure 1). Arterial blood gas analysis and electrolytes were normal. The chromosomal study was compatible with 46, XX karyotype and female phenotype. Abdominopelvic ultrasonography and magnetic resonance imaging (MRI) demonstrated a large cystic midline mass (15x10 cm) extending from the pelvic floor to the liver, which contained moderately echogenic fluid and bilateral hydronephrosis (Figure 2). In our patient presented with upper respiratory infection, we detected congenital laryngomalacia in the fiber optic

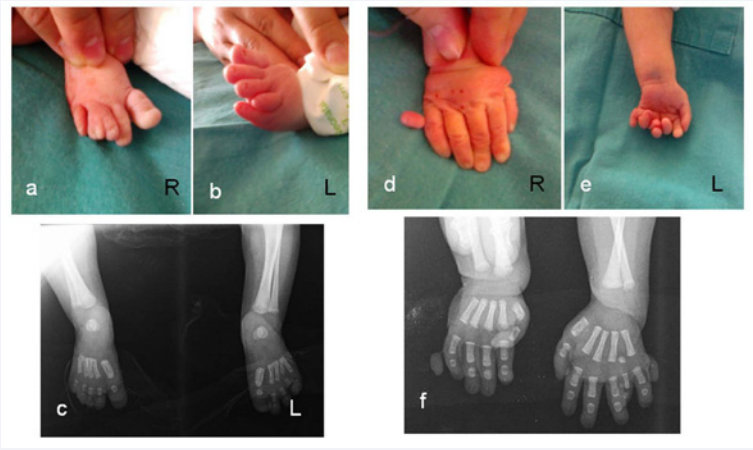

Figure 1 Central polydactyly-syndactyly on the lower extremity (a,b,c) and postaxial polydactyly on both upper extremities (d,e,f) was present.

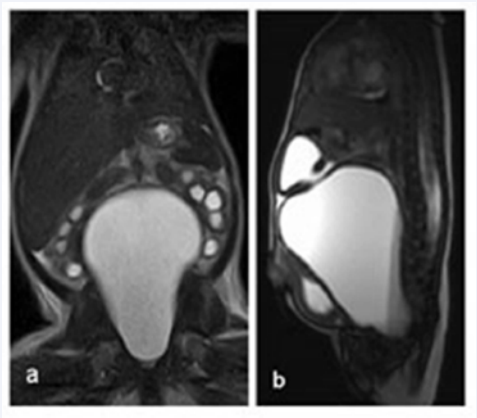

Figure 2 Coronal (a) and sagittal (b) magnetic resonance imaging shows a large cystic mass compatible with hydrometrocolpos.

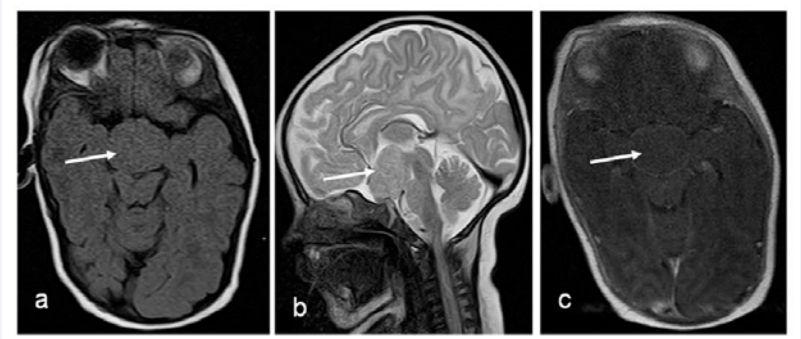

Figure 3 Brain magnetic rezonance imaging shows a hypothalamic hamartoma in the supra- and parasellar region with a homogeneous signal pattern similar to the normal brain on axial T1 (a) and sagittal T2 (b) weighted sequences, without contrast enhancement (c).

bronchoscope examination. In the patient operated by the child surgery department detected the cystic mass in 1/3 upper site of vagina and atresia in lower vagina. In addition, malrotation (cecum mobile and it was adjacent to stomach) was found. The cyst was discharged and vaginoplasty was performed.

To rule out possible congenital anomalies of central nervous system she went brain MRI showed a hypothalamic tumour 3 cm in diameter in the supra- and parasellar region with a homogeneous signal pattern similar to the normal brain on T1 and T2 pulse sequences, without contrast enhancement, presumably a hamartoma (Figure 3). Based upon these results, we considered that McKusick-Kaufman syndrome was available in the patient with congenital laryngomalacia, intestinal malrotation and hypothalamic hamartoma.

DISCUSSION

McKusick-Kaufman syndrome is characterised the postaxial

polydactyly, hydrometrocolpos and congenital heart disease [1,4]. HMC in infancy is dilatation of the vagina and uterus caused by the accumulation of cervical secretions from maternal estrogen stimulation [3]. HMC in the newborn usually presents as a large cystic mass that arises from the pelvis and contains complicated fluid [1]. The mass can be large enough to cause intestinal obstruction, hydroureter, hydronephrosis, inferior vena caval obstruction and elevation of the diaphragm in breathing difficulties.

Postaxial polydactyly, meso-axial polydactyly or syndactyly is present in 90% of cases. Developmental dysplasia of the hips and lower extremity edema are other manifestations involving the limbs [5]. Other digital anomalies described in MKS include metacarpal/tarsal anomalies, postaxial minimus, brachydactyly, absent phalanges, interstitial polydactyly and heptadactyly [3]. In our patient, postaxial polydactyly of both under extremities, syndactyly of the right hand and central polydactyly-syndactyly on the left lower extremity was present. Congenital heart disease is seen in 10-20% of cases [2,5,6]. Cardiac malformations described in patients with MKS include atrioventricular canal defect, atrial septal defect, ventricular septal defect, hypoplastic left ventricle, tetralogy of Fallot, patent ductus arteriosus, subvalvular aortic stenosis and atrioventricularis communis with a left-sided superior vena cava [2,3]. Echocardiography examination results were normal. Different from the other patients in the literature, we detected that our patient had intestinal malrotation and congenital laryngomalacia, as well. Additionaly, the other main finding in our patient was the hypothalamic hamartoma. Congenital brain neoplasms are rare, especially in the diencephalic region. Individuals with hypothalamic hamartomas may have neurologic symptoms, although most are asymptomatic. The best-described neurologic complication of hypothalamic hamartoma is gelastic epilepsy, a partial complex seizure manifest by clonic movements of the chest and diaphragm that simulate laughing [7].

HMC and postaxial polydactyly are common to both MKS and Bardet-Biedl syndrome (BBS). BBS is a genetically heterogeneous autosomal recessive condition characterized by retinitis pigmentosa, postaxial polydactyly, central obesity, learning disability, hypogonadotrophichypogonadism, complex female genitourinary malformations and renal dysfunction [3,8,9]. BBS differs from MKS in that many of its features do not become manifest until age 10 to 20 years. Slavotinek and Biesecker [2] reviewed reported cases of both syndromes presenting with HMC and postaxial polydactyly and they did not find any phenotypic features that allowed reliable differentiation between the 2 syndromes in the neonatal period, and concluded that sporadic cases in female patients cannot be diagnosed as MKS until age of 5 years [1].

Antenatal diagnosis of MKS can be established in case of HMC, polydactyly and congenital heart disease are seen all together with sonography, and is important for appropriate counseling of parents and planning of delivery conditions [1]. Prompt surgical intervention can be done to relief vaginal obstruction and the HMC. Close follow-up of patients with MKS is required because of recurrent urinary tract infections and restenosis of the vagina [1,10]. As in our patient, in cases with MKS should be kept in mind that laryngomalacia, intestinal malrotation and hypothalamic hamartoma.

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