



MCKUSICK KAUFMAN SYNDROME HEMAMALINI B

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Abstract : McKusick Kaufman syndrome is an Autosomal recessive syndrome characterized by post axial polydactyly, hydrometrocolpos and congenital heart defects. We report a case of a five months old girl child second born of non consanguineous marriage who presented with complaints of abdominal distension and decreased urine output. She had syndactyly and polydactyly of all four limbs, USG and CT abdomen showed large midline cystic lesion measuring 1086 cm, ECHO findings of common AV valve with moderate pulmonary hypertension, Laparotomy was done. Per operative findings were pyometrocolpos, vaginal atresia, persistent urogenital sinus and normal ovaries.

Keyword : Polydactyly, heart defects, pyometrocolpos, Bardet biedl syndrome, MKKS.



POLYDACTYLY

INTRODUCTION:

McKusick-Kaufman syndrome is a condition that affects the development of the limbs, heart, and reproductive system characterized by a triad of polydactyly, heart defects and genital abnormalities. This condition was first described in the Old Order Amish population, where it affects an estimated 1 in 10,000 people. The incidence of McKusick-Kaufman syndrome in non-Amish population is unknown.

CASE HISTORY:

A five months old girl baby second born of non consanguineous marriage presented with complaints of abdominal distension for 1 day duration, associated with decreased urine output. The child also had history of fever for one day. She had no loose stools or vomiting. There were no swellings elsewhere in the body. She had no history of previous hospital admission or similar illness in the past. She had an uneventful antenatal, natal and perinatal history. She was developmentally normal and immunized appropriate for age. The elder sibling was 3 years old, apparently normal.

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There was no history of similar illness in the family. On examination, she was hemodynamically stable with syndactyly and polydactyly of all four limbs. Systemic examination showed a mass palpable per abdomen measuring 11*9 cm, periumbilical in position, not moving with respiration and a systolic murmur heard over left parasternal area. Her complete hemogram, Renal function tests, liver function tests, chest x ray and urine routine examinations were normal. USG abdomen showed right kidney measuring 6.8*3.2cm and Left kidney 5*2.5cm; distended urinary bladder with segments and a large midline cystic lesion measuring 10*8*6cm suggesting cystitis with bilateral hydronephrosis and possible mesenteric cyst. CT abdomen showed bilateral ovarian cyst. ECHO showed Common AV valve with Moderate Pulmonary hypertension. Her ophthalmological examination was normal.

During course of hospital stay child developed increasing abdominal distension with signs of shock, ovarian torsion was suspected and emergency laparotomy was done. Her preoperative findings were: Distended bladder with pyometrocolpos, vaginal atresia, persistent urogenital sinus and normal ovaries. Colpocystostomy and Suprapubic catheterization was done. Patient was treated with antibiotics. Post-operative period was uneventful. She was discharged with anti-failure drugs, Suprapubic catheter and Foley's bulb in situ in the vagina.

Patient came for follow up after self-expulsion of SPC. Urine drained normally via urethral opening. Follow up USG showed distended bladder with cystitis left hydronephrosis grade 1, Right kidney was normal. Foley's bulb was in vagina [no collection]. Foley's catheter in uterus expelled spontaneously one month later. On further follow up baby had no complaints. All three orifices were present normally and child was passing urine normally through urethral opening. Systemic examination was normal.

DISCUSSION:

McKusick Kaufman syndrome affects the development of the limbs, heart and reproductive system. It is a triad characterized by polydactyly, heart defects and genital abnormalities. In females it presents with Hydrometrocolpos, Vaginal agenesis, Trans vaginal membrane. In males presentation includes hypospadias, Chordee, Cryptorchidism. The syndrome is more common in Amish population. 1-3% of

Amish in Lancaster country and Pennsylvania carry one mutation of MKKS gene on Chromosome 20p12 which is inherited as autosomal recessive. Protein predicted by MKKS is similar to chaperonin family of proteins. They protect cells from damage by reshaping abnormal proteins. Mutation of MKKS gene affects the protein processing during the development of limb, heart and genitals. The signs and symptoms of McKusick-Kaufman syndrome overlap significantly with those of another genetic disorder, Bardet-Biedl syndrome. Bardet-Biedl syndrome has several features that are not seen in McKusick-Kaufman syndrome, however. These include vision loss, delayed development, obesity, and renal failure. Because some of these features are not apparent at birth, the two conditions can be difficult to tell apart in infancy and early childhood.

Complications include Hydrometrocolpos, recurrent infections of genitourinary tract and cardiac failure. Management includes symptomatic treatment, vaginoplasty, cardiac surgery, and regular follow up. Our patient presented with the typical triad of MKKS namely, polysyndactyly, Congenital heart disease and genital abnormality in the form of vaginal atresia leading to pyometrocolpos. Though it is an uncommon entity, the case is being reported to emphasise the fact that methodical and systematic clinical examination can aid us in arriving at a diagnosis.

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