Mckusick-Kaufman Syndrome and its Anaesthetic Management

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Abstract

Mckusick-Kaufman Syndrome (MKKS) is a distinct pan-ethnic genetic entity inherited in an autosomal recessive multiple malformation syndromes. The cardinal features are polydactylyandhydrometrocolposin a female and in male glandular hypospadias, prominent scrotal raphe, cryptorchidism, congenital heart defects and polydactyly. This syndrome is diagnosed most frequently in the old order Amish population as an autosomal recessive pattern and reduced penetrance and variable expressivity. So far less than 100 cases have been reported in English literature. Very few cases on anaesthetic management of MMKS is reported from India and none from Karnataka.Thisneonate with a huge abdominopelvic mass for emergency laparotomy carry grave anaesthesia risk. This report is aimed at describing the anaesthetic management adopted for a MKKS.

Key words :hydrometrocolpos, Mckusick-Kaufman Syndrome, polydactyly.

Introduction

McKusick-Kaufman Syndrome was first described by McKusick et al in 1964 in two Amish subships and rapidly confirmed. It is often reported as 'hydrometrocolpos-polydactyly syndrome'. Hydrometrocolpos is present in 80-95% of females and results from either vaginal atresia or imperferate hymen which leads to the development of abdomino-pelvic mass with regional compression of ureters and secondary hydronephrosis. Post axial polydactyly or rarely mesoaxialpolydactyly or syndactyly is present in 90% of cases. Congenital heart defects are seen in 10-20% of cases. A locus for MKKS has recently been mapped to $20p^{12}$ close to the jagged gene [1]. The only differential diagnosis being Bardet-Biedl Syndrome. These neonates have to be evaluated carefully for congenital cardiac anomalies, cardiorespiratory, central nervous system and retinal disorders. These neonates may also present with oesophageal atresia, distal tracheoesophageal fistula, intestinal obstruction, circulatory obstruction, ano-rectal, recto-vaginal fistula, genitourinary and gastrointestinal malformation [2,6]. Hence a detailed preoperative evaluation is done for appropriate anaesthetic management to prevent morbidity and mortality.

Case Report

A 21day old female baby born out of consanguineous marriage weighing 3000 g and 45 cm long presented with h/o fever, urinary obstruction, abdominal distension, nausea and vomiting for last 7 days. On examination polydactyly of all four limbs, tachypnoea, visible veins on the distended abdomen, palpable abdominal mass extending from suprapubic region to epigastrium and vaginal atresia were noted.Airway was normal and no cardiac anomalies were detected clinically.

Baby was further evaluated with digital X-ray of abdomen, pelvis and chest, ultrasound and Computerized Tomography (CT) of abdomen and pelvis.Routine hemogram and urine analysis was done.Despite low urine output blood urea,serum creatinine and serum electrolytes were within normal limits. Sonography showed large abdomino-pelvic cystic mass with significant indentation of bladder base with bilateral moderate to gross hydronephrosis. CT study suggested 10x8x6.9cm sized cystic focal lesion with fluid debris and lesion appearing pear shaped with diagnosis of pyometra and mass effect on bladder and ureter. This baby (figure1) with hydrometrocolpos and polydactyly was diagnosed as

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Mckusick-Kaufmann Syndrome because she has two typical clinical abnormalities [3]. Baby was kept nil by mouth for 4 hours and was taken up for emergency exploratory laparotomy.

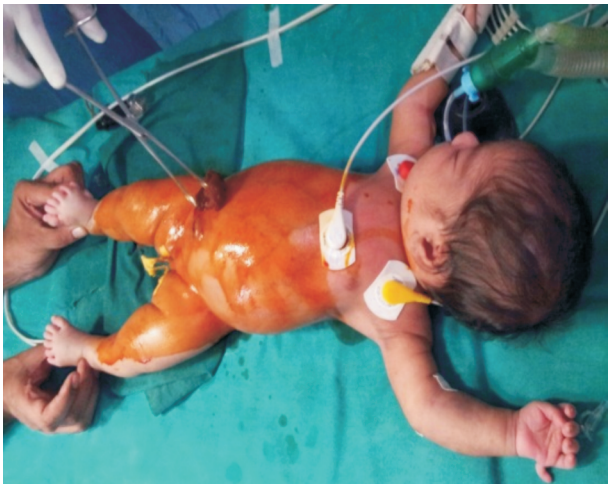

Figure1. Abdominopelvic distension and polydactyly of limbs

Anaesthetic Management

After securing intravenous line vitals were monitored by multipara monitor for ECG, SPO_2, NIBP. Stomach was decompressed with number 8 ryles tube, Inj. atropine 0.02mg/kg was given intravenously. Baby was pre-oxygenated, induced and intubated by inhalational anaesthesia with O_2, N_2O and halothane usingplain EnodotachealTube 3.0mm I.D. maintained with inj. Atracurium 0.3mg/kg body weight for skeletal muscle relaxation and tramadol hydrochloride 1mg/kg body weight was used for intraoperative analgesia.N_2O was cut-off during exploration as it would distend the intestinal coils further displacing the diaphragm and reducing the functional residual capacity.

On exploration peritonitis with varying degree of intestinal adhesions (Figure-2) seen which was released . There was gross utero vaginal distension (Figure-3) extending into the abdomen with both fimbriae filled with chocolate coloured debris. Further exploration revealed recto-vaginal fistula, which was repaired and abdomen was washed with saline and closed. Sensorcaine 2ml of 0.125% was infiltrated subcutaneously along the incisional line and suppository of Acitaminofen 30mg/kg body weight was administered for postoperative pain relief. Neuromuscular blockade was reversed with injection neostigmine 40µg/kg body weight and atropine 20µg/kg body weight. After assuring good urine output,normothermia and breathing attempts baby was extubatedand shiftedto neonatal intensive care for further monitoring of body temperature, accurate fluid management, urine output, cardio-respiratory monitoring and analgesia.

Discussion

Neonatal syndromes are associated with various congenital anomalies involving various system of the body. Because MKKS is a variable association of congenital defects, evaluation is individualised. Preanaesthetic care should include thorough airway assessment because these neonates may present defects such as tracheo-oesophageal fistula, congenital tracheal stenosis or tracheal ring hence may present with difficult intubations[4]. Careful cardiologic evaluation should be performed aiming at identifying structural heart changes for which appropriate perioperative anaesthetic technique is planned. The huge abdominopelvic mass may determine compressive symptoms such as respiratory distress and hydronephrosis and should be immediately surgically treated to prevent uterine rupture.Major anaesthetic problem faced are rapid desaturation at the time of intubation because of the large abdomino-pelvic mass pressing on the diaphragm and reducing FRC further causing ventilatory and circulatory dysfunction. Aspiration of the stomach contents is another danger because of huge hydrometrocolpos pressing on the stomach and intestines. Acute decompression of HMC leads to hypovolemia, hypotension and cardiovascular collapse. Careful monitoring of urinary function by urinary catheterisation to prevent and treat acute renal failure is advised. Prolonged exposure may lead to fluid loss and alteration in the body temperature.

Several other symptoms have been described associated with MKKS namely intestinal obstruction, circulatory obstruction, congenital heart disease, ano-rectal, recto-vaginal fistula, genitourinary and gastrointestinal malformation [5]. Hence a detailed preoperative evaluation and close intraoperative monitoring and smooth recovery from anaesthesia are the rule to prevent morbidity and mortality in MKKS.

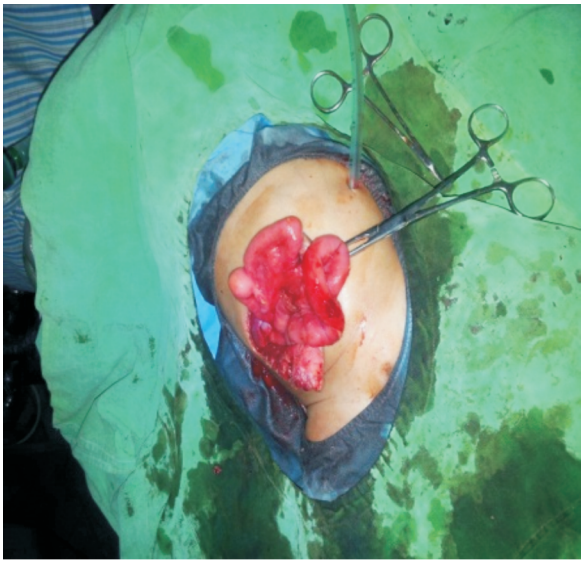

Figure 2. Intestinal adhesions

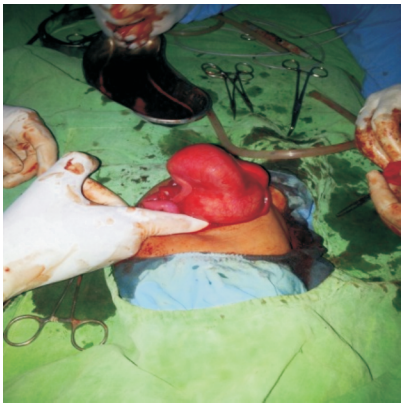

Figure 3. Uterovaginal distension

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