OBSTRUCTED UMBILICAL HERNIA IN A PATIENT OF HURLER'S SYNDROME - A CASE REPORT

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ABSTRACT

BACKGROUND

Hurler's syndrome is a type of Mucopolysaccharidoses (MPS I-H), where there is accumulation of mucopolysaccharides like dermatan sulfate and heparan sulfate, inside the lysosomes of cells, leading to its dysfunction. Clinically, it presents as child with developmental delay, short stature, hepatosplenomegaly, skeletal abnormalities, cloudy cornea, short neck, stiff joints and mental retardation. Because of various physical, mental and developmental anomalies, these children meet a premature end. One of the common presentations is the presence of inguinal and/or umbilical hernia. Obstruction of umbilical hernias in such patients is rare. We present here a case of Hurler's syndrome with obstructed umbilical hernia and its peculiar anaesthetic and surgical challenges while dealing with this case. To understand the anaesthetic and surgical challenges in cases with Hurler's syndrome.

MATERIALS AND METHODS

Primary tissue repair was performed under intravenous sedoanalgesic mixture of low-dose midazolam and ketamine with local infiltration of a mixture of lignocaine with adrenaline (1:200,000) and bupivacaine 0.25%.

CONCLUSION

Anaesthesia and surgery in patients with Hurler's syndrome can be safely performed by giving a combination of low dose intravenous sedoanalgesic and local anaesthetic with mask ventilation.

KEYWORDS

Hurler's Syndrome, Mucopolysaccharidoses, Umbilical Hernia, Primary Tissue Repair, Sedoanalgesic Mixture.

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BACKGROUND

Hurler's syndrome is a rare lysosomal storage disorder having incidence of around 1:1,00,000 births.⁽¹⁾ It is a subtype-I of mucopolysaccharidosis (MPS Type I). The molecular biology of all the various subtypes has been described.⁽²⁾ In this condition there is accumulation of mucopolysaccharides, dermatan sulfate, heparin sulfate in the lysosomes leading to cellular dysfunction. This cellular dysfunction leads to many medical problems like short stature, progressive developmental delay, obstruction of airways, cardiovascular disease, severe restriction of joints and hepatosplenomegaly. Most of the affected children die young.⁽³⁾ One of the common presentations is the development of hernias (umbilical or inguinal). In view of the many anaesthetic and surgical risks for such patient, and also due to short lifespan, non-obstructive hernias are managed conservatively. But if the patient presents with obstructive features of hernia, we have to deal with it surgically. Such presentations although rare are not uncommon.⁽⁴⁾

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We present here a case of irreducible umbilical hernia with Hurler's syndrome, who had two previous episodes of intestinal obstruction in the last one year, which resolved spontaneously. As there was a high risk of developing another episode of intestinal obstruction or a possibility of its going into strangulation, we took the case for surgery. Herein, we report our experience in dealing with the various anaesthetic and surgical challenges while managing this case.

CASE REPORT

A 6-year-old girl with known Hurler's syndrome presented to our outpatient department with Umbilical Hernia since two and a half years. Initially, it was small in size of about 2*2 cms, but gradually progressed to the present size of 15*15 cms. In the initial days of presentation it used to be reducible, but since last one year it has failed to reduce. On top of it she had two episodes of excessive crying associated with vomiting, feeling of tightening in the swelling (noticed by parents). These symptoms were relieved after symptomatic treatment by a consultant paediatrician. There is a history of consanguineous marriage of her parents. Her elder sibling is also having some of these features, but with less severity.

On examination, the child was short statured with height of 96 cms and weight 16 kg with marked developmental delay. She had many of the features of Hurler's syndrome like large head, coarseness of facial features, nasal bridge was depressed, cornea was hazy, tongue was thick, contractures in the joints causing restricted mobility, in the chest there was pectus excavatum and wide ribs (Figure 1).

There was a large umbilical hernia of around 15*15 cms, skin stretched and ulcerations over the right lower aspect of the skin over the swelling (Figure 2). Hernia was only slightly reducible and the bulge used to slightly increase in size when the child cried. The excoriated portion of the skin was dangerously thin having a high chance of formation of rent and exposure of the contents to the surroundings. This was also one of the reasons for taking the child for surgical correction. Rest of the skin over the hernia had areas of hyperpigmentation.

Laboratory investigations like Haemoglobin, Complete blood count, Random blood sugar, Blood urea, Serum creatinine, Urine examination for albumin, sugar and microscopy were normal. Liver function tests were within normal limits. Ultrasound of the abdomen showed large umbilical hernia with bowel loops and omentum as its content. The hernial defect was found to be 5 cms. Hepatomegaly was also reported in the ultrasound report. Echocardiography was normal except for slight thickening of mitral valve. There were bilateral coarse crepitations in the lungs. Therefore, she was admitted a day before and given intravenous antibiotic and nebulisation for the control of lung infection and secretion.

Patient was posted for surgery on the next day (after taking informed consent from the parents). Because endotracheal intubation many a times is fraught with danger,⁽⁵⁾ we did surgery under sedoanalgesic mixture of intravenous low-dose Midazolam 0.02 mg/kg and low-dose Ketamine 0.5 mg/kg.⁽⁶⁾⁽⁷⁾ Although, the necessity of adding adjunct atropine is a controversial issue, we opted to add atropine 0.15 mg/kg to the above intravenous mixture as a prophylaxis to decrease hypersalivation effect of ketamine,⁽⁸⁾⁽⁹⁾ because of the presence of significant secretions in the lungs of our patient.

Local infiltration of Local anaesthetic was also done with a solution of Lignocaine with adrenaline (1:200000) 5 cc with Bupivacaine 0.25% 5 cc, both were mixed and in this mixture 5 cc of distilled water was added. The solution was infiltrated into the skin, subcutaneous tissue and muscular planes around the hernia. A mixture of nitrous oxide/oxygen (50:50) was given throughout the procedure as an inhalational sedative analgesic. This 50:50 mixture is safe to use in paediatric patients.⁽¹⁰⁾

During surgery we found small intestine and omentum as contents of the sac with adhesions between the loops and between the loops and omentum and both of these contents were also adherent to the sac. All the adhesions were released meticulously by sharp dissection. The main aim of the surgery was to do a quick, less time consuming procedure to relieve the obstruction and prevent the environmental exposure of contents due to total necrosis of the covering skin with the closure of hernia defect. For this reason, primary tissue repair was preferred over Mayo's double breasting or meshplasty. Redundant skin was removed. Intraoperative period was uneventful.

Postoperatively, the patient was started on oral fluids after 3 hours and on semi-solid diet from the evening and was discharged on the second day. Patient came for regular followup visits. Sutures were removed on sixth post-operative day. There was no evidence of wound infection at the surgical site.

Figure 1. Showing Typical Features of Hurler's Facies

Figure 2. Shows Large Umbilical Hernia with Excoriated Skin

DISCUSSION

Mucopolysaccharidoses (MPS) are a heterogeneous group of rare inheritable lysosomal storage disorders, characterised by intracellular accumulation of glycosaminoglycans (GAGs) in the lysosomes, because of deficiency of various lysosomal enzymes required for the breaking down of these glycosaminoglycans molecules. This leads to progressive physical and mental deterioration. Hurler's syndrome (MPS I-H), which is a prototype of MPS is caused by a deficiency of

alpha-L-iduronidase due to a loss of function mutation leading to excessive accumulation of partially degraded GAGs, dermatan sulphate and heparan sulphate within the cells.⁽¹¹⁾

Usually, a patient of Hurler's syndrome presents with the clinical features of hepatosplenomegaly, deformity of the skeleton and stiffness of joints, clouding of cornea, large head, coarseness of facial features, broad nose with flared nostrils, large tongue, mental retardation and short physical stature.

There will also be repeated episodes of respiratory tract infection. One of the constant features in such patients is the presence of Umbilical and/or Inguinal hernia.⁽⁴⁾ In suspected cases, urinary screening tests is done and definitive diagnosis can only be established by enzyme assay.⁽¹²⁾

Selecting the type of anaesthesia for these kind of patients will be a major challenge, because of progressive involvement and functional derangement of many important organs and tissues causing anaesthetic difficulties.⁽¹³⁾⁽¹⁴⁾ Due to high prevalence of odontoid dysplasia and upper cervical vertebral subluxation, excessive flexion or extension of the neck may cause cervical spinal damage.⁽¹⁵⁾ In addition to these, thick tongue and thick palate make the endotracheal intubation a challenging task. A good knowledge of different types of intravenous, inhalational sedation and analgesia is very important in dealing with patients with mucopolysaccharidoses requiring surgery.

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