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Case Report

Anaesthetic challenges faced in a child with Morquio syndrome- An orphan disease

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ABSTRACT

Introduction: Morquio syndrome is a progressive lysosomal storage disorder leading to accumulation of glycosaminoglycans in the soft tissue, cartilage, and bone causing skeletal dysplasia.
Case Report: We present a four-year-old child diagnosed with right side inguinal hernia posted for herniotomy. He had a large head with short neck and kyphoscoliosis. Pre-operative MRI whole spine shows a hypoplastic dens causing canal stenosis, indenting the cord at C1 level. General Anesthesia was planned. With a video laryngoscope on standy by, patient was induced after stabilizing the neck with a custom neck collar and the airway was secured with a Proseal Laryngeal Mask Airway.
Discussion: The airway management can be challenging because of accumulation of mucopolysaccharides in the soft tissue of upper airway and limited mouth opening due to involvement of temporo mandibular joints.
Conclusion: A thorough pre-operative evaluation of airway must be done and a well planned anaesthetic technique must be executed.

1. Introduction

Morquio-Brailsford syndrome is a type IV Mucopolysaccharidoses (MPS) syndrome. It is an autosomal recessive disorder caused by deficiency of enzyme n- acetylgalactosamine-6-sulphate which is responsible for degradation of glycosaminoglycans. It results in the defect in the breakdown of keratan sulfate and chondroitin-6 sulfate resulting in the accumulation of mucopolysaccharides in the connective tissue and skeletal system.[1] Cartilages and cornea are majorly affected by this syndrome which is rich in keratin sulfate.[2] The office of rare diseases of National Institute of Health listed Morquios syndrome as "rare disease".[3] The incidence of this syndrome in India is unknown but the estimated prevalence of MPS in Canada, Columbia and British is about 1 per 20,000 live births and in Europe the incidence varies from 1 per 76,000 to 1 in 4,50,000 live births.[4] At birth, the patient with Morquios syndrome may appear healthy but later develop with wide spectrum of abnormality. The spectrum of this syndrome includes macrocephaly, coarse facial features, broad mouth, prognathism, anteverted nares with flat bridge, spinal deformity, macroglossia, short stature, odontoid hypoplasia, dental abnormalities, kyphoscoliosis, acoustic deafness, dwarfism, corneal opacities, pectus carnitum, hepatomegaly and cardiac abnormalities.[5] This manifestation is primarily due to mesenchymal abnormalities and the mental intelligence is usually normal.[1]

Anaesthetic challenges in such cases are due to deposition of mucopolysaccharides in the various structures leading to defects in the thoracic cage like kyphoscoliosis which may reduce the lung volume which causes ventilation-perfusion mismatch. Upper airway obstruction

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can occur due to macroglossia and excessive subglottic tissue during induction. Compression of cervical cord due to hypoplasia of dens and atlantoaxial instability can complicate the intubation process.[6] Here we present a case of 4 years old child, known Morquio syndrome posted for right side inguinal herniotomy.

2. Patient information

Four year old male child who is a known case of Morquio syndrome, born out of third-degree consanguineous marriage, was admitted for an elective right herniotomy. The child presented with history of abnormal chest shape and spine curvature since the past two years of age and not attaining the appropriate morphological features as compared with the same age group children. There is no history of breathing difficulty. The antenatal history revealed the history of pregnancy induced hypertension at the time of delivery. The post natal and development history was normal.

3. Clinical Findings

Clinical examination demonstrated the child of short stature with frontal bossing. The child presented with low set ears, short neck, gargoilism, widened wrist, chest abnormality, crowding of toes, knock kneed and short fingers and toes. Other findings in the musculoskeletal system are coarse facies, kyphoscoliosis and flat foot. The anthropometry was inappropriate with weight 12.9kg (-2z score)[7] and height 89 cm (-3z score)[7] with mid-arm circumference 14.5cm (-1 to-2z score) and head circumference 48cm (-1 to -2z score).[7] Examination of cardiovascular, respiratory and neurological systems was within normal limits. Right inguinal swelling were present.

4. Diagnostic Assessments

Pre-anaesthetic workup was done. All baseline blood investigations was done and found to be normal. 2D echo showed trivial mitral regurgitation with normal left ventricular function. Ultrasonography of abdomen and pelvis was normal. Chest X-ray revealed crowding of the ribs and wrist X-ray has visible widening. Orthopaedic opinion was obtained and radiological examination of spine suggested of lumbar lordosis and kyphosis. MRI whole spine showed prominent post dense tissue, thickening and posterior ligamentous thickening causing canal stenosis indenting the cord with subtle cord edema at C1 level, diffuse platyspondyly with anterior beaking, kypho-scoliotic deformity of thoracolumbar junction, mild indenting of cord at multiple dorsal levels more prominent at D10-D11 levels. No evidence of subluxation /dislocation of vertebrae was present. Bedside Pulmonary Function Test (PFT) was done for the child and was within normal limits. Airway examination revealed adequate mouth opening with Mallampati grade I and short neck, anticipating difficult airway.

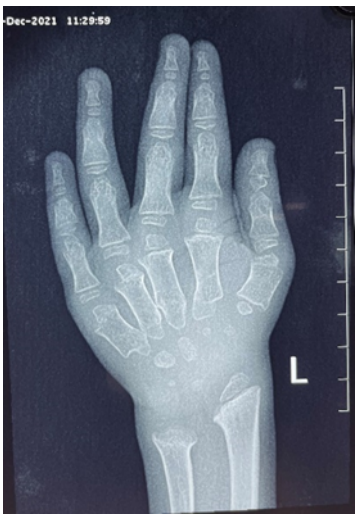

Fig. 1: X-ray of left hand Widening of wrist

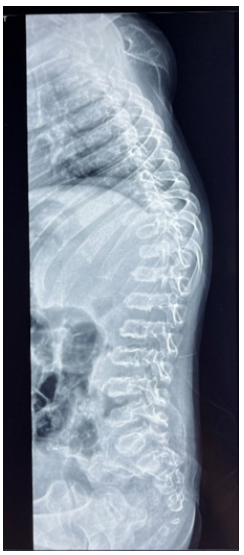

Fig. 2: Lumbo-sacral spine (Lateral view)- Kyphoscoliosis

5. Anaesthetic Management

General anaesthesia was planned after obtaining informed and written consent from the parents. Peripheral venous access was secured for the child pre-operatively. Premedication was not given. A video laryngoscope was kept on stand-by for this patient. After stabilizing the neck with the help of a custom made rigid neck collar and using manual in-line stabilisation technique, patient was induced with Inj.Propofol 2mg/kg and Inj. Fentanyl 1.5mics/kg through peripheral venous access. Airway was secured with size 2.5 Proseal Laryngeal Mask Airway (LMA).

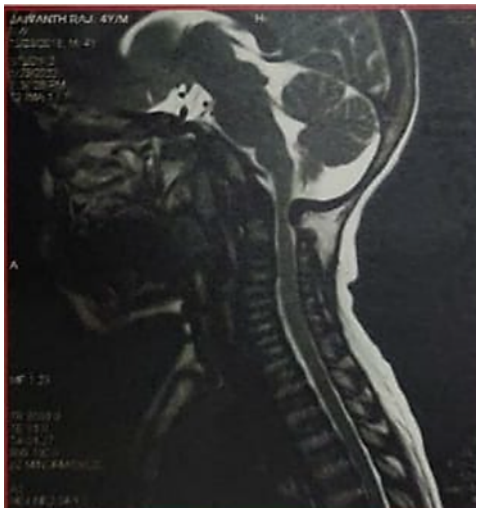

Fig. 3: MRI Spine- Hypoplastic Dens with post dens soft tissue thickening causing canal stenosis and indenting the spinal cord at C1 level

The anaesthesia was maintained with Sevoflurane 2%, oxygen and nitrous oxide in a ratio of 2L:2L. Patient was not paralyzed throughout the surgery. The surgery lasted for an hour and intra-operative period was uneventful. For post-operative analgesia, under ultrasound guidance, right side ilioinguinal nerve block was administered. Patient was extubated after complete recovery and was shifted to Post Anaesthetic Care Unit (PACU) where child was monitored. The post-operative period was uneventful as well.

Fig. 4: View of child after securing LMA

6. Discussion

Morquio syndrome is termed by Dr.Morquio in Urgquay and Brailsford in UK, in which his study among 932 patients with Mucopolysaccharidoses underwent procedure showed risk of 0.7% death /procedure and 4.2% death/patient rate

Fig. 5: Rolls are kept on either side of head to keep it in neutral position

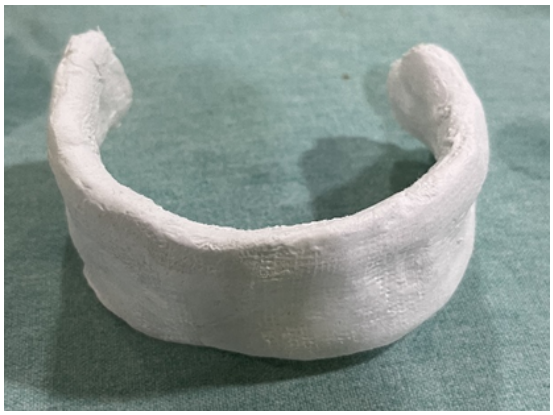

Fig. 6: Custom made cervical collar

respectively. In a recent study of Frawely et al including 17 MPS patients showed 25% difficult ventilation and 1.6% failed ventilation.[8] In planning surgery for a MPS patients it is necessary to understand the benefits against the risk of the procedure.

Despite the regular pre anaesthetic assessment, it is important to assess the anatomy of upper and lower airways and functional abnormality by doing a complete ENT examination, pulmonary function test, coronal and axial MRI or CT scan of the neck in order to rule out tracheal stenosis and glottis narrowing in patient with MPS, both of which can cause a difficult airway situation. Furthermore, this can be complicated by risk of subluxation and instability of cervical spine. Presence of Kyphoscoliosis in some patient leading to restricted lung disease which compromises the respiratory functions.[9] These patients have accumulation of mucopolysaccharides in the tissues often resulting in macroglossia, limited mouth opening due to involvement of temporo-mandibular joints and often

presents with difficulty in intubation sometimes causing a "cannot intubate, cannot ventilate" situation.

In our patient, we noticed coarse facial features like frontal bossing and kyphoscoliosis and hence anticipated difficult airway. The decision regarding the airway management can be based on radiological findings. In our patient the chest X-ray and CT neck reveals no evidence of laryngeal or tracheal stenosis. The custom-made collar made of Plaster of Paris took care of cervical spine by limiting the neck movement and stabilizing it during induction and securing airway. Induction with inhalational agent like sevoflurane provides a controlled situation and maintenance of spontaneous ventilation until laryngoscopy is performed. Sevoflurane is chosen as inhalational agent because it is well tolerated by children and non-irritant and it provides faster recovery [10] and same was chosen for our case. Some anaesthesiologists prefer the use of intravenous agents as in the management of this patient, but the use of muscle relaxants is avoided as sometimes relaxation of subglottic tissue can cause difficulty in ventilation. For securing airway, usage of LMA is a good option as there will be less manipulation of the cervical spine when compared to tracheal intubation but there is always a risk of inadequate ventilation in patient with MPS. Flexible Fiberoptic bronchoscope (if available in the centre) being used for endotracheal intubation is a highly recommended method for MPS patients. Apart from this, difficult airway cart must be ready and kept next to the patient till airway is secured.

Extubation threats another major anaesthetic complication as the child can develop laryngospasm. Extubation should be performed early after surgery before the patient is fully awake or coughing vigorously. [11]

Postoperative neurological monitoring is mandatory for MPS patients undergoing any procedure as they are at increased risk of spinal cord compression. [12] Another serious post-operative anaesthetic complication includes pulmonary compromise which indicates importance to monitor postoperative airway obstruction and respiratory failure. [11]

As there are evident risks involved in general anaesthesia, it is advised to opt for regional technique whenever possible. However, spinal anaesthesia can also be difficult to perform in patients with scoliosis.

7. Conclusion

Morquios syndrome or MPS affects multiple systems and patients should be assessed thoroughly pre-operatively. Understanding the Morquios syndrome, the anticipated anaesthetic risk can be reduced depending on the expertise, resource availability and an individual's anaesthetic plan. A complete preoperative evaluation of the airways, careful assessment of cervical spine, cardiac, respiratory and neurological functions are paramount in the anaesthetic care of such patients.

8. Source of Funding

None.

9. Conflict of Interest

No conflicts of interest declared.

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References

1. Morquio L. Sur une forme de dystrophie osseuse familiale. *Arch Med Enfants*. 1929;32:129–40.
2. Walker PP, Rose E, Williams JG. Upper airway abnormalities and tracheal problems in morquio's disease. *Thorax*. 2003;58(5):458–9.
3. Gösele S, Dithmar S, Holz FG, Völcker HE. Late diagnosis of Morquio syndrome. Clinical histopathological findings in a rare mucopolysaccharidosis. *Klin Monbl Augenheilkd*. 2000;217(2):114–7.
4. Nelson J. Incidence of the mucopolysaccharidoses in Northern Ireland. *Hum Genet*. 1997;101(3):355–8.
5. Jones KL. Morquio syndrome. In: Smith's Recognizable Patterns of Human Malformation. Elsevier Saunders; 2006. p. 538–9.
6. Morgan KA, Rehman MA, Schwartz RE. Morquio's syndrome and its anaesthetic considerations. *Paediatr Anaesth*. 2002;12(7):641–4.
7. World Health Organization. WHO AnthroPlus for personal computers Manual: Software for assessing growth of the world's children and adolescents. Geneva: WHO; 2009.
8. Arn P, Wraith JE, Underhill L. Characterization of Surgical Procedures in Patients with Mucopolysaccharidosis Type I: Findings from the MPS I Registry. *J Pediatrics*. 2009;154(6):859–64.
9. Gösele S, Dithmar S, Holz FG, Völcker HE. Late diagnosis of Morquio syndrome. Clinical histopathological findings in a rare mucopolysaccharidosis. *Klin Monbl Augenheilkd*. 2000;217(2):114–7.
10. Bartz HJ, Wiesner L, Wappler F. Anaesthetic management of patients with mucopolysaccharidosis IV presenting for major orthopaedic surgery. *Acta Anaesthesiol Scand*. 1999;43(6):679–83.
11. Frawley G, Fuenzalida D, Donath S, Yaplito-Lee J, Peters H. A retrospective audit of anesthetic techniques and complications in children with mucopolysaccharidoses . *Paediatr Anaesth*. 2012;22(8):737–44.
12. Davison JE, Kearney S, Horton J, Foster K, Peet AC, Hendriksz CJ. Intellectual and neurological functioning in Morquio syndrome (MPS IVa). *J Inherit Metab Dis*. 2013;36(2):323–8.

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