Annals of Medical and Clinical Case Reports
Volume 1| Case Series

Navigating the Maze: Anesthetic Management of Mucopolysaccharidosis - A Case report

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Citation: Adethen Gunasekaran, Navigating the Maze: Anesthetic Management of Mucopolysaccharidosis – Case series., Ann Med Clin Case Rep, 2025; 1(7): 1-4.

Keywords: Airway Management,Mucopolysaccharidosis, Pediatric Anesthesia, Sanfilipo, Morquio

Published Date: 31-08-2025 Accepted Date: 28-08-2025 Received Date: 23-08-2025

Abstract

Background

MPS are genetic disorders characterized by the accumulation of glycosaminoglycans, leading to multi-system involvement and increased anesthetic risk. The primary concern for anesthesiologists in these patients is airway management due to features like thickened soft tissues, enlarged tongue, short neck, retrognathia, and immobile cervical joints. This case series details the anesthetic management and challenges encountered in three cases of Mucopolysaccharidosis (MPS) Type III (Sanfilippo syndrome) and Type IV (Morquio syndrome). Specific challenges highlighted are difficult intubation due to upper and lower airway obstruction, excessive secretions, and skeletal abnormalities. The importance of meticulous airway planning, availability of advanced airway devices, and consideration of altered drug metabolism due to end-organ dysfunction are stressed. The series concludes by underscoring the necessity of recognizing the specific MPS type for tailored anesthetic planning and experienced decision-making to ensure successful outcomes in these complex patients.

1. Introduction

Mucopolysaccharidosis (MPS) is a group of genetic disorders characterised by the accumulation of Glycosaminoglycans (GAGs) due to lysosomal enzyme deficiencies. This accumulation can lead to autophagy, mitochondrial dysfunction, and cell death, resulting in a multi-system disorder with varying symptoms. MPS encompasses 7 types (excluding types 5 and 7) with multiple subtypes. This case report focuses on the anaesthetic management and challenges encountered in two different MPS types (Type III and IV) across three cases. As anesthesiologists, our primary concern is the airway, which can present challenges due to thickened soft tissues, an enlarged tongue, and immobile cervical joints. This series highlights the strategies and techniques employed for successful anaesthetic administration.

2. Clinical description

2.1. Case 1

An 11-year-old male with Mucopolysaccharidosis Type III (Sanfilippo syndrome) and attention deficit hyperactivity disorder (ADHD) presented with an obstructed inguinal hernia. His airway examination revealed a large head, coarse facies, a short neck, and retrognathia. (Figure 1). Patient was posted for inguinal herniotomy. Left inguinal exploration was performed uneventfully. A USG-guided Transverse Abdominis Plane block was administered post-procedure. The child was extubated on the table with a difficult airway cart readily available. He was discharged from the PACU to the ward and advised to follow up.

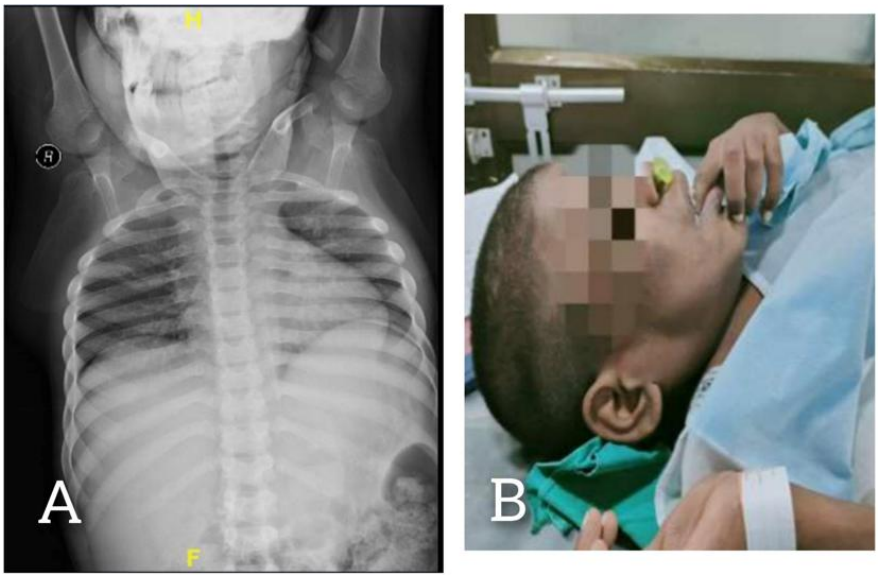

Figure 1: 1A X-ray showing cardiomegaly in Sanfilipo syndrome. 1B- Image showing coarse facies, short neck and retrognathia in Sanfilippo syndrome

2.2. Case 1

A 3.5-year-old male, a known case of MPS type IV (Morquio syndrome), presented with an irreducible right groin and umbilical swelling, pain, and constipation. Echocardiography suggested a retroaortic left circumflex artery. Airway evaluation showed retrognathia and a short neck. Patient was posted for inguinal and umbilical hernia repair. General anesthesia and regional anesthesia (caudal block) were given. Anaesthesia was maintained with an air and oxygen mixture (FiO2 0.4-0.5) and Sevoflurane (up to MAC 1.2). A 0.25% Bupivacaine caudal block was used for postoperative analgesia. The intraoperative course was uneventful. The child was extubated with stable vitals and transferred to the PACU for monitoring.

2.3. Case 1

A 6-year-old female diagnosed with Morquio syndrome (MPS Type IV) presented with wrist widening, bowing of legs, and short stature noted at age 3. She had a history of recurrent upper respiratory tract infections. MRI of the brain revealed narrowing of the cervical spinal cord and platyspondyly. Airway evaluation indicated retrognathia and a short neck. She was scheduled for C1 posterior arch excision in the prone position. After general anaesthesia, maintenance was done with Propofol at 100-200 mcg/kg/hr for neuromonitoring, supplemented with Fentanyl and paracetamol suppositories for analgesia. Due to anticipated airway and spinal cord oedema, the child was electively mechanically ventilated and extubated the following day with complete sensorimotor recovery.

All the details regarding demographic management and case management had been described in (Table 1).

Table 1: Case details and management of all 3 cases

S no	Syndrome	Type of MPS	Diagnosis	Positive findings	Surgical done	Anesthesia plan	Videolaryngoscope
1	Sanfilippo syndrome	Type 3	Obstructed inguinal hernia	Large head, coarse facies, short neck, Retrognathia	Herniotomy	GA+ TAP Block	MAC #3 Tuoren
2	Morquio Syndrome	Type 4	Umbilical hernia & inguinal hernia	Retrognathia , short neck	Hernia repair.	GA+ Caudal block	MAC #1 BESDATA®BD -DF
3	Morquio syndrome	Type 4	C1 spinal cord compression	Retrognathia, Short neck, C1 spinal cord compression	C1 posterior arch excision	GA with i,v analgesics.	C-MAC #2 (Karl Storz)

3. Discussion

Mucopolysaccharidoses are a group of storage disorders caused by deficiencies in enzymes responsible for GAG degradation. This accumulation leads to various enzymatic defects and biochemical alterations, resulting in a broad spectrum of clinical presentations

including hepatosplenomegaly, mental retardation, corneal clouding, coarse facial features, and multi-organ dysfunction. Due to the involvement of multiple systems (airway, respiratory, cardiovascular, and central nervous system), these children face increased anaesthetic risk from both regional and general anaesthesia. This heightened risk is attributed to airway obstruction, excessive airway secretions, frequent respiratory tract infections (complicating general anaesthesia), and skeletal abnormalities like restricted cervical spine movement and kyphoscoliosis (hindering regional anaesthesia). These issues necessitate a carefully planned anaesthetic approach.

The incidence of all genetic diseases is less than 0.1% [1,2]. MPS has 7 types (1-9, excluding 5 and 7) with varying prevalence based on region and ethnicity [3]. Clinical symptoms depend on the deficient enzyme. Overall, common features include respiratory issues, musculoskeletal abnormalities, and other features ranging from facial dysmorphisms to coagulation abnormalities like thrombocytopenia and clotting factor deficiencies. Early diagnosis and treatment, through hematopoietic stem cell transplantation or enzyme replacement therapy, are crucial. The deposition of GAGs in various anatomical structures such as the airway, liver, and spleen affects normal organ function. As anesthesiologists, these pathophysiological changes are a significant concern as they impact induction, intubation, and drug metabolism.

Given that these patients undergo a variety of surgeries, from simple elective procedures like hernia repair to complex procedures like kyphoscoliosis correction, anaesthetic management presents significant challenges due to the multi-organ involvement and altered anatomy and physiology.

Our first case, Sanfilippo syndrome (MPS type III), is a neurodegenerative disorder with worsening behavioural disturbances due to heparan sulfate accumulation. These children often present with facial dysmorphisms, hyperkinesia, aggression, attention deficit, dysphagia, and swallowing difficulties [4]. A major challenge for anesthesiologists in MPS patients is difficult intubation due to both upper and lower airway involvement. Upper airway obstruction is caused by excessive secretions, macroglossia, cervical spine involvement, and sometimes vocal cord thickening. Lower airway obstruction can result from malformed tracheal cartilage and airway oedema [4]. Avoiding neck extension and flexion is crucial due to potential odontoid dysplasia, which can complicate airway management. Therefore, meticulous airway planning is mandatory, and a difficult airway cart with videolaryngoscopes and fiberoptic bronchoscopes should always be ready. In our case, airway difficulty was due to a short neck and retrognathia [5]. Rapid sequence induction was employed due to the risk of obstruction and aspiration. Given the increased risk of post-obstructive pulmonary oedema in these patients, emergency re-intubation and tracheostomy kits were kept readily available [6].

The second and third cases involved Morquio syndrome (MPS Type IV). This type is characterised by defective degradation of keratan sulfate, leading to end-organ dysfunction and anatomical distortions. Most reported individuals with Morquio syndrome are anticipated to have a difficult airway due to anatomical distortion of the upper airway, including temporomandibular joint involvement, a short neck, retrognathia, cervical spine involvement, and tracheal narrowing, often with combined tracheal and thoracic inlet narrowing, resulting in a difficult airway and positioning during laryngoscopy [5,7]. End-organ dysfunction leads to altered metabolism and excretion of drugs and their metabolites, complicating drug dosage. Thick tracheobronchial secretions also affect perioperative care, potentially leading to recurrent respiratory tract infections and pneumonia [8].

4.Conclusion

Mucopolysaccharidoses present a spectrum of anaesthetic challenges, ranging from mild systemic disease features like short stature, sleep disturbances, cognitive dysfunction, and delayed milestones, to severe complications such as cervical spine abnormalities, scoliosis, cardiac anomalies, and a difficult airway. Recognising the specific MPS type is crucial for understanding the basic pathophysiology and identifying potential risk factors for better anaesthetic planning. Anaesthesia planning and management should incorporate experienced decision-making, including the use of a full array of advanced airway devices.

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