

## CASE REPORT

# Awake airway endoscopy in mucopolysaccharidosis: a case report



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### KEYWORDS

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**Abstract** Mucopolysaccharidosis (MPS) are a group of rare genetic inherited diseases with a progressive course due to the accumulation of glycosaminoglycans resulting in anatomic abnormalities and organ dysfunction, including the respiratory, cardiovascular, skeletal, and neurological systems that can increase the risk of anesthesia complications. Clinical manifestations are variable, multisystemic, and include severe morphological changes. The anesthetic management of these patients is complex, particularly airway management, which can be planned to include a fiberoptic airway investigation prior to surgery. We present two cases of patients with MPS type VI and VII who underwent fiberoptic airway mapping under conscious sedation, with no complications. Since MPS is a rare but challenging disease concerning the airway management, we propose a safe and effective anesthetic technique that could be used for fiberoptic bronchoscopy and allow fiberoptic-assisted tracheal intubation at the time of surgery.

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## Introduction

Mucopolysaccharidosis (MPS) are inherited lysosomal storage diseases caused by the deficiency of enzymes required for the stepwise breakdown of Glycosaminoglycans (GAGs),

being associated with the widespread tissue accumulation of partially degraded GAGs. The characteristic patterns and age of presentation form the basis of MPS classification into seven types.

Typical clinical manifestations include coarse facial features, ear-nose-throat problems, skeletal dysplasia, growth impairment, cervical instability, organomegaly, impaired vision and hearing, joint contractures, hernias, and cardiorespiratory disease.<sup>1</sup> Respiratory complications affect all

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types of MPS and contribute to death and disability. Airway obstruction result from respiratory abnormalities, excessive secretions, skeletal restriction, organomegaly, infections, and neurologic compromise.<sup>2</sup>

Patients with MPS I, II, VI, and VII often develop upper airway obstruction due to an enlarged tongue, thickened gums, and engorged soft tissues of the nasopharynx.<sup>2</sup> Airway problems may be worsened by excessive thick secretions due to recurrent infections.

Most MPS patients require anesthesia for multiple interventions to help manage the disease and complications from anesthesia are commonly related to the airways. Abnormal airway anatomy associated with the above-mentioned risk for airway obstruction can result in extreme difficulty in tracheal intubation.<sup>3</sup> Older patients and sleep apnea are at increased risk of difficult intubation. Anesthesia should be administered at centers experienced in the management of MPS patients. A comprehensive plan should be drawn up before performing general anesthesia.

The exact incidence of MPS VI and VII is unknown, although MPS VI is estimated to occur in 1 of 250,000 to 600,000 newborns,<sup>4</sup> and MPS is estimated to occur in 1 of 250,000 newborns, being one of the rarest types of mucopolysaccharidosis.<sup>5</sup>

In this report, we would like to share two cases of patients with MPS type VI and VII listed to corneal transplant and aortic prosthesis revision surgery, respectively. They underwent preoperative fiberoptic airway mapping. Informed consent for publication was obtained from the patient and his parents.

## Case report

A 23-year-old man (40 kg, 116 cm) diagnosed with MPS type VI Maroteaux-Lamy syndrome (Patient 1, Fig. 1a), and a 28-year-old female (37 kg, 134 cm) with MPS type VII Sly syndrome (Patient 2, Fig. 1b) were listed for a corneal transplant and aortic valve prosthesis revision surgery under general anesthesia, respectively.

Patient 1 was diagnosed with MPS at 21 months of age and had a medical history of heart failure (NYHA class III), mitral and aortic valve disease, restrictive lung disease, sleep apnea (CPAP device during the night), and stable cervical spinal stenosis. Physical examination revealed macrocephaly, hearing loss (conduction type), saddle nose, macroglossia, gingival hyperplasia, prominent costal margin, short limbs, and clawed hands. Cognition was not affected. He was submitted to enzyme replacement therapy with galsulfase (once a week) from the age of 9, with improvement of respiratory infection and quality of life. Past surgeries included myringotomy, and tonsillectomy and adenoidectomy, with a difficult airway requiring multiple intubation attempts.

Patient 2 was diagnosed with MPS at 16 years of age and had a medical history of heart failure (NYHA III), and developmental delay and progressive intellectual disability. Physical examination showed macrocephaly, saddle nose, irregular shaped teeth, macroglossia, short neck, and hepatosplenomegaly. Past surgeries included adenoidectomy with myringotomy, glossectomy reduction surgery, aortic valve replacement, unilateral total hip replacement, and

cervical decompression surgery, also with a difficult airway requiring multiple intubation attempts, and one postponed surgery.

Given the past difficult airway management, both were proposed for fiberoptic airway mapping, with the aim of predicting further airway complications at the time of surgery, as per our institution's protocol for managing MPS patients.

The patients were premedicated with midazolam intravenous (0.1 mg.kg<sup>-1</sup>), positioned (seated) and monitored with a pulse oximeter, electrocardiogram, noninvasive blood pressure, and Bispectral Index Monitor (BIS). Intranasal phenylephrine (2.5 mg ×2) and topical (1 mL ×2) and nebulized lidocaine 2% (8 mL at 5 L.min<sup>-1</sup>) were applied to the airway (nasopharynx and oropharynx) twenty minutes prior to the procedure. Antisialagogues were not prescribed. Pre-oxygenation was achieved with 100% O<sub>2</sub> via an endoscopy mask (an explorer endoscopy face mask with three one-way valve). A remifentanil Target-Controlled Infusion (TCI) targeting a predicted plasma-site concentration (Minto model) of 1.0 ng.mL<sup>-1</sup> was started. The fiberscope was nasally introduced, with lidocaine 2% applied according to the spray-as-you-go technique. Remifentanil TCI targeting 3–5 ng.mL<sup>-1</sup> reduced the airway reactivity, with a BIS objective of 75–80.

The procedures lasted for 15 minutes, and the patients remained awake, calm, and cooperative, with spontaneous ventilation and no sign of breathing difficulty, oxygen saturation of 96–99% with FiO<sub>2</sub> 100%. There were no airway obstruction or desaturation episodes during both procedures. They remained hemodynamically stable with mean arterial pressure in the range of 80–95 mmHg and a heart rate of 88–110 beats per minute. The airway images of each patient are presented in Figure 1.

After the procedure was finished, remifentanil infusion was stopped, and the patients were transferred to the recovery area fully conscious and with adequate breathing. Both patients were discharged home, with no complication.

## Discussion

This report presents the anesthetic management of two adult patients with a rare genetic condition with major structural abnormalities of the upper airway in whom awake fiberoptic airway mapping was performed due to the high risk of difficult intubation and ventilation.

The life expectancy of patients with MPS continues to increase due to improvement in therapy. MPS patients have multiple comorbidities, many of which require surgical interventions. Very little literature is available about anesthesia in adult patients with this condition.<sup>1–3</sup> Besides that, the process of aging can be associated with severe narrowing of the larynx and trachea.

Due to MPS being associated with specific phenotypic facial and airway characteristics which have an impact on ventilation, substantial challenges for perioperative anesthetic management may be expected. There is an increased anesthetic risk due to a difficult airway, cervical spine disease, and a higher prevalence of cardiovascular manifestations.<sup>2</sup> Restrictive or obstructive lung disease,



**Figure 1** (a) Patient 1 (b) Patient 2. (c) Patient 1: Fiberoptic bronchoscopy demonstrating tracheal rings, carina and subglottic region. (d) Patient 2: Fiberoptic bronchoscopy demonstrating vocal folds and subglottic region.

recurrent lung infections, and obstructive sleep apnea are also common.

It has been reported that 25–50% of MPS patients have problematic airways and 82% of MPS patients receiving anesthesia require urgent airway interventions.<sup>1</sup>

Various airway issues may impact intubation and ventilation of these patients. Supraglottic abnormalities are common due to cranial and spinal deformations and GAG deposition in the mouth, nose, and throat. These include flattened nasal bridge, maxillary hypoplasia, impaired opening of the mouth, high-arched palate, macroglossia, gingival hyperplasia, mucosal oedema, mucoid secretions, narrow hypopharynx, short neck, abnormal cervical vertebrae, and high epiglottis. Excessive tissue at arytenoid cartilages and aryepiglottic can cause stridor and airway compromise in extreme cases. Infraglottic airway abnormalities include tracheobronchomalacia due to GAG deposition in the tracheobronchial cartilage and tracheal collapse due to decreased tracheal traction from decreased lung volume.<sup>6,7</sup>

In the two cases presented in this report, awake fiberoptic airway mapping under local anesthesia plus conscious sedation with low-dose midazolam and plasma-effect TCI remifentanyl infusion was undertaken without airway, respiratory or cardiovascular complications in high-risk patients with a known history of difficult airways.

The authors suggest that this technique is safe and effective and could be used for fiberoptic bronchoscopy and allow fiberoptic-assisted tracheal intubation at the time of surgery. No major problems were found and based on this experience, the authors suggest that this technique is safe and effective and could be used for fiberoptic bronchoscopy, allowing fiberoptic-assisted tracheal intubation at the time of surgery. Careful planning and experienced support for difficult airway management are important when anesthetizing such patients.

In MPS patients, the high prevalence of perioperative complications and critical problems related to anesthesia (difficult intubation and airway control) underlies the critical role of a multidisciplinary careful evaluation before the procedure, namely evaluation of the airways. The risk of difficult intubation must always be suspected and requires experienced, expert staff, and the use of advanced airway management.

### Implication statement

In mucopolysaccharidosis patients, the most critical problems related to anesthesia are difficult intubation and airway control. Therefore, careful evaluation of anesthetic risk factors should be made before the procedure, namely evaluation of airways.

### Conflicts of interest

The authors declare no conflicts of interest.

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