Case Report

Navigating Airway Management in Adult Hurler's Syndrome: A Clinical Case Report

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Abstract

Hurler syndrome, also known as mucopolysaccharidosis type I (MPS I), is a rare lysosomal storage illness that involves many systems and is characterised by the tissue accumulation of glycosaminoglycan. Although it usually manifests in children, adult instances are less widely known. We describe a case report of an adult patient with Hurler syndrome. The patient presented with short stature, obesity, and a challenging airway with macroglossia, short neck, and restricted cervical mobility. A steeple sign was noted on X-ray, indicating possible airway narrowing. Furthermore, the diagnosis of empty sella syndrome added complexity to the case and impacted the anaesthetic management strategy. This case underscores the critical need for comprehensive preoperative evaluation, careful intraoperative monitoring, and strategic airway management planning in patients with Hurler syndrome to ensure safe and effective anaesthetic care.

Key words

Adult Hurler, Genetic Disorder, Difficult Airway, Multisystem Involvement.

Introduction

Hurler's syndrome, also known as type 1 mucopolysaccharidosis (MPS I-H), is a rare hereditary disorder caused by a deficiency of the lysosomal enzyme α-L-iduronidase. This enzyme is crucial for the proper metabolism of glycosaminoglycans, leading to their progressive accumulation in various organs.

The condition is characterized by significant craniofacial, skeletal, and cardiac involvement, which heightens the risks associated with anaesthesia. Patients with MPS I-H typically present with various somatic issues and progressive cognitive decline. Common skeletal deformities include short stature, distinctive facial features, reduced joint mobility, growth retardation during childhood, and cervical spine instability.[1]

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These symptoms collectively manifest as dysostosis multiplex, which is characterized by a short, rigid neck among other findings. Additional complications may include recurrent respiratory infections, various hernias, communicating hydrocephalus, spinal cord compression, corneal clouding, hearing loss, cardiovascular issues (such as cardiomyopathy and valvular dysfunction), restrictive lung disease, and organomegaly. Soft tissue hypertrophy can lead to anatomical changes, such as a narrowed nasal passage, adenotonsillar hypertrophy, and thickening of laryngeal and pharyngeal structures, resulting in airway obstruction at multiple levels.[2, 3]

Patients often experience severe obstructive sleep apnea (OSA). Other associated findings can include a stiff temporomandibular joint, limited mouth opening, and odontoid dysplasia. Informed and written consent was taken from patient and guardians in accordance with the case report.

Case Report

A 37-year-old male with Hurler's syndrome presented for surgical intervention for acute corneal hydrops, see Figure 1,2.

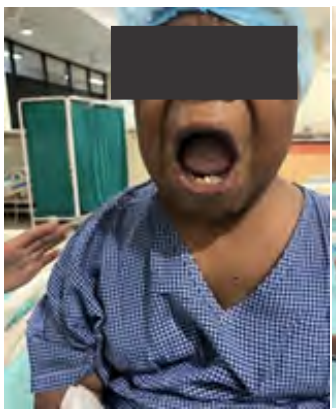

Figure 1

Figure 2

The patient had a notable medical history including short stature, intellectual disability, juvenile hypothyroidism, features suggestive of partial empty sella syndrome, and dyspnoea on exertion. The collective assessment of clinical symptoms led to the confirmation of the diagnosis by the concerned multidisciplinary evaluation involving genetics, neurology, paediatrics, medicine, and endocrinology. At present, the patient was not receiving enzyme replacement therapy (ERT). It was not initiated due to reduced expected benefit at this advanced age with established multisystem involvement. Management is therefore mainly supportive and targeted toward organ-specific issues.

The preoperative physical examination revealed a distinctive individual with short stature, weighing 40 kg and standing 115 cms tall. The patient also had grade I obesity, having a BMI of 30.1 kg/m.^{2} The patient's appearance was notably inconsistent with his chronological age, suggesting a discrepancy between physical development and age. The patient had macroglossia classified as Friedmann grade 3, with a mouth opening of more than two fingers. Additionally, he had a short, bulky neck.

Systemic examination and vitals were normal. Laboratory investigations such as hemogram, urine routine and liver function tests were within normal limits. Endocrine evaluation revealed hypothyroidism, with a significantly elevated TSH level of 25mIU per litres. The patient was started on Thyronorm 100 mcg, resulting in a decreasing trend in TSH levels.

Chest X-ray showed kyphoscoliosis, cardiomegaly, broadened anterior portion of ribs and slight tracheal narrowing in the lateral view, revealing a steeple sign, see Figure 3.

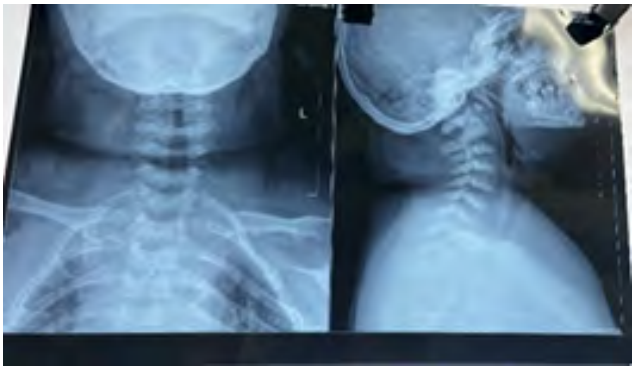

Figure 3

An ENT consultation was taken, and their evaluation suggested the likelihood of difficult intubation, a potential need for tracheostomy, and challenges with extubation. 2D echocardiogram showed concentric left ventricular hypertrophy (LVH) with mild pericardial effusion. MRI brain indicated empty sella syndrome and flattened cervical vertebrae. Although empty sella was noted on imaging, the patient remained clinically asymptomatic. His endocrine evaluation-including blood sugars and pituitary hormone profile-was within normal limits, and he has not required any steroid or hormone replacement therapy.

Given these parameters, significant anaesthetic concerns arose, particularly regarding airway management. A comprehensive plan for a potentially difficult airway was developed in collaboration with the ENT team, including the option for emergency tracheostomy.

Plan A: Utilize spontaneous ventilation with a volatile agent and intravenous agents. Avoid neuromuscular blockers until the airway is secured.

Plan B: Employ a laryngeal mask (LM) either as a definitive airway or as a conduit for fiberoptic bronchoscopic (FOB) intubation or use of C-MAC blade video laryngoscope for intubation.

Plan C: Prepare for emergency tracheostomy with ENT backup.

Management

After thorough discussions with the ophthalmology team regarding their procedural requirements, the patient was transferred to the operating theatre. Preoperative sedation was withheld. Based on the physical examination, a difficult intubation was anticipated. As a precaution, a difficult airway cart was prepared and kept ready in the operating room. Upon the patient's arrival, a 20-gauge IV line was secured, and basic monitors were applied 15 minutes prior to induction. Anaesthesia was induced with fentanyl 40 mcg and propofol 40 mg IV, titrated to maintain spontaneous respiration following preoxygenation with 100% oxygen. An oral Guedel airway size 2 was inserted to confirm mask ventilation. Given the short and minimally painful nature of the procedure, we chose to maintain spontaneous ventilation using face mask with administration of intravenous propofol, fentanyl, and ketamine, along with low dose of inhalational agent sevoflurane (1-3%) combined with oxygen and nitrous oxide.

The adequacy of anaesthesia was confirmed using standard monitoring techniques, ensuring that the patient showed no response to pain or surgical manipulation. The surgical team then commenced the double bubble pneumo descemetopexy procedure. Before the crucial step of injecting the air bubble in the eye, intravenous fentanyl was administered to alleviate potential pain or discomfort. The surgery lasted for 15 minutes and the patient was transferred to the post-anaesthesia recovery unit after confirming that he was fully awake and had regular, adequate and spontaneous respiration.

Discussion

Hurler's syndrome often presents with several challenges, including abnormal upper airway anatomy, poor thoracic compliance, and atlantoaxial instability. These factors contribute to a heightened risk of airway-related complications, with unsuccessful intubation attempts leading to potentially fatal outcomes.

The difficulties associated with airway management in patients with Hurler's syndrome arise from features such as macrocephaly, macroglossia, odontoid hypoplasia, etc. Due to these anatomical variations, face mask ventilation can be challenging; however, the inverted mask technique may help achieve an adequate seal.[4]

It is crucial to have a difficult airway trolley readily available in the operating theatre before the administration of anaesthesia. Additionally, sedative premedication should generally be avoided to minimize respiratory compromise. Options for airway control include maintaining spontaneous ventilation during induction, use of supraglottic devices as a primary airway or as a conduit for fibreoptic intubation, video laryngoscopy, and awake or sedated fibreoptic intubation-which remains the gold standard. It allows airway visualization while maintaining spontaneous ventilation. In older patients, awake fiberoptic intubation or intubation following deep inhalation induction is preferred, ensuring spontaneous ventilation is maintained until the airway is secured. Given the risk of failed mask ventilation and intubation, emergency tracheostomy preparedness with ENT backup is mandatory. Securing the airway is crucial in these patients because progressive glycosaminoglycan deposition results in multilevel airway obstruction, high risk of airway collapse under anaesthesia, and increased likelihood of post-operative airway compromise.

Anaesthesia for short procedures can be effectively administered using either a face mask with spontaneous breathing or a laryngeal mask airway (LMA). Intubation is not always required for these brief interventions, thereby reducing the potential challenges associated with intubation and extubation. The nature of the surgical procedure obviated the need for airway instrumentation altogether, thereby preventing potentially catastrophic airway complications. Ketamine can help maintain a patent airway at light levels of anaesthesia, facilitating spontaneous without significant airway obstruction.[5]

Spontaneous ventilation techniques using oxygen along with a high-concentration volatile anaesthetic are also frequently employed; however, these methods necessitate a skilled team ready to manage any potential airway obstruction that may arise. Extubation can be associated with various issues, including partial or complete airway obstruction, which may lead to negative pressure pulmonary edema and hypoxemia.6

Face-mask ventilation was selected because the ophthalmic procedure was short and minimally stimulating. In Hurler syndrome, maintaining spontaneous ventilation is advantageous as it reduces the risk of airway collapse during induction. Mask ventilation also avoids manipulation of a known difficult airway, thereby minimizing trauma, bleeding, and airway edema, and provides a simple, non-invasive means of maintaining anaesthesia for brief procedures.

However, mask ventilation has notable limitations in Hurler syndrome. Macroglossia, craniofacial abnormalities, and multilevel airway narrowing may lead to poor mask seal or obstruction. A face mask does not secure the airway, offering no protection against aspiration and increasing the risk of sudden airway compromise with deeper anaesthesia. Therefore, while used safely in this case, face-mask ventilation should be reserved for carefully selected situations, with readiness to escalate to a supraglottic device, fibreoptic intubation, or surgical airway if required.

The primary goal in managing these patients is to avoid a "can't ventilate, can't intubate" scenario, which underscores the importance of careful planning and preparation. Therefore, critical decisions regarding anaesthetic management should ideally be made by a multidisciplinary team within a tertiary referral center experienced in the perioperative care of MPS patients.

Conclusion

Patients with mucopolysaccharidosis (MPS) present significant challenges for anaesthetists. The anaesthetic risks can be substantially mitigated if the anaesthetist proactively anticipates potential complications that may arise during and after the procedure. Also, it is worth mentioning that airway and anaesthetic management should be tailored to the surgical procedure in potentially difficult airways.

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Conflicts of interest

There are no conflicts of interest.

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