

Anesthetic Considerations for a Patient with Mucopolipidosis type III Intermediate and Chiari I Malformation

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INTRODUCTION

This case discusses a patient with both mucopolipidosis (ML) type III intermediate and Chiari I malformation.

BACKGROUND

Mucopolipidosis (ML) comprises a group of **autosomal recessive lysosomal storage disorders**. Type II and III are caused by mutations in **GNPTAB** or **GNPTG**, which encode **N-acetylglucosamine-1-phosphotransferase**. This enzyme adds the **mannose-6-phosphate (M6P)** tag directing hydrolases to lysosomes. Loss of M6P tagging causes misrouting of hydrolases to the extracellular space, leading to **lysosomal substrate accumulation** and **progressive tissue injury**.

The mucopolipidoses are divided into **types I-IV**, with **type II (I-cell disease)** and **III (pseudo-Hurler polydystrophy)** sharing similar pathophysiology. **Type III intermediate** represents a form between the severe early-onset ML II and the milder ML III variant.

The coexistence of **Chiari I malformation** further complicates care due to risks of **cervical cord compression**, **increased intracranial pressure**, and **restricted neck mobility**.

PATIENT INFORMATION

Age / Sex: 21-year-old female presenting for right total hip arthroplasty.

Past Medical History:

- Mild aortic regurgitation
- Mild mitral stenosis and prolapse
- History of difficult intubation
- Chiari I malformation
- Cervical stenosis
- Mild intellectual delay
- Mucopolipidosis type III intermediate

Anesthesia / Surgical History:

- Adenotonsillectomy
- Left total hip replacement
- Prior difficult airway

CASE DESCRIPTION

The patient was premedicated with **acetaminophen**, **celecoxib**, **midazolam**, and a **scopolamine patch**. On examination, she was **Mallampati III** with **limited neck range of motion**. One year prior, she had an uneventful video-laryngoscopic intubation. General anesthesia was induced with **fentanyl 100 µg**, **lidocaine 60 mg**, **propofol 100 mg**, and **rocuronium 50 mg**. Intubation was performed successfully using video-assisted laryngoscopy with minimal cervical extension. Anesthesia was maintained with a **propofol infusion**, and **phenylephrine** was administered to manage transient hypotension. At the end of the procedure, paralysis was reversed with **sugammadex 2 mg/kg**. Multimodal analgesia included **ketamine 30 mg** and **hydromorphone 0.6 mg**. She received dual antiemetic prophylaxis with **dexamethasone** and **ondansetron**. The patient was extubated uneventfully and spent one night in the hospital.



Figure 1. Patient MRI with Chiari I malformation



Figure 2. female with ML IIIα/β reproduced with permission © 1993-2025 University of Washington

ANESTHETIC CONSIDERATIONS

Mucopolipidosis

The primary concern in ML is the **global stiffening of connective and soft tissues**, leading to:

- **Difficult airway** due to limited cervical mobility and narrowed upper airway structures.
- **Restrictive lung disease** from chest wall rigidity and mucosal thickening.
- **Cardiac valve abnormalities:** thickening of the **mitral** (and less commonly aortic) valve, often presenting as regurgitation.
- **Skeletal deformities (dysostosis multiplex):** thoracic deformities, kyphosis, clubfoot, and abnormal long bones, which can complicate **regional anesthesia**.

Chiari I Malformation

- Defined by **downward displacement of the cerebellar tonsils (>4 mm)** through the foramen magnum into the cervical canal; often associated with **syringomyelia** or **hydromyelia**.
- **30–50%** of patients exhibit **fusion of the skull base with C1** or **partial fusion of C1–C2**.
- Special care is required in **positioning** and **intubation** to prevent neurological injury.
- Depending on severity, anesthesiologists should avoid maneuvers that **increase intracranial pressure (ICP)**.

DISCUSSION

• **ML II and III** are rare, with an incidence of **0.22–2.7 per 100,000 live births**.

• ML III results in slower **progressive stiffening and thickening** of tissues, affecting the **airway**, **cardiac valves**, and **skeletal system**.

• **Preoperative evaluation** of pulmonary and cardiac function is essential, anticipating progressive functional decline.

• Structural changes can lead to **difficult mask ventilation and intubation**, even in patients with previously uneventful anesthetics, due to the **progressive nature of tissue deposition and musculoskeletal contractures**.

• **Chiari I malformations** require careful airway management and avoidance of ICP-elevating stimuli.

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