

# orphanainnesthesia

## Anaesthesia recommendations for patients suffering from **Mucolipidosis II and III**

**Disease name:** Mucolipidosis II and III

**ICD 10:** E77.0

<b>Synonyms:</b> Mucolipidosis II	I-cell disease
	N-acetyl-glucosamine 1-phosphotransferase deficiency
Mucolipidosis III	Pseudo-Hurler polydystrophy

Prevalence is estimated at 0.3/100,000.

Mucolipidosis II (ML II) and Mucolipidosis III (ML III) are inherited metabolic diseases classified as lysosomal storage diseases. Due to a defective N-acetylglucosamine-1-phosphotransferase, growing amounts of carbohydrates lipids and byproducts accumulate in various tissues and organs leading to characteristic deformities and organ insufficiencies. The phenotype resembles Hurler syndrome but in case of ML II with an earlier onset. Why ML III shows a more benign progression than ML II is poorly understood. Whereas in ML II death often occurs by the age of 5 to 8 years, in patients with ML III there is a great variability among patients, and individuals can survive into their fourth or fifth decade.

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Medicine in progress

 Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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### **Typical surgery**

Adenoidectomy, tonsilectomy paracentesis and drainage, herniotomy, carpal tunnel syndrome, PEG (percutaneous endoscopic gastrostomy).

### **Type of anaesthesia**

High complication rates of general anaesthesia in children with ML II have been reported. This leads to the recommendation of checking carefully alternative methods if possible at all.

Carpal tunnel syndrome might be manageable with regional anaesthesia of the axillary plexus. As a result of cutaneous depositions of metabolic byproducts and stiff joints, an approach with ultrasound if available seems to be more promising.

Whether sedation combined with analgesics for less invasive procedures like paracentesis might be advantageous has to be considered on a case-by-case basis.

### **Necessary additional diagnostic procedures (preoperative)**

#### Airway/pulmonary function:

Jaw and neck may be stiff. The neck can also be short. The gingivae are usually hypertrophic and the teeth easily damaged. The base of the tongue, larynx and epiglottis may be thickened as well as the tracheal wall. Adenoids and tonsils can be enlarged. Thoracic deformities, increased secretions and ineffective coughing lead to respiratory infections. Obstructive sleep apneas frequently occur with the need of oxygen therapy or even the use of CPAP-masks.

If there is a history of obstructive sleep apneas then polysomnography is indicated.

#### Cardiovascular:

Rarely cardiomyopathy (dilated or hypertrophic), thickened valves, pulmonary hypertension, coronary artery occlusion; cardiac evaluation including echocardiography (valvular pathology, cardiomyopathy).

#### Miscellaneous:

Various degree of mental and motor developmental delay; dwarfism, corneal opacities, deafness; kyphoscoliosis, stiff joints, cutaneous infiltrations leading to difficult venous access. Hepatosplenomegaly leading to abdominal distension.

### **Particular preparation for airway management**

Direct laryngoscopy and tracheal intubation may be very difficult and become even more difficult as patients grow up. It may be difficult to maintain a patent airway with a face mask, even with an oral airway. In patients with Hurler syndrome, laryngeal masks airways are sometimes difficult or impossible to insert. In ML II successful management with assisted spontaneous ventilation, laryngeal mask airway and fiberoptic laryngoscopy has been reported.

For a safe airway management, it is essential to use a difficult airway algorithm and have a skilled theatre team and difficult airway management devices available.

These include various paediatric face masks, oral airways, nasal airways, laryngeal mask airways of different sizes and a small size fiberoptic scope. As the patient's tracheal wall may be thickened, an endotracheal tube that is smaller than predicted may be required. Due to anteverted small nostrils and adenoid hyperplasia, the insertion of a nasal airway may be impossible or even cause bleeding. A carefully secured iv access should be established before anaesthetic induction (in the author's experience).

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#### **Particular preparation for transfusion or administration of blood products**

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Not reported.

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#### **Particular preparation for anticoagulation**

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Not reported.

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#### **Particular precautions for positioning, transport or mobilisation**

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Not reported.

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#### **Probable interaction between anaesthetic agents and patient's long-term medication**

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Not reported.

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#### **Anaesthesiologic procedure**

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Sedative premedication is relatively contraindicated in patients with airway difficulties, especially in those with obstructive sleep apnea and/or chronic respiratory insufficiency. Both conditions often present in ML children.

Intravenous access in ML children might be very difficult due to cutaneous depositions of metabolic byproducts. Therefore if it is considered to induce general anaesthesia via face mask without previous iv access it is recommended to have devices for intraosseous needle access available.

Antimuscarinics: In children with severely compromised respiratory function and/or airway difficulties additional secretions might increase the risk of respiratory complications. Hence the use of atropine or glycopyrrolate might be beneficial in these children, especially in patients undergoing airway investigations, oropharyngeal surgery or requiring fiberoptic intubation. Notably if sedation with ketamine is considered, the antisialogogue action of antimuscarinics is well appreciated.

Airway management: Maintaining spontaneous ventilation after inhalational induction or intravenous titration of an anaesthetic agent (e.g. propofol) and checking intermittently that control ventilation is possible seems to be a validated approach. If the airway is partially obstructed during induction, early use of CPAP allows deepening of the anaesthetic, while

maintaining spontaneous respiration. Due to anteverted small nostrils and adenoid hyperplasia, the insertion of a nasal airway may be impossible or even cause bleeding. Therefore the use of an oral airway in case of pharyngeal obstruction might be the better choice.

If controlled ventilation by bag is sufficient, the use of a short acting muscle relaxant might optimise conditions for direct laryngoscopy. If conventional laryngoscopy is not successful, fiberoptic intubation can be performed through a laryngeal mask airway.

#### **Particular or additional monitoring**

Children with difficult airways or compromised respiratory function are prone to respiratory complications even during moderate sedation or less invasive procedures. Hence they should be fully monitored, and end-tidal capnography should be immediately available.

#### **Possible complications**

Difficult airway management. Respiratory insufficiency due to chronic pulmonary disease.

#### **Postoperative care**

Patients with chronic airway obstruction or obstructive sleep apneas should be observed closely postoperatively. Due to difficulties in swallowing, even small bleeding complications after oral or pharyngeal surgery may lead to severe respiratory impairment.

If postoperative ventilation is required, early extubation should be an aim in order to minimize ventilator associated complications.

#### **Information about emergency-like situations / Differential diagnostics**

*caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease*

Not reported.

#### **Ambulatory anaesthesia**

Because of the high risk of postoperative respiratory complications, ambulatory surgery cannot be recommended (especially when surgery involved the airway or if the trachea was intubated).

#### **Obstetrical anaesthesia**

Not reported.

### Literature and internet links

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6. Black A. Management of the difficult airway. 315-328, Hatch & Sumner's Textbook of Paediatric Anaesthesia,3rd Ed.,2008,Hodder Arnold.

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**Last date of modification:** June 2011

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