orphanaesthesia

Anaesthesia recommendations for

Mucolipidosis II and Mucolipidosis III

Disease name: Mucolipidosis II, Mucolipidosis III

ICD 10: E77.0

Synonyms: I-Cell Disease (Mucolipidosis II), Pseudo-Hurler Dystrophy (Mucolipidosis III)

Disease summary: The 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. These autosomal recessive diseases are related to the mucopolysaccharidoses. 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.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Typical surgery

Adenoidectomy, Tonsillectomy, Paracentesis and Drainage, Gingivectomy (ML II), Herniotomy, Carpal Tunnel Syndrome, PEG (Percutaneous endoscopic gastrostomy).

Type of anaesthesia

High complication rates of general anesthesia in children with ML II have been reported. This lead 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 metabilc 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 pre-operative testing (beside standard care)

Airway/Pulmonary Function: Coarse Facies. Jaw and neck may be stiff. The neck can also be short. The gingivae in ML II 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 (OSA) frequently occur with the need of oxygen therapy or even the use of CPAP-masks.

If there is a history of OSA, then polysomnography may be indicated.

Cardiovascular: Rarely hypertrophic cardiomyopathy, thickened valves with mitral or aortic valve regurgitation, pulmonary hypertension as well as coronary artery occlusion have been described.

Cardiac evaluation including echocardiography.

Miscellaneous: Various degree of mental and motor developmental delay. The patient may have hearing loss. Dwarfism, corneal opacities. 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 to 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).

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Patients must be carefully positioned and padded secondary to stiff joints. Unstable atlantoaxial joints with atlantoaxial dislocation and consecutive spinal cord injury in children with ML II have been described. Therefore a careful positioning of the cervical spine during airway management and surgery could be advised.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Sedative premedication is relatively contraindicated in patients with airway difficulties, especially in those with OSA 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, in the case induction of general anaesthesia via face mask without previous iv.-access is considered, it is recommended to have devices available for intraosseous needle access.

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 glycopyrolate might be beneficial in these children, especially in patients undergoing airway investigations, oropharyngeal surgery or requiring fiberoptic intubation.

Notably if sedations with ketamine are 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 the 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 and tracheal intubation is required, 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

Post-operative care

Patients with chronic airway obstruction or OSA 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.

Disease-related acute problems and effect on anaesthesia and recovery

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).

Not reported.

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