

## Anaesthesia recommendations for patients suffering from **Sanfilippo disease**

**Disease name:** Mucopolysaccharidosis type III

**ICD 10:** E76.2

**Synonyms:** MPS III, Sanfilippo syndrome, Mucopolisaccaridosi type III

Mucopolysaccharidosis type III (MPS III) disease is an autosomal recessive disease, belonging to the group of mucopolysaccharidoses characterized by accumulation of heparin sulfate and characterized by severe and rapid intellectual deterioration. There are recognized four enzymatically distinct forms, with the prevalence of subtypes; type IIIA – IIIB – IIIC and IIID. The disorder is underdiagnosed (due to the generally very mild dysmorphism); it is the most frequent MPS in the Netherlands and Australia with respective prevalences of 1:53,000 and 1:67,000. The frequency of the different subtypes varies between countries: Subtype A is more frequent in England, the Netherlands and Australia and subtype B is more frequent in Greece and Portugal, whereas types IIIC and IIID are much less common. The first symptoms appear between the ages of 2 and 6 years, with behavioral disorders (hyperkinesia, aggressiveness) and intellectual deterioration, sleep disorders and very mild dysmorphism. The neurological involvement becomes more prominent around the age of 10 years with loss of motor milestones and communication problems. Seizures often occur after the age of 10. A few cases of attenuated forms have also been reported. The neurological degradation accompanied by multiple complications requires a multidisciplinary management to allow adapted symptomatic treatment. The prognosis is poor with death occurring in most cases of type IIIA at the end of the second decade. Longer survival times (30/40 years) have been reported for the B and D subtypes. When these patients undergo surgical procedures, often intended to improve their quality of life, they have serious anaesthetic difficulties due to skeletal, neurologic and cardio-respiratory alterations.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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Find more information on the disease, its centres of reference and patient organisations on Orphanet: [www.orpha.net](http://www.orpha.net)

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### **Typical surgery**

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International literature is very poor about these patients, but likely all kinds of surgery can occur in these patients also in emergency situations, as (adeno-) tonsillectomy, tympanostomy, carpal tunnel release, trigger-finger release, spinal fusion, hip replacement.

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### **Type of anaesthesia**

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General anaesthesia, central blocks or local anaesthesia as periferic nerve blocks in slight sedation (if behavioural problems do not interfere) may be performed, due to surgery.

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

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Cardiac evaluation whit ECG is mandatory due to possible prolonged QT syndrome and other cardiac disorders of conductions (Spontaneous development of complete atrioventricular block has been described). Also a deep pneumological evaluation and assessment is important because frequently heart and lungs impairment, most often seen in patients suffering from MPS III (spirometry, evaluation by a (paediatric) lung specialist). Accurate preoperative anaesthesiological evaluation is necessary to assess clinical conditions and anatomical limitations particularly of mouth, throat and spine.

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

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Skeletal malformation particularly of mouth, troath and cervical spine may cause difficult airways management. Accumulation of heparin sulfate in the tissues surrounding the upper respiratory tract in the nasopharynx, oropharynx, hypopharynx and larynx plays a role in the difficulty of airway management, so it is mandatory to have all devices for difficult intubation as well as video-laryngoscope available.

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

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Stem cell transplantation candidates require special blood components, such as leukocyte-reduced cellular, cytomegalovirus seronegative, and/or gamma-irradiated components. Transplantation patients may require a large number of transfused blood products, as a result of pancytopenia and organ and tissue damage sustained during the procedure. After successful stem cell transplantation, blood type changes to the blood type of the donor.

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### **Particular preparation for anticoagulation**

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None.

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

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Positioning should be undertaken with care due to the possibility of osteonecrosis of the femoral heads, hip dysplasia, scoliosis with thoracolumbar kyphosis and dysplastic/misshapen vertebral bodies. If is concomitant a respiratory disease and is performed an central block is appropriate to place these patients in the best way to allow spontaneous respiration, or give them a few amount of oxygen supplementation.

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

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Sometimes these patients take anticonvulsivant drugs that can interfere with anaesthetics, opioids or hypnotic drugs.

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### **Anaesthesiologic procedure**

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Patients with MPSIII should only undergo anaesthesia/surgery in centres experienced with the perioperative management of individuals with this disease. This needs more detail on planning, selection of anaesthetic induction and the use of airway adjuncts. However, no particular procedures are necessary performing induction and maintenance of general anaesthesia. Take care about medications that increase QT-interval, particularly in patients with long-QT syndrome before anaesthesia.

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### **Particular or additional monitoring**

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None.

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### **Possible complications**

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- Inability to ventilate or intubate the patient
- Complete airway obstruction
- Failure to maintain airway after extubation, stridor, upper or lower airway collapse.
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### **Postoperative care**

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Prevention of lower respiratory tract infections as pneumonia, by physiotherapeutic treatment. Take care of the risk of respiratory failure and/or apnea after anaesthesia, more specifically after general anaesthesia requiring a prolonged (24hs?) medical supervision, due to the risk of airway obstruction episodes and desaturation.

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

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

None.

## **Ambulatory anaesthesia**

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These kind of patients are always assisted and accompanied by a tutor because their mental and physical impairment, so if a slight anaesthesia or sedation is performed they are assisted after ambulatory surgery.

## **Obstetrical anaesthesia**

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These patients are unlikely to exceed the second decade of life and if it happens, neurological and mental conditions are such not to provide a state of pregnancy. In the very off chance that this could happen, the preferred standard is subarachnoid anaesthesia.

## Literature and internet links

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7. Cleary MA, Wraith JE. Management of mucopolysaccharidosis type III. *Arch Dis in Childhood* 1993;69:403-406
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**Peer revision 2**

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