

A&I

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Nemaline myopathy

Niemann-Pick Type C

orphan**anesthesia**

a project of the German Society
of Anaesthesiology and Intensive Care Medicine

SUPPLEMENT NR. 9 | 2022

OrphanAnesthesia –

ein krankheitsübergreifendes Projekt des Wissenschaftlichen Arbeitskreises Kinderanästhesie der Deutschen Gesellschaft für Anästhesiologie und Intensivmedizin e.V.

Ziel des Projektes ist die Veröffentlichung von Handlungsempfehlungen zur anästhesiologischen Betreuung von Patienten mit seltenen Erkrankungen. Damit will OrphanAnesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patienten mit seltenen Erkrankungen benötigen für verschiedene diagnostische oder therapeutische Prozeduren eine anästhesiologische Betreuung, die mit einem erhöhten Risiko für anästhesieassoziierte Komplikationen einhergehen. Weil diese Erkrankungen selten auftreten, können Anästhesisten damit keine Erfahrungen gesammelt haben, so dass für die Planung der Narkose die Einholung weiterer Information unerlässlich ist. Durch vorhandene spezifische Informationen kann die Inzidenz von mit der Narkose assoziierten Komplikationen gesenkt werden. Zur Verfügung stehendes Wissen schafft Sicherheit im Prozess der Patientenversorgung.

Die Handlungsempfehlungen von OrphanAnesthesia sind standardisiert und durchlaufen nach ihrer Erstellung einen Peer-Review-Prozess, an dem ein Anästhesist sowie ein weiterer Krankheitsexperte (z.B. Pädiater oder Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, so dass die Handlungsempfehlungen grundsätzlich in englischer Sprache veröffentlicht werden.

Ab Heft 5/2014 werden im monatlichen Rhythmus je zwei Handlungsempfehlungen als Supplement der A&I unter www.ai-online.info veröffentlicht. Als Bestandteil der A&I sind die Handlungsempfehlungen damit auch zitierfähig. Sonderdrucke können gegen Entgelt bestellt werden.

OrphanAnesthesia –

a project of the Scientific Working Group of Paediatric Anaesthesia of the German Society of Anaesthesiology and Intensive Care Medicine

The target of OrphanAnesthesia is the publication of anaesthesia recommendations for patients suffering from rare diseases in order to improve patients' safety. When it comes to the management of patients with rare diseases, there are only sparse evidence-based facts and even far less knowledge in the anaesthetic outcome. OrphanAnesthesia would like to merge this knowledge based on scientific publications and proven experience of specialists making it available for physicians worldwide free of charge.

All OrphanAnesthesia recommendations are standardized and need to pass a peer review process. They are being reviewed by at least one anaesthesiologist and another disease expert (e.g. paediatrician or neurologist) involved in the treatment of this group of patients.

The project OrphanAnesthesia is internationally oriented. Thus all recommendations will be published in English.

Starting with issue 5/2014, we'll publish the OrphanAnesthesia recommendations as a monthly supplement of A&I (Anästhesiologie & Intensivmedizin). Thus they can be accessed and downloaded via www.ai-online.info. As being part of the journal, the recommendations will be quotable. Reprints can be ordered for payment.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

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orphan^aesthesia

Anaesthesia recommendations for Niemann-Pick Type C

Disease name: Niemann-Pick Type C

ICD 10: E75.242

Synonyms: Juvenile Niemann-Pick disease

Disease summary: Niemann-Pick disease type C (NPC) is an autosomal recessive lipid lysosomal storage disorder with incidence of approximately 1:120,000 live births. NPC involves alterations in the intracellular transport of endocytosed cholesterol and accumulation of unesterified cholesterol in lysosomes and endosomes due to mutations in either the NPC1 or NPC2 genes. Clinically, patients with NPC can present with a spectrum of neurological deficits including vertical gaze palsy, dystonia, dysphagia, seizures, and progressive dementia. With systemic disease, hepatosplenomegaly can be severe. Lung involvement can be present and often result from severe neurological impairment and associated dysphagia, recurrent aspirations, and decreased thoracic muscle strength. The disease is associated with a significant decrease in life expectancy, and unfortunately effective therapy is lacking. Given the spectrum of severity, the broad spectrum of clinical presentation, and the involvement of various organs, it is highly likely that many NPC patients will require diagnostic and therapeutic procedures during the course of their disease, which will require anaesthesia.

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

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Typical surgery

Patients with NPC may require a multitude of diagnostic and therapeutic procedures including, but not limited to: imaging studies, lumbar punctures, intrathecal therapeutic injections, vascular or intrathecal access device placement, percutaneous feeding tube placement, auditory brainstem response, skin biopsies.

Type of anaesthesia

Given the risk of aspiration, general anaesthesia with intratracheal intubation is often required, especially for patients who have severe lung disease, recurrent aspiration and chronic cough. Inhalational anaesthetics (sevoflurane and N₂O), sedatives and propofol have all been administered without complications. Intravenous sedation without airway instrumentation for patients who are not at high risk of aspiration can and should be entertained. Cognitive impairment is frequently present and should be taken into consideration when regional or local anaesthesia are being used, as patients may be unable to cooperate with such procedures. Respiratory depression can be a concern in patients with NPC who have pulmonary involvement. Therefore, the risk associated with the use of opioids and sedatives should be weighed against its benefits.

Necessary additional pre-operative testing (beside standard care)

Hepatosplenomegaly is invariably present in NPC. Patients may have liver function abnormalities and ascites. In addition, in part due to hypersplenism, thrombocytopenia may be present, with platelet dysfunction sometimes further exacerbated by the use of anticonvulsants such as sodium valproate. In patients with NPC, the evaluation of liver function tests, complete blood count, and coagulation profile need to be considered before invasive procedures. Correction of potential abnormalities should be thought of in the context of the planned invasive procedure. A chest radiograph might be helpful in patients with a history of frequent aspirations.

As many patients may be fed by gastric or jejunostomy tubes, feeds should be discontinued 8 hours before any procedure.

Particular preparation for airway management

NPC is not associated with craniofacial defects, therefore airway abnormalities are not to be expected.

Particular preparation for transfusion or administration of blood products

As indicated above, these patients may have thrombocytopenia. Therefore, platelet transfusions may be required.

Particular preparation for anticoagulation

Patients with NPC might have thrombocytopenia, elevated PT, and/or PTT elevation if severe liver dysfunction is part of the disease. Therefore, the use of anticoagulants should be carefully monitored.

Particular precautions for positioning, transportation and mobilisation

Patients with NPC are at increased risk for recurrent aspiration and might have pulmonary dysfunction due to recurrent aspirations and infections. Therefore, oxygen supplementation is often required during transportation and recovery of anesthetised patients.

Interactions of chronic disease and anaesthesia medications

None known.

Anaesthetic procedure

Volatile anaesthetics, like sevoflurane and N₂O can be safely administered. However, as these patients can have a history of seizures, sevoflurane may precipitate seizure activity, especially if patients are not compliant with their prescribed anticonvulsant regimen.

Opiates and propofol usually can be safely used as part of the anaesthetic management of these patients.

Particular or additional monitoring

Routine ASA monitors, including HR, BP, respiration rate, O₂ saturation, EtCO₂, body temperature are needed. Additional monitoring will depend on the nature of the planned procedure.

Possible complications

NPC patients are at high risk for post-operative respiratory complications, hypoxia and may require prolonged endotracheal intubation to protect the airway and prevent aspiration. Patients with NPC are at risk of intraoperative hypothermia. In one report, despite efforts to rewarm the patients, hypothermia (34.4 and 35.2° C) was significant.

Post-operative care

For patients with pre-operative severe lung disease, an intraoperative hypoxic episode, seizure episode or possible perioperative aspiration, ICU admission should be considered for further monitoring and management.

Disease-related acute problems and effect on anaesthesia and recovery

NPC and anaesthesia can both lead to aspiration pneumonia, seizure episode or hypoxia.

Ambulatory anaesthesia

Patients with early disease stages may be suitable for ambulatory anaesthesia and discharge home on the same day. In contrast, patients with severe NPC may require careful monitoring during the peri-operative period especially if there is a suspicion of aspiration, hypoxia or ongoing seizures.

The decision to discharge the patient on the same day of the surgery should be discussed with the anaesthetist, surgical team, and patient family members when objective discharge criteria are met.

Obstetrical anaesthesia

Not reported.

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