

Anaesthesia recommendations for patients suffering from

Niemann-Pick Type C

Disease name: Niemann-Pick type C

ICD 10: E75.242

Synonyms: Juvenile Niemann-Pick disease

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 unsterified 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 significant decrease in life expectancy, and unfortunately effective therapy is lacking. Given the spectrum of severity, broad spectrum of clinical presentation, and 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 in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong



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

Typical surgery

Patient 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 diagnostic procedures (preoperative)

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 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 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, transport or 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 required during transportation and recovery of anesthetized patients.

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

None known.

Anaesthesiologic 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 postoperative 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.

Postoperative care

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

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

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 perioperative 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.

Literature and internet links

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