Fucosidosis

Giant axonal neuropathy
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.


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.

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Anaesthesia recommendations for

**Fucosidosis**

**Disease name:** Fucosidosis

**ICD 10:** E77.1

**OMIM 230000**

**Synonyms:** Alpha-L-Fucosidase Deficiency

**Disease summary:** Fucosidosis is an extremely rare lysosomal storage disease, characterised by a deficiency of the enzyme alpha-L-fucosidase. Fucosidosis is inherited as an autosomal recessive genetic trait. Fucosidosis types 1 and 2 may occur in the same family. The gene has been localised at 1p36-p34 (locus FUC1), but there is also a pseudogene on chromosome 2, and the FUC2 gene, localised on chromosome 6, regulating the activity of alpha-L-fucosidase in fibroblasts. Over 20 mutations have been identified so far.

Low levels of the alpha-L-fucosidase enzyme lead to the abnormal accumulation of certain fucose-containing complex compounds (i.e., glycosphingolipids, glycolipids, and glycoproteins) in many tissues of the body. There are two relevant types of fucosidosis, determined mainly by the time of onset and severity of clinical symptoms. Some scientists theorise there are three types, with the age of onset and the disease severity being the determining factors.

The symptoms of fucosidosis type 1, the most severe form of the disease, may become apparent between 3 and 18 months of age. Symptoms may include progressive deterioration of the central nervous system, mental retardation, loss of previously acquired intellectual skills, seizures and growth retardation leading to short stature. Other abnormalities become apparent over time including multiple deformities of the bones (dysostosis multiplex), ovoid breaking vertebrae with kyphoscoliosis, coarse facial features (Hurler-like appearance), cardiomegaly and hepatosplenomegaly. Additional symptoms may include malfunction of the gallbladder, salivary and sweat glands that produce sweat with high NaCl content. Death usually occurs during the first decade of life.

In fucosidosis type 2, deterioration of the central nervous system becomes apparent after the first years of life; symptoms may be similar but milder and progress more slowly than in type 1, and sweat salinity is normal. Dystonia, progressive deafness and tortuous conjunctival vessels are present. The most noticeable feature distinguishing fucosidosis type 1 from type 2 is the appearance of angiokeratomas on the skin around 10 years of age in those individuals with type-2 disease.
Medicine is in progress

或许新的知识

每个患者都是独一无二的

或许诊断是错误的

更多关于疾病的更多信息，其中心和患者组织，可以在Orphanet上找到：www.orpha.net
Typical surgery

Reported cases are rare and patients may present at different ages for different types of surgeries/procedures and examinations, for example, magnetic resonance imaging, CT scans, bone marrow transplantation, dental, ophthalmologic, orthopaedic and urologic procedures among many others.

Type of anaesthesia

Each patient should be evaluated on an individual basis. The abnormal accumulation of certain fucose-containing complex compounds makes many tissues of the body affected by the disease (e.g. vacuolated lymphocytes). The anaesthetic management of patients with fucosidosis may be complicated by airway problems such as difficult laryngoscopy secondary to dysmorphic features and difficult visualisation of the vocal cords. A careful evaluation and management of a possible difficult airway should be planned. Induction of anaesthesia with maintenance of spontaneous ventilation is therefore highly recommended.

Preoperative examination should also look for cardiomegaly and/or hepatosplenomegaly and determine the function of these organs. Anaesthesia medications’ choice and dosages should be adjusted accordingly.

As some patients may have metabolic disorders such as hypothyroidism, an endocrinological evaluation may be indicated. Patients with fucosidosis may be predisposed to fluctuations in body temperature secondary to sweat glands malfunctioning. Special caution should be exerted in maintaining adequate hydration and normothermia. Careful positioning adapted to bone deformities should be applied.

Hyper- or hyposialorrhoea secondary to salivary glands disturbances may be present.

There are no reports about regional anaesthesia in patients with fucosidosis. The use of regional anaesthesia could be considered after excluding any neurological or anatomical anomalies (kyphoscoliosis) that may exclude this technique.

Both intravenous and inhalational anaesthesia techniques are suitable for patients with fucosidosis.

Necessary additional pre-operative testing (beside standard care)

Findings to be considered prior to planning anaesthesia care are the involvement of vital organs. The presence and severity of cardiomegaly, hepatosplenomegaly, renal, urological, endocrine and metabolic disturbances should be documented.

Baseline neurological/mental status and skeletal deformities or spasticity should be assessed, including visual and hearing loss.

Type, frequency, severity of seizures should be documented and their treatment should be optimised preoperatively.

The presence of angiokeratomas and bone deformities should be documented.
Particular preparation for airway management

Careful evaluation for difficult mask ventilation and tracheal intubation secondary to dysmorphic features: facial and mandibular deformities and possibly macroglossia.

Particular preparation for transfusion or administration of blood products

None reported.

Particular preparation for anticoagulation

None reported.

Particular precautions for positioning, transportation and mobilisation

Prevention of risk of injury from seizures and careful positioning secondary to bone deformities and spasticity. Neurological/mental developmental delay and visual disturbances require help with mobilisation and transport.

Interactions of chronic disease and anaesthesia medications

There may be possible interactions between anaesthetic agents and patient’s chronic medication such as anti-seizures therapy, secondary to their effect on CYP450. Special pharmacological considerations for this syndrome are related to involvement of vital organs such as liver and kidney that may alter the metabolism and clearance of medications. Severe cardiomegaly and excess sweating may affect cardiac output and hydration status. The only discussed specific treatment is bone marrow allograft. Strict sterility techniques should be applied for patients undergoing or having undergone a bone marrow allograft.

Anaesthetic procedure

Consideration and preparation for a possible difficult airway. Possibility of cardiomegaly and other vital organs involvement. Possibility of hypothyroidism and seizures.

Particular or additional monitoring

Individual basis in the presence of cardiac or liver/kidney involvement. Temperature monitoring with the goal of achieving normothermia is important in these patients.
Possible complications

Give special attention to the following points: Cardiomegaly/hepatomegaly/neurological problems: Seizures/potential difficult airway.

Postoperative care

Documentation and stabilisation of:

- Airway patency: obstruction caused by macroglossia, risk of obstructive sleep apnoea.
- Haemodynamic stability in the presence of cardiac or involvement of vital organs.
- Management of seizures, neurological involvement: bladder problems as well as help with visual impairment, if present.
- Positioning adapted to bone deformities.
- Prevention of hypo- and hyperthermia and regular assessment of hydration status secondary to sweat glands abnormalities in type 1.
- Hyper- or hyposialorrhoea secondary to salivary glands disturbances.

Disease-related acute problems and effect on anaesthesia and recovery

Not reported.

Ambulatory anaesthesia

Each patient must be evaluated carefully for co-morbidities and/or airway issues. Anaesthesia and surgery have to be performed in a medical facility with a capacity of taking care of potential complications: difficult airway, cardiac and hepatic dysfunction, hypothyroidism, seizures.

Obstetrical anaesthesia

Anaesthesia and surgery have to be performed in a medical facility with a capacity of taking care of potential challenges and complications: difficult airway, cardiac and hepatic dysfunction, hypothyroidism, seizures.
Literature and internet links


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This recommendation has been prepared by:

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