orphanaesthesia

Anaesthesia recommendations for

Fucosidosis

Disease name: Fucosidosis (OMIM 230000)

ICD 10: E77.1

Synonyms: Alpha-L-Fucosidase Deficiency

Disease summary: Fucosidosis is an extremely rare lysosomal storage disease, characterrized 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 localized at 1p36- p34 (locus FUCA 1), but there is also a pseudogene on chromosome 2, and the FUCA 2 gene, localized on chromosome 6, regulates 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 theorize 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 beaking 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 constant progress; new clinical knowledge may not be in this guideline.

Recommendations are not rules or laws; they are a framework for clinical decision-making.

Every patient is unique; individual circumstances must guide clinical care.

The diagnosis may be wrong; if questionable, the diagnosis should be confirmed.

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

Typical surgery

Reported cases are rare and patients may present at different ages for different types of surgeries/procedures and examinations: such as 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 anesthetic management of patients with Fucosidosis may be complicated by airway problems such as difficult laryngoscopy secondary to dysmorphic features and difficult visualization of the vocal cords. A careful evaluation and management of a possible difficult airway should be planned. Induction of anesthesia 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. Anesthesia 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 hyposialorrhea 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 optimized 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 have 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

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.
- Hemodynamic stability in the presence of cardiac or vital organs' involvement.
- 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 hyposialorrhea 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 to be performed in a medical facility with capacity of taking care of potential complications: difficult airway, cardiac and hepatic dysfunction, hypothyroidism, seizures.

Obstetrical anaesthesia

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

Literature and internet links

1. Soltani AE, Moharari RS, Ghaffari R, Zahedi H, Hajmahmoodi M. Fucosidosis and anesthesia. Saudi Med J. 2007;28(9):1446-1448
2. Abdallah C, Hannallah R, McGill W. Anesthesia for fucosidosis. Paediatr Anaesth. 2007;17(10):994-997
3. http://rarediseases.org/rare-diseases/fucosidosis/ accessed on 03/25/2016.

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