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Friedreich's ataxia

Glycogen storage disease type I

orphanaesthesia

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

SUPPLEMENT NR. 4 | 2017

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 Orphan Anesthesia 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.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

www.ai-online.info/Orphsuppl
www.orphananesthesia.eu

OrphanAnesthesia –
a common 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 recommenations 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.

A survey of until now in A&I published guidelines can be found on:

www.ai-online.info/Orphsuppl
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orphananesthesia

Anaesthesia recommendations for patients suffering from

Friedreich's ataxia

Disease name: Friedreich's ataxia

ICD 10: G11.1

Synonyms: -

Friedreich's ataxia (FRDA) is the most common autosomal recessive ataxia in the Caucasian population characterized by ataxia, predominantly sensory neuropathy, cardiomyopathy, and diabetes mellitus. The incidence in Caucasians has been estimated between 1:29,000 to 1:50,000 in different populations. Men and women are affected equally [1,2]. The primary pathology involves degeneration of the dorsal root ganglia, posterior columns, corticospinal, ventral and lateral spinocerebellar tracts, and the dentate nuclei of the cerebellum. FRDA segregates as an autosomal recessive trait, and patients have mutations in the gene FXN that encodes the protein frataxin. The typical mutation found in 96% of the patients is an abnormal expansion of the trinucleotide GAA (guanine, adenine, adenine trinucleotide) in the first intron. Frataxin is a mitochondrial protein and has a role in iron homeostasis and antioxidation [3]. The mutation leads to reduced levels of frataxin, with subsequent accumulation of iron and impaired electron transport in the respiratory chain in the mitochondria. The resulting impairment in mitochondrial function causes pathology in the peripheral and central nervous system, the heart myocardial fibers and the pancreatic islets of Langerhans [4]. Initial symptoms of FRDA typically occur before the age of 25, and the typical presentation includes varying degrees of ataxia in all four limbs, absent lower extremity reflexes, and pyramidal signs. Most patients have an abnormal electrocardiogram due to hypertrophic cardiomyopathy. Other signs are pes cavus, saccadic intrusions, optic atrophy, deafness, diabetes mellitus or glucose intolerance. Death is usually due to cardiac dysfunction, including arrhythmias or heart failure [1,5].

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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Typical surgery

Typically, anaesthesia may be required for orthopaedic surgeries such as correction of pes cavus, Achille's tendon tenotomy or correction of kyphoscoliosis [6].

Type of anaesthesia

There is no definite recommendation for either general or regional anaesthesia.

The patients with FRDA usually have bulbar symptoms and therefore are at risk for aspiration, and peri- and postoperative respiratory complications. This risk is additionally raised due to thoracic kyphoscoliosis with restrictive respiratory function [7].

Anaesthesia concerns must be focused on the safe use of muscle relaxants and on the prevention of possible peri- or postoperative cardiac involvement. Depolarizing muscle blockers (e.g. succinylcholine) should be avoided because there is an increased risk of hyperkalaemia. Non-depolarizing muscle relaxants can safely be used, but patients may have an increased sensitivity, so monitoring of the neuromuscular blockade is compulsory [7-11].

General anaesthesia can be performed as total intravenous technique [7] or balanced anaesthesia. It has been suggested that propofol-based total intravenous anaesthesia (TIVA) should not be used in these patients because of its depressant effects on mitochondrial metabolism, and the possible mitochondrial role in propofol-infusion syndrome [12].

Regional or local anaesthesia have been reported without complications [13-16].

Necessary additional diagnostic procedures (preoperative)

The patients with FRDA have a significantly increased risk for cardiomyopathy, and congestive heart failure. One third of patients develop impaired glucose tolerance or diabetes mellitus. Due to the frequency of neurologic, cardiac, pulmonary, and endocrine disorders in the patients, preanaesthetic evaluation should be carried out carefully [1,7].

Consider echocardiography, electrocardiogram, and blood gas analysis preoperatively.

Particular preparation for airway management

Not reported.

Particular preparation for transfusion or administration of blood products

Not reported.

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Particular preparation for anticoagulation

There are no particular recommendations.

Particular precautions for positioning, transport or mobilisation

The severe kyphoscoliosis can lead to difficult positioning.

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

Due to impaired glucose tolerance and increased risk of diabetes mellitus, avoid steroid substitution and dextrose infusion.

Some patients may use botulinum toxin, and it is recommended to avoid the use of botulinum toxin injections ahead of general anaesthesia. Some patients may also use baclofen [18].

Anaesthesiologic procedure

Avoid succinylcholine due to the risk of hyperkalaemia.

Consider to avoid the use of nitrous oxide because of its cardio-depressant and neuropathic effects.

Consider rapid sequence induction due to the increased risk of aspiration.

There are reports about the safe use of volatile anaesthetics, opiates and propofol, even some authors recommend to avoid propofol.

The use of non-depolarizing muscle relaxants is safe but monitoring of the neuromuscular blockade is mandatory.

Some authors recommend using Bispectral Index Monitors to assess the depth of anaesthesia and to promote a faster recovery [11].

About 10% of FRDA patients have diabetes mellitus, and 30% have impaired glucose tolerance. Whenever glucose-containing solutions with electrolytes are used, the blood glucose concentration should be monitored to avoid hyperglycaemia [5,15].

Patients with FRDA have a significantly increased risk for cardiomyopathy, and congestive heart failure. Hypertrophic cardiomyopathy leads to increased risk of serious arrhythmias, and most patients will develop electrocardiographic and echocardiographic abnormalities during the disease course [17,7,18].

Some authors prefer the use of regional anaesthesia instead of general anaesthesia whenever possible [12]. There are reports of spinal, epidural, combined spinal-epidural and peripheral nerve blocks for anaesthesia without any complications [13-16]. However, it has been reported that technical difficulties may arise in patients with spasticity, especially when spasm occur in flexion and/or extension. There are no known contraindications to local anaesthesia/dental anaesthesia in FRDA [17].

Particular or additional monitoring

The blood glucose concentration should be monitored to avoid hyperglycaemia due to the metabolic disturbance and impaired glucose metabolism associated with FRDA [14].

Monitoring of the neuromuscular transmission is mandatory if neuromuscular blocker are used [7].

One should pay close attention to the monitoring of fluid balance and cardiovascular function in people with FRDA undergoing anaesthesia (because of heart disease).

Possible complications

There are increased risk of aspiration and respiratory insufficiency [7]. Succinylcholine can induce hyperkalaemia that may cause cardiac arrest.

Postoperative care

Consider close postoperative monitoring of cardiac arrhythmias and cardiac function. In case of cardiac and pulmonary impairment, postoperative intensive or intermediate care is necessary.

Because of impaired glucose tolerance or diabetes mellitus, the glucose concentration should be monitored [14].

Confinement to bed and prolonged immobilisation prior to or during surgery can lead to aggravation of neurological difficulties, which must not be unreasonably attributed to the anaesthesia. Difficulties are also frequently underestimated before surgery, and postoperatively brought to the fore and wrongly attributed to the actual anaesthesia [16].

Whenever possible, early mobilization and institution of physical rehabilitation after surgery avoid complications and prevent secondary muscle atrophy.

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

None reported.

Ambulatory anaesthesia

None reported.

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Obstetrical anaesthesia

There are previous reports using epidural, spinal, and general anaesthesia for elective caesarean section [13,19,20]. Close fetal monitoring during delivery is strongly recommended.

Literature and internet links

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