orphanaesthesia

Anaesthesia recommendations for patients suffering from

Kearns Sayre syndrome

Disease name: Kearns Sayre syndrome

ICD 10: H49.8

Synonyms: chronic progressive external ophthalmoplegia and myopathy, chronic progressive external ophthalmoplegia with ragged red fibers, CPEO with myopathy, CPEO with ragged red fibers, KSSS (Kearns Sayre Shy syndrome), mitochondrial cytopathy, occulocraniosomatic syndrome (absolute), ophthalmoplegia, pigmentary degeneration of the retina and cardiomyopathy, ophthalmoplegia plus syndrome

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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

Disease summary

Kearns-Sayre syndrome (KSS) is a rare mitochondrial myopathy caused by deletion of mitochondrial DNA. It is a disease with a wide continuum of phenotypes ranging from mild forms of ptosis to multisystemic disorders resulting in early death.

The original characterisation as presented by Kearns in 1958 comprised three core findings: 1. Chronic progressive external ophthalmoplegia [CPEO] caused by advancing weakness of the levator palpebrae, orbicularis oculi and other extra-ocular muscles. 2. Atypical retinitis pigmentosa ("Salt-and-pepper" fundus of depigmentation, hyperpigmentation and chronic inflammation). 3. Cardiac conduction disorders (AV-blockage, pre-excitation syndromes).

Disease onset is typically before the age of 20 and symptoms may appear as early as infancy. Other findings may include cerebellar ataxia, hearing loss, a wide range of neuro-endocrine (growth retardation) and gastrointestinal (intestinal dysmotility and gastroparesis) dysbalances, furthermore general muscle weakness including dilated cardiomyopathy and consecutive heart failure.

Deletion of mitochondrial DNA will impair oxidative/aerobic production of cellular energy and usually affect those organs more, which have intensive energy consumption such as the central nervous system or (cardiac) muscles. Defects are often distributed unevenly between cells, tissues and organs, and this "heteroplasmic pattern" of dysfunction explains for the variety of phenotypes. Diagnosis may be facilitated by genetic testing or muscle biopsy which can reveal so called "ragged red fibers" after trichrome staining.

Typical surgery

Ophtalmosurgery to correct ptosis.

Pacemaker insertion for brady-arrhythmias, defibrillators for pre-excitations syndromes and cardiomyopathies.

Type of anaesthesia

Literature describes both general and regional anaesthesia in patients with KSS.

However, interactions of anaesthetic agents and impaired cellular biochemistry in mitochondriopathies are only poorly understood.

Necessary additional diagnostic procedures (preoperative)

Anaesthesia management of the patient with KSS should begin with a thorough review of the patients history and previous investigations - preferably in consultation with the attending internist/paediatrician – in order to grasp the extend of impairment.

Anamnesis and physical examination must look for any hints of impairment of airway or respiratory muscles.

An ECG and echocardiogram are advisable to identify conduction disorders and cardiomyopathies.

Particular preparation for airway management

Weakness of pharyngeal muscles may result in difficult mask/bag-ventilation.

Gastroparesis may be present and a rapid-sequence-induction may be indicated.

Particular preparation for transfusion or administration of blood products

Transfusion triggers must be reconsidered individually in patients with mitochondriopathies.

Particular preparation for anticoagulation

n/a

Particular precautions for positioning, transport or mobilisation

n/a

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

n/a

Anaesthesiologic procedure

Peri-operative fasting should be minimized to avoid katabolic stress. Normoglycaemia and normovolaemia should be checked and maintained, e.g. via supplementary intravenous glucose and infusions. But beware: Lactate from infusions may overload the oxidative cellular capacity! Elevated levels of pyruvate and lactate may indicate increased anaerobic metabolism.

Adequate oxygen delivery, normothermia and stable cardiovascular functions during anaesthesia are imperative. Adequate anxiolysis and analgesia will help avoid increased energy needs.

General anaesthesia has been described repeatedly in literature. However, patients may require smaller doses of agents – for induction and maintenance of narcosis.

The clinical relevance of the effects even of single doses of propofol on mitochondria (e.g. propofol infusion syndrome (PRIS)) is not clear. Literature suggests the avoidance of this substance due to a variety of alternatives.

In cases of muscle weakness, reduced doses of muscle relaxants should be considered. Otherwise, the use of rocuronium and subsequent reversal with sugammadex seems highly beneficial.

Always exclude known triggers of rhabdomyolysis. There is no known direct link between KSS and malignant hyperthermia (MH) per se, but severe cases of MH-like rhabdomyolysis have been described in patients with mitochondriopathies.

In cases of respiratory impairment or cases that are suggestive of a difficult airway, general anaesthesia must be taken with great care. Local or (ultrasound guided!) regional anaesthesia should be considered.

The use of regional anaesthesia eliminates the risk of central nervous system depression, prolonged muscle relaxation, and the possibility of MH-like complications, and has the least impact on the patient's metabolic state. (Prilocaine should be avoided due to its haemoglobin oxidizing properties that may aggravate cellular energy depletion.)

Particular or additional monitoring

Monitoring of the neuromuscular blockade and temperature are advisable.

Possible complications

External pacing and/or defibrillation must be available for arrhythmic complications.

Postoperative care

Skeletal muscle weakness may compromise postoperative ventilation, especially after upper abdominal or thoracic surgery.

In cases of major surgery and/or increased duration/dosis of anaesthesia, post-operative surveillance in a ICU environment is advisable.

Information about emergency-like situations / Differential diagnostics

Beware of cardiac arrhythmias and cardiac failure in underdiagnosed patients!

Treat MH-like symptoms as if it was malignant hyperthermia!

Ambulatory anaesthesia

Ambulatory anaesthesia is usually not suitable for these patients. It can only be considered with minor procedures in very mildly affected individuals.

Obstetrical anaesthesia

KSS has equal prevalence in males and females. Fertility is not necessarily compromised and you may encounter KSS patients in obstetrics. However, literature does not provide any information for this setting.

Literature and internet links

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Last date of modification: June 2011

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