orphanaesthesia

Anesthesia recommendations for patients suffering from

Joubert syndrome

Disease name: Joubert syndrome

ICD 10: Q04.3

Synonyms: CPD IV, Cerebelloparenchymal disorder IV, Classic Joubert syndrome, Joubert syndrome type A, Pure Joubert syndrome, Vermis-Agenesia, Joubert-Boltshauser syndrome

The Joubert syndrome (JS) is a rare autosomal recessive disorder whose main clinical signs are hypotonia, ataxia, mental retardation, abnormal eye movements and a respiratory pattern of alternating hyperpnea-apnea.

Since its first description in 1969 more than 100 cases have been documented. The prevalence is estimated to be 1 in 100.000.

JS is characterized by partial or complete agenesis of the cerebellar vermis – the structure connecting both parts of the cerebellum. Furthermore other bordering parts of the cerebellum can be involved, too.

Clinical course shows abnormal breathing pattern (episodic tachypnea and/or apnea) and nystagmus with onset during neonatal period. The breathing pattern is characterized by effortless hyperventilation, which is more conspicuous in the awake state and intensifies after stimulation. Paroxysmal hyperventilation is often punctuated by intermittent central apnea. However, abnormal respiratory pattern is not a consistent finding and respiratory distress in the proper sense is not a feature. During infancy muscular hypotonia can be observed and later on cerebellar ataxia (staggering gait and imbalance) may develop. Delayed motor function development is common and cognitive function ranges from normal intelligence to severe deficits. Oculomotor apraxia and seizures may occur.

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

Physical characteristic are: large head, prominent forehead, high rounded eyebrows, epicanthal folds, an upturned nose with prominent nostrils, palate malformations, laryngomalacia, micrognathia, an open mouth (with oval, 'rhomboid' and finally triangular shape), tongue protrusion and rhythmic tongue motions, and occasionally low-set and tilted ears. Other features sometimes present in Joubert syndrome include retinal dystrophy, nephronophthisis, liver fibrosis and polydactyly.
Diagnosis is based on the main clinical features. These must be accompanied by the presence of a neuroradiological hallmark, designated as the “molar tooth sign'' (MTS) on magnetic resonance imaging (MRI). In view of the complex genetic heterogeneity mutations are only found in about 50% of patients at present.
Management is symptomatic and should be multidisciplinary, prognosis for mild or moderate forms is favorable, and therapy of severe forms should be carried out at a specialized reference centre.

Typical surgery

Oral and maxillofacial surgery: maxillary aberrations, dental malposition, dental extractions; Neurosurgery: ventricular shunts (rare); Ophthalmology: strabismus and retinal surgery; General surgery: implantation of peritoneal dialysis catheter, liver and renal transplantation due to loss of function. Orthopedic surgery: scoliosis surgery.

Type of anaesthesia

Up to now there are only some reports about anesthesia in patients with JS in the literature. Those were basically reported to be uneventful. Although volatile anesthetics are discussed to increase postoperative breathing problems, so far a recommendation for either volatile or intravenous anesthetic agents cannot be made.

Regional anesthesia is strongly recommended, but mostly in combination with sedation, because of the psychological disturbances of the patients.

This is especially true because emotional stress can trigger breathing difficulties like tachypnea or apnea in these patients.

Necessary additional diagnostic procedures (preoperative)

Routine preoperative testing depends on comorbidity of the patients. Mostly no particular tests are necessary, beside normal routine approach.

In case of renal or hepatic involvement renal and hepatic function has to be assessed.

Particular attention should be turned to medical history and physical examination. History of hyper-, tachy- and apnea in the past has to be evaluated. Furthermore tongue protrusion, small mouth opening and mobility of the cervical spine as signs for difficult intubation conditions have to be recognized.

Particular preparation for airway management

Airway management should not be a special problem in the majority of patients. If there are any physical signs for possible difficult intubation conditions, the usual precautions must be taken and common tools to secure the difficult airway should be held for immediate use.

Awake fiberoptic intubation is not recommended because of the often diminished or absent patient compliance. If necessary, fiberoptic intubation should be performed in the anesthetized patient.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

Not reported.

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

Not reported.

Anaesthesiologic procedure

Avoid premedication with drugs depressing respiratory function.

Propofol was used without any adverse effects. For volatile anesthetics possible prolongation of postoperative breathing problems has been discussed. Inhalational anesthesia induction can force apnea.

Depth of anesthesia can be monitored by using electroencephalographic based devices.

The use of long acting opioids should be avoided because of the depression of the respiratory function.

In case of immobilization succinylcholine should be avoided.

Non depolarizing muscle relaxants should be administered with caution or avoided, because of existing muscular hypotonia.

Regional or local anesthesia is strongly recommended for postoperative pain management and to avoid the administration of opioids. Because of the reduced or absent patient compliance, regional and local anesthesia has often to be combined with general anesthesia or sedation.

Clonidine may lead to an increasing incidence of apnea and therefore its use should be restricted for strong indications.

Caffeine and theophylline are discussed to be beneficial by reducing the incidence and severity of the apneic episodes. However, results seem to be rather limited.

Particular or additional monitoring

There are no reports for particular monitoring in patients with JS besides routine monitoring.

Monitoring of the depth of anesthesia using electroencephalographic based devices may be helpful.

Possible complications

Patients with JS are at risk for the need of difficult airway management, mainly because of macroglossia.

Hyper- and tachypnea and apneic episodes are common and may influence and prolong postoperative course.

Postoperative care

There is a need of postoperative monitoring of breathing frequency and oxygen saturation.

Duration of postoperative monitoring has to be discussed for every single case. It depends of the type of surgery, the need of postoperative pain therapy (especially if opioids have to be used) and the administered anesthetic drugs.

In some cases postoperative intensive care may be necessary, nevertheless in some cases ambulatory setting may also be possible.

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 diseases, e.g.:

Hyper- and tachypnea as well as apneic episodes are typical disease characteristics and possible side effects of anesthetics and opioids. Differentiation of the causative trigger may be difficult.

Ambulatory anaesthesia

Ambulatory anesthesia (according to common guidelines) is possible under certain circumstances. These include the avoidance of neuromuscular blocking agents and long acting opioids, the performance of a potent regional anesthesia, stable postoperative respiratory conditions and a competent home care.

Obstetrical anaesthesia

Up to now there are no reports about obstetrical anesthesia in patients with JS.

Literature and internet-links

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Last date of modification: January 2013

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