orphanaesthesia

Anaesthesia recommendations for

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-agenesis, Joubert- Boltshauser syndrome

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

First described in 1969, 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 the 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 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. Additional findings can include renal disease, ocular colobomas, occipital encephalocele, hepatic fibrosis, oral hamartomas, and endocrine abnormalities.

Physical characteristic are: Large head, prominent forehead, high rounded eyebrows, epicanthal folds, an upturned nose with prominent nostrils, palate malformations, laryngo-malacia, 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, 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 favourable, and therapy of severe forms should be carried out at a specialized reference centre.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

Typical surgery

Diagnostic procedure: MRI. 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. Orthopaedic surgery: scoliosis surgery.

Type of anaesthesia

Up to now, there are only some reports about anaesthesia in patients with JS in the literature. Those were basically reported to be uneventful. Although volatile anaesthetics and opioids are discussed to increase postoperative breathing problems, a recommendation for either volatile or intravenous anaesthetic agents cannot be made so far. Preferred are short-acting substances, the use of α-2-agonists is reported.

Regional anaesthesia 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 pre-operative testing (beside standard care)

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 have 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 anaesthetized patient.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Not reported.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Avoid premedication with drugs depressing respiratory function.

Propofol was used without any adverse effects. For volatile anaesthetics possible prolongation of postoperative breathing problems have been discussed. Inhalational anaesthesia induction can force apnea.

Depth of anaesthesia 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 and appropriate monitoring or be avoided because of existing muscular hypotonia.

For postoperative analgesia and to avoid opioids, local or regional anaesthesia is strongly recommended. This often has to be performed under (or combined with) general anaesthesia or sedation.

Clonidine may increase the incidence of apnoeic episodes and should only be used with appropriate monitoring. There are reports about the successful administration of dexmedetomidin.

Possibly caffeine or theophylline decrease the incidence of apnoeic episodes, but data is limited.

Particular or additional monitoring

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

Monitoring of the depth of anaesthesia 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.

Post-operative 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 on the type of surgery, the need of postoperative pain therapy (especially if opioids have to be used) and the administered anaesthetic drugs.

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

Disease-related acute problems and effect on anaesthesia and recovery

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

Ambulatory anaesthesia

Ambulatory anaesthesia (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 anaesthesia, stable postoperative respiratory conditions and a competent home care.

Obstetrical anaesthesia

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

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