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Stress cardiomyopathy

Sturge-Weber syndrome

orphanaesthesia

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

SUPPLEMENT NR. 15 | 2018

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

Sturge-Weber syndrome

Disease name: Sturge-Weber syndrome

ICD 10: Q85.8

Synonyms: Dmitri disease, encephalofacial angiomatosis, enchephalotrigeminal angiomatosis, fourth phacomatosis, leptomeningeal angiomatosis, meningeal capillary angiomatosis, Sturge-Kalischer-Weber syndrome, SWS, SWS type 1-facial and leptomeningeal angiomas, SWS type 2 - facial angioma alone - no CNS involvement, SWS type 3 - isolated leptomeningeal angiomas.

Sturge-Weber is one of the rare phacomatosis or neurocutaneous syndromes, which consists of abnormal capillary malformations that can involve the face, eyes and leptomeninges of the brain. The syndrome was first described by W.A. Sturge in 1969. It has been recently demonstrated by Shirely et al. that it is caused by a somatic activating mutation in guanine nucleotide-binding protein G(q) (GNAQ) in the majority of cases.

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

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Disease summary

The facial capillary angiomas are centered along the distribution of the V1 (ophthalmic), V2 (maxillary) and V3 (mandibular) branches of the trigeminal nerve and, if present in an infant or child presenting with seizures, a diagnosis of SWS should be considered. It should be noted that SWS may be present in patients without any facial angiomas and that not all patients with facial angiomas have SWS.

Central neuroaxial imaging may reveal characteristic angiomas along with calcification of the leptomeninges ipsilateral to the facial naevus. These may lead to atrophy of the cerebral cortex along with variable neurological and cognitive impairment.

Characteristically, the main ocular manifestations include glaucoma, varicosities of the retinal vessels, haemangioma of the choroid and retinal detachment. Optic neuropathy and bubphthalmos secondary to the raised intraocular pressure can occur in untreated cases of raised IOP. Occipital brain involvement is possible. These may lead to varied visual field defects and even blindness.

Clinical features include seizures, which may be generalised or focal in origin, most often occurring contralateral to the facial naevus. Developmental delay and cognitive impairment, along with headache, stroke-like events, hemiparesis and hemicerebral atrophy may be present. These may occur secondary to the ischaemic and destructive effect of cerebral angiomas and associated seizures. Facial angiomas may vary in colour from light pink and flat to dark purple and raised. Cardiac lesions, which may be occasionally associated with Sturge-Weber Syndrome, include septal defects, valvular anomalies, transposition of the great vessels, aortic coarctation and rarely deep arteriovenous malformations.

The mainstay of treatment is seizure control. Seizures may worsen any associated cortical hypoperfusion with the potential to further impair both neurological and developmental delay. Intraocular pressure reduction in glaucoma can sometimes be achieved with both carbonic anhydrase inhibitors and beta-blockers, however, surgery may be required to control the elevate eye pressure. Facial naevi can be treated with laser to varying degrees of results.

Typical surgery

Caesarean section, dental procedures, ocular surgery, examination under anaesthesia or glaucoma surgery such as trabulectomy or trabulotomy, oral surgery for the removal of friable haemangiomas, hemispherectomy, focal cortical resection (typically of the posterior quadrant), callosotomy or surgical ablation of the affected cerebral hemisphere for seizure control, laser therapy for facial naevus formation have all been described.

Type of anaesthesia

Patients with Sturge-Weber syndrome tolerate anaesthesia well. Initial management includes appropriate investigations for associated anomalies, as the varied presentation and extent of the disease process from pure cutaneous manifestations to extensive systemic and airway involvement.

Regional anaesthesia: Poor patient compliance and the presence of existing neurologic deficits may preclude the use of central neuroaxial blockade. Neuraxial anaesthesia is not specifically contraindicated unless other contraindications exist. It is also important to note that there is the theoretical risk of spinal cord angiomas which may be present in possible overlap syndromes such as with Klippel-Trénaunay syndrome and if these are suspected, neuroimaging may be appropriate.

General anaesthesia: From an airway point of view, facial asymmetry, large tongues and lips may make mask ventilation difficult. Friable airway haemangiomas may engorge during stressful times and may bleed easily if traumatised, potentially further obscuring laryngoscopic view. Intracranial pressure may also be further raised from baseline by not only laryngoscopy itself, but multiple events from induction to emergence, which could theoretically lead to angioma rupture as these abnormal vessels are not only thin-walled, but also may have impaired autoregulation. Low blood pressure is thought to increase the risk of stroke in patients with impaired brain perfusion It is important, therefore, to maintain perioperative BP within preoperative normal limits. It should also be noted that if large cerebral AV shunts are present, this could lead to a prolonged inhalational induction time due to the associated increased cardiac output.

Necessary additional diagnostic procedures (preoperative)

Patients with Sturge-Weber syndrome should be evaluated for any associated airway, cardiac and CNS anomalies.

Airway anomalies and their investigation are described in the next section.

Cardiac anomalies are rare in SWS. Cardiac investigations such as an ECG and ECHO may be required to look for possible cardiomegaly and cardiac failure if clinical suspicion exists. These may occur secondary to the cardiac anomalies associated with Sturge-Weber syndrome, which include septal defects, valvular anomalies, transposition of the great vessels and aortic coarctation. Arteriovenous shunting from large arteriovenous angiomas in the skin, subcutaneous tissues, muscle and intracerebral haemangiomas may lead to a high-output cardiac failure. An ECG may also help elucidate any cardiac conduction defects secondary to possible elevated levels of anticonvulsants, e.g. phenytoin. GNAQ polymorphisms have been linked with increased myocardial injury in female patients undergoing cardiac bypass.

Contrast CT and MRI of the central neuroaxis may be used to demonstrate, if present, any of the characteristic cerebral features of angiomas. These include enlarged choroid plexi, cortical atrophy underlying the angioma, calcification and abnormal draining of medullary and subependymal veins. Neuroaxial imaging may also be useful to demonstrate any associated raised ICP or potential spinal leptomeningeal angiomas. While spinal leptomeningeal angiomas have not been reported in Sturge-Weber syndrome to date, there is always the potential for overlap syndromes, for example, Klippel- Trénaunay syndrome, so caution would be recommended prior to performing central neuroaxial blockade.

Laboratory testing should include blood levels of anticonvulsants to ensure levels that are within the therapeutic range. A full blood count may help exclude any possible bone marrow depression secondary to anticonvulsant therapy. A platelet count is also important in light of the increased frequency of thrombosis and or bleeding. Patients with heart failure on diuretic or digoxin therapy should also have their electrolytes checked.

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Particular preparation for airway management

In preparation for a potentially unidentified difficult airway, a variety of facemask sizes, endotracheal tubes, laryngoscopy blades and handles should be prepared. The use of video laryngoscopes and fibre-optic intubation may help prevent injury to any airway angiomas. There should also be access to a difficult airway trolley for alternative oxygenation strategies should the need arise.

Emotional stress with its potential haemodynamic effects can cause swelling of angiomas with increased risk of perioperative bleeding. This can be partially avoided by ensuring a good preoperative rapport with the patient, and with adequate premedication.

Facial asymmetry may make head positioning, mask ventilation and intubation difficult. It is important to take time to adequately position the patient into the optimal position using pillows and or blankets, if required.

If examination or imaging is suggestive of airway angiomas, consultation with ear, nose and throat specialists may be useful for further elucidation of lesions by upper airway endoscopy. These angiomas may involve the mucous membranes of the mouth including the lips, gingiva, tongue or palate and may also be present as far down as the larynx and trachea. These may not only directly affect mask ventilation or visualisation of the vocal cords, but they can also bleed if traumatised during any airway manipulation further affecting laryngoscopic view.

Careful and gentle tracheal intubation should be performed to minimise both the hypertensive response and rise in ICP that can potentially lead to rupture of any cerebral angiomas. These haemodynamic effects can be minimised by means of gentle laryngoscopy, adequate depth of anaesthesia along with the use of an appropriate opioid dose and adequate vocal cord topicalisation. Intubation should be performed with well-lubricated endotracheal tubes. Nasal intubation should only be performed once nasopharyngeal angioma involvement has been excluded.

Particular preparation for transfusion or administration of blood products

Significant blood loss and difficulty with haemostasis can be encountered during excision or resection of any vascular malformations. Depending on the proposed surgery, type and screen should be performed with a crossmatch required for any surgery at higher risk of bleeding.

Particular preparation for anticoagulation

Recurrent thrombotic episodes within the abnormal angiomas may require use of antiplatelet agents. Antiplatelet therapy may impact further on perioperative haemostasis and should be taken into account when planning surgery. Aspirin use is increasingly common in patients with this condition.

Particular precautions for positioning, transport or mobilisation

None identified.

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

The mainstay of treatment in Sturge-Weber syndrome involves the prevention of seizures with anticonvulsants. These have the potential to affect enzyme metabolic pathways, which could subsequently affect the metabolism of the anaesthetic agents used. Conversely, certain anaesthetic agents themselves may induce hepatic enzymes and lead to lowering of serum anticonvulsant medications in the postoperative period below therapeutic levels.

Anaesthesiologic procedure

Adequate premedication, in addition to anxiolysis, may help with seizure prophylaxis, but may also allow the placement of an intravenous line prior to induction of anaesthesia in cognitively impaired children. It should be noted that ketamine, which may both lower the seizure threshold and raise the ICP should be avoided, if possible.

Induction and emergence from anaesthesia should be as smooth as possible as any light plane of anaesthesia; straining, coughing or even laryngoscopy itself may increase ICP, or blood pressure theoretically risking angioma rupture due to their abnormal autoregulation. Blood pressure should also be well controlled throughout the perioperative period.

Inhalational induction may be preferred to intravenous inductions for older children, those with facial deformities or in any patient in whom a difficult airway is considered. Intravenous induction may be preferred in cognitively impaired children or those with seizures difficult to control.

The use of video laryngoscopes and/or fibre-optic intubation may help navigate carefully around airway angiomas and may be considered the safer option in a potentially difficult airway if the patient is compliant.

Suxamethonium administration may result in hyperkalaemia in patients with long-standing denervation lesions such as hemiplegia.

The location of any neuromuscular monitoring should take into account existing neurological deficits as misleading resistance to non-depolarising muscle relaxants and may be observed.

Endocarditis prophylaxis should be considered according to current guidelines.

Particular attention should be paid to hydration as dehydration may increase the risk of intravascular thrombosis.

Particular or additional monitoring

None identified.

Possible complications

Complications can be grouped into airway, circulatory, CNS and metabolic complications.

Airway complications as discussed above can include problematic mask ventilation, hypocarbia, intubation difficulties or bleeding into the airway from angioma damage.

Perioperative sleep deprivation, hyperthermia, electrolyte disturbance, hypoxia, hypoglycaemia, and hypotension may precipitate seizures and should be avoided. Preparation should be made to treat seizures if they occur.

Vascular insufficiency might occur in organs such as the pituitary, thymus, lung, liver, spleen or pancreas, where other angiomas may be present. This may lead to secondary metabolic and associated clinical disorders.

Kossoff described outcomes for 32 patients following hemispherectomy. 47% experienced immediate post-operative complications which consisted of bleeding (4), infection (4), and re-operation was required in a further 3 patients because of seizures (1), hypertension (1) and shunt (1). Perioperative deaths due to uncontrollable bleeding from diploic veins during burr hole creation, hyperkalemia from blood products, postoperative fluid shift and autonomic instability have been described after neurosurgical procedures.

Postoperative care

Seizure prophylaxis is of vital importance. As stated above, perioperative sleep deprivation, electrolyte disturbance, hypoxia, hypocarbia, hypoglycaemia, and hypotension should be avoided as abnormalities here may precipitate seizures. It is important to ensure that oral diet along with regular anticonvulsant medication are resumed as soon possible in the postoperative period. Patients in whom oral intake cannot be established, consultation with a neurologist may be required in order to adequately titrate nasogastric, rectal or even intravenous anticonvulsants. Hydration should be maintained.

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.

Seizures may be related to underlying cerebral pathology. Seizures may also be precipitated by any of the factors mentioned above. Potential for cerebral herniation in patients undergoing neuraxial anaesthesia with concomitant raised ICP should also be considered.

Ambulatory anaesthesia

None identified.

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Obstetrical anaesthesia

Any parturient with port wine naevus in the ophthalmic division of the trigeminal nerve should have a contrast MRI to rule out any cerebral angioma. This will be especially important if the patient has a history of seizure disorders as there is the potential for angioma rupture during labour secondary to both the hypertensive pain response and Valsalva manoeuvres during labour.

While regional anaesthesia is safe in the majority of cases neuroimaging assessment will help clarify any possible concerns to central neuroaxial blockade such as raised ICP or vertebral canal angioma. As stated above, there should be awareness to the possibility of inadvertent dural puncture and potential cerebral herniation in those with raised ICP when performing epidural anaesthesia. An excessive absorption or intravenous injection of local anaesthetic may also provoke seizures. It should be noted that pre-existing neurological deficits of the spinal cord or peripheral nerves is a relative contraindication to central neuroaxial blockade.

Regional anaesthesia shall be preferred to general anaesthesia for caesarean section if time permits, ICP is normal and the patient agrees.

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Please note that this guideline has not been reviewed by an anaesthesiologist but by two disease experts instead.

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