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Jarcho-Levin syndrome

orphananesthesia

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

SUPPLEMENT NR. 4 | 2024

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 Patientinnen und Patienten mit seltenen Erkrankungen. Damit will OrphanAnesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patientinnen und 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 Anästhesistinnen und Anästhesisten damit keine Erfahrungen gesammelt haben, sodass 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 eine Anästhesistin bzw. ein Anästhesist sowie eine weitere Krankheitsexpertin bzw. ein weiterer Krankheitsexperte (z. B. Pädiaterin bzw. Pädiater oder Neurologin bzw. Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, sodass 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:

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OrphanAnesthesia –
a 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.

Find a survey of the recommendations published until now on:

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orphananesthesia

Anaesthesia recommendations for

Jarcho-Levin syndrome

Disease name: Jarcho-Levin syndrome

ICD 10: Q76

Synonyms: Spondylocostal dysostosis

Disease summary: Jarcho-Levin syndrome (JLS) is an inherited costovertebral dysplasia. It is inherited in both an autosomal and a recessive pattern [1]. Saul Jarcho and Paul M Levin first described this disorder in 1938 [2]. Global incidence of JLS is 1/40,000 births which makes it a rare genetic disorder [1]. It is related to the SSRT/ARS2 (Serrate RNA effector molecule homolog or arsenite-resistance protein 2 gene. It is caused by a mutation in DLL3 gene mapped on 19q13 sequence [3]. The intelligence levels (IQ) remained normal in all reported cases. Its exact incidence in the Asian continent is unknown, but a few cases of JLS have been reported in India [4]. JLS has two phenotypes: Spondylothoracic dysplasia (STD), which is autosomal recessive and Spondylocostal dysplasia/dysostosis (SCD), which is autosomal dominant, both being associated with multiple other anomalies. Both types are due to a failure of vertebral segmentation. Infants usually present with vertebral and rib anomalies along with a plethora of non-skeletal abnormalities including hydrocephalus, neural tube defects, tracheal abnormalities, cardiac, renal, gastrointestinal and urinary tract abnormalities [4,5].

Axial skeleton growth defects manifest in the form of vertebral column malformations, intrinsic rib anomalies, small thoracic cavity, short stature, scoliosis and kyphosis, leading to a protuberant abdomen and severe pulmonary complications [6].

Respiratory insufficiency, pulmonary hypertension and other multisystem anomalies are the major causes of mortality in these patients.

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

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Typical surgery

Patients with JLS generally present in infancy with neurological problems which may require neurosurgical procedures (emergency or elective), like ventriculoperitoneal (VP) shunt insertions. Thorough pre-operative evaluation should be done, with detailed systemic examination for multi-system afflictions of this rare syndrome.

They may also present for thoracospinal corrective surgeries, performed as staged operations in prone position with associated complications.

They can also present for orthopaedic correction surgeries, requiring repeated anaesthesia exposures. Rarely, they can also present for obstetric anaesthesia if there is uneventful survival till adulthood.

Type of anaesthesia

General anaesthesia is most commonly employed in these children. All considerations for neonatal/paediatric anaesthesia are required, including the possibility of difficult airway and respiratory dysfunction. Controlled mechanical ventilation is usually preferred for adequate oxygenation. Central neuraxial anaesthesia is difficult due to spinal abnormalities and not recommended. Ultrasound-guided nerve blocks can be given to supplement anaesthesia for perioperative analgesia.

Necessary additional pre-operative testing (beside standard care)

Besides standard pre-anaesthetic investigations, these children will need neurological workup, including magnetic resonance imaging (MRI) of the brain and spine. Cardiac and abdominal evaluation may be required in children with extensive multisystem disease. However, emergency procedures may not allow for adequate time for detailed assessments.

Chest x-ray films may show crowding of ribs in a crab-like pattern and dextrocardia. Advanced radiological investigations like MRI of the spine and the brain may reveal fusion defects in the lower back and upper trunk region with herniation of nerve roots, Arnold Chiari malformation and/or chronic hydrocephalus [7].

Particular preparation for airway management

Children with JLS have a difficult airway due to skeletal growth defects, hydrocephalus and craniofacial abnormalities. In addition, due to severe scoliosis and kyphosis, they have minimal respiratory reserves and increased risk of hypoxia during apnoeia episodes. In addition, airway access can be challenging because of the thoracic lordosis and the compensatory cervical kyphosis that may make intubation very challenging even with proper positioning. Difficult airway cart needs to be prepared beforehand. A silicon or cotton donut with padding needs to be used to position the patient supine on operation table prior to induction for accommodating the meningomyelocele sac. Paediatric video laryngoscopes and fibre-optic bronchoscopes must be available. Adequate pre-oxygenation and presence of expert anaesthesiologist help are needed. Standard difficult airway guidelines and algorithms must be followed to ensure patient safety.

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Particular preparation for transfusion or administration of blood products

There are no specific recommendations for the transfusion of blood or blood products. Adequate, cross-matched blood must be arranged in case of spinal corrective procedures.

Particular preparation for anticoagulation

There are no particular recommendations for anticoagulation in these patients. A coagulation profile may be done before major surgery in these children.

Particular precautions for positioning, transportation and mobilisation

Children with JLS have multiple deformities, hence positioning needs to be done carefully to avoid injuries. Padding of pressure points is mandatory to prevent neuropathies and pressure sores. Particular positioning difficulties can arise in the presence of meningomyelocele, where donuts (silicon or cotton) are used to cushion the posterior swelling during induction, to prevent their rupture.

Interactions of chronic disease and anaesthesia medications

There have been no reports in the literature on interactions between chronic diseases and anaesthetic medications in JLS. Nevertheless, it is better to avoid histamine-releasing agents.

Anaesthetic procedure

Due to distorted airway anatomy, difficult positioning and recurrent infections, the challenges of anaesthetic management are manifold. Anaesthetic management of any JLS case is challenging for the anaesthesiologist due to the distorted anatomy of the patient's rib cage, a high incidence of lower respiratory tract infections and anomalies of the tracheal and bronchial lumen (e.g., missing tracheal rings, narrow bronchial lumen, wider carina, short neck and small or distorted thorax) [8].

If dextrocardia [9] is present, one must remember to place the precordial stethoscope on the right chest for auscultation, not on the left side as usual. Oto-rhino-laryngology and paediatric consultation must be obtained to rule out other congenital anomalies. A difficult airway cart must be prepared preoperatively for both induction and extubation. The airway of a JLS patient is very sensitive to infection because of its distorted anatomy and the high risk of aspiration. Intubation in JLS patients should always be considered difficult and all necessary arrangements should be made prior to taking the patient on the table. Positioning such children for induction and surgery might also pose great difficulties. Gentle handling of these children especially during laryngoscopy is warranted as is diligent postoperative monitoring. Care and padding of spinal deformities and the meningomyelocele sac is advisable.

Controlled mechanical ventilation under general anaesthesia is usually recommended, especially in case of paediatric patients undergoing spinal or neurosurgery. In addition,

ultrasound-guided nerve blocks can be given by trained anaesthesiologists in the scope of perioperative pain management.

Particular or additional monitoring

Standard ASA (American Society of Anesthesiologists) monitoring must be applied to all patients. In addition, neuromuscular monitoring, invasive arterial and central venous monitoring line can be used in major surgeries and corrective spinal procedures. Arterial blood gas measurements can be monitored serially to enable correction of metabolic and electrolyte derangement. Temperature monitoring is mandatory, especially in paediatric patients. A strict intake-output charting with hourly urine output monitoring is recommended.

Possible complications

The presence of vertebral, rib and tracheal anomalies often results to an increased incidence of difficult airway and post-operative respiratory complications. Affected patients may require post-operative mechanical ventilation, a prolonged ICU stay and attendant complications. Neurological problems in JLS patients can have a protracted course. The longest survival of a reported case of JLS is 33 years [9].

Paediatric patients with JLS undergoing spinal, neurosurgical and orthopaedic procedures may have multisystem disorders like respiratory failure, cardiovascular complications, renal dysfunction, problems due to blood transfusion and metabolic derangements. Other concerns like perioperative laryngospasm and bronchospasm are common due to airway anomalies and frequent respiratory tract infections in these children.

Post-operative care

JLS patients of will generally require post-operative ICU care and continued monitoring post-operatively. Post-operative care can be protracted due to prolonged post-operative mechanical ventilation and the management of other multisystem abnormalities. Padding of pressure points is required to prevent pressure sores. Care of invasive monitoring lines needs to be continued in the ICU along with diligent monitoring.

Disease-related acute problems and effect on anaesthesia and recovery

Disease-related acute problems in JLS patients include distorted airway anatomy resulting in a difficult airway and “cannot-ventilate, cannot-intubate” (CVCI) situation; frequent respiratory tract infections leading to increased incidences of laryngospasm and bronchospasm; rib, chest wall and vertebral anomalies causing prolonged mechanical ventilation, hypoxia and metabolic derangements; dextrocardia or other cardiac anomalies resulting in difficulty in cardiovascular monitoring and arrhythmias; spinal abnormalities resulting in neurological problems, stunted growth, pressure ulcers and repeated surgical/anaesthetic exposures. Anaesthesia can be quite challenging in these children and recovery can be delayed.

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Ambulatory anaesthesia

It is best to avoid day-care or ambulatory surgery in paediatric patients with JLS, as there is a possibility of severe perioperative complications due to multisystem involvement, restrictive lung disease and neurological defects. There can also be incidences of post-operative apnoeas in preterm children with JLS.

Obstetrical anaesthesia

There has been some recent literature [10] regarding obstetric anaesthesia in patients with JLS. Though there have been no studies regarding fertility in these patients, pregnancy may be possible if patient survives till reproductive age. There are several challenges for obstetric anaesthesia in them. All standard precautions, monitoring and perioperative care concerns with detailed pre-operative evaluation must be expedited for a safe maternal and foetal outcome. The difficult airway risk associated with the obstetric patient is accentuated in patients with JLS and a difficult airway cart needs to be kept ready in all these cases. In view of vertebral anomalies, a central neuraxial blockade may be difficult to perform for operative deliveries. Aspiration prophylaxis, adequate pre-oxygenation, meticulous monitoring, expert airway manger, post-operative ICU care and an alternative plan of anaesthesia should be expedited on a case-to-case basis.

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