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Pierre Robin sequence

Schwartz-Jampel syndrome

orphanaesthesia

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

SUPPLEMENT NR. 11 | 2023

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:

www.ai-online.info/Orphsuppl
www.orphananesthesia.eu

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

Pierre Robin sequence

Disease name: Pierre Robin sequence

ICD 10: Q87.0

Synonyms: Pierre Robin syndrome, Robin syndrome, anomalad, complex, deformity, triad

Disease summary: A diagnosis of Pierre Robin sequence (PRS) is established when a patient exhibits the three clinical hallmarks of micrognathia (small mandible), glossoptosis (backward downward displacement of the tongue base) and airway obstruction present from birth. Cleft palate commonly occurs, but is not a prerequisite for a diagnosis. Pierre Robin sequence may be isolated (20–40 %) or associated with a syndrome, the commonest being Stickler, foetal alcohol, Treacher-Collins and velocardiofacial syndrome. The anatomical features cause a variable degree of airway obstruction and patients may present with stridor, respiratory distress, cyanosis and signs of obstructive sleep apnoea (OSA). Patients may also exhibit other airway pathology such as laryngomalacia and subglottic stenosis. Patients are at risk of inadequate nutrition, aspiration and gastro oesophageal reflux disease. Incidence varies between 1:5,000 to 1:85,000, the range a reflection of the variable clinical presentation.

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

Flexible nasolaryngoscopy and direct rigid laryngobronchoscopy are performed to assess the patient's airway anatomy and degree of obstruction. The ease of orotracheal intubation can be performed during the assessment.

Tracheostomy placement in patients with a critical airway typically in syndromic PRS with multilevel obstruction and children < 2 kg who may be too small for surgical intervention.

Tongue-lip adhesion (glossopexy) is performed in the neonatal period and aims to correct the glossoptosis, relieve airway obstruction and give time for the mandible to grow. Surgical attachment of the tongue to the lingual surface of the lower lip is performed. Patients are kept sedated and paralysed on paediatric intensive care unit (PICU) to minimise dehiscence for a number of days before being woken and extubated. The sutures remain until a year old and removal can coincide with cleft surgery during the same period.

Mandibular distraction osteogenesis aims to elongate the mandible and move the tongue forward thus relieving the airway obstruction. Bilateral osteotomies are performed and a distraction device is applied. The patient may remain sedated and paralysed on PICU for up to a week according to local protocol. Once extubated the device will remain in for a number of weeks to allow consolidation of new bone.

Cleft palate surgery.

Dental surgery.

A proportion of patients with PRS require a gastrostomy feeding tube and refractory reflux disease may require a Nissen's Fundoplication.

Anaesthesia for radiological computed tomography (CT) and magnetic resonance imaging (MRI).

Type of anaesthesia

Sevoflurane is one of the most commonly used anaesthetic volatile agents, widely used in paediatrics, and is recommended if there is uncertainty about the ease of airway management. Patients with PRS may exhibit opioid sensitivity secondary to chronic airway obstruction and hypoxia.

Alpha-2 agonists such as dexmedetomidine or clonidine provide analgesia and may reduce opioid requirements.

Regional anaesthesia can be utilised as an opiate sparing modality and in older patients may be considered as an alternative to general anaesthesia. Inferior alveolar and infra-orbital nerve blocks have been performed for postoperative pain control after mandibular distraction and cleft surgery commonly performed by oral surgeons.

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Necessary additional preoperative testing (beside standard care)

History of delivery, initial APGAR scores, frequency of apnoea and cyanotic episodes will provide information on the severity of airway and respiratory compromise.

The degree of feeding difficulties correlates with the severity of airway obstruction. Aspiration is common in patients with severe gastric reflux disease and should be appropriately treated prior to elective surgery.

Physical examination focused on airway and respiratory signs and the presence of associated features.

If breathing is stable whilst sitting, awake flexible nasolaryngoscopy may be performed by ear, nose and throat surgeons. This will provide useful information about airway anatomy and the level and severity of oropharyngeal obstruction.

Review of CT and MRI images will provide information on airway anatomy and should include evaluation of the cervical spine. Patients with Stickler syndrome are at increased risk of cervical spine instability, however, there have been a small number of published cases of occipito-atlanto-axial instability in children with PRS who present with musculoskeletal abnormalities such as skeletal dysplasia.

Polysomnography will measure the Apnoea Hypopnea Index (AHI) in patients with OSA. AHI >10 indicates severe OSA. The presence of central apnoeas indicates a neurological component, which increases the risk for requiring a tracheostomy. Severity of OSA correlates with intraoperative and postoperative respiratory complications.

Echocardiography is required for patients with syndromic Pierre Robin Sequence to exclude associated cardiac anomalies. Patients with Treacher-Collins and velocardiofacial syndrome commonly have a congenital heart disease. Stickler syndrome patients have a marfanoid appearance with joint laxity and may have a mitral valve prolapse. Patients with fetal alcohol syndrome may have a ventricular septal defect.

Particular preparation for airway management

Airway obstruction may occur at multiple levels and under anaesthesia the patients may be difficult to ventilate, oxygenate and intubate. Anaesthesia should be undertaken by anaesthetists experienced in difficult airway management with airway surgeons and theatre teams available to assist when required. Preparation for a difficult airway is vital with appropriate equipment checked and ready to use. Difficult airway trolleys with standardised equipment should be located in close proximity. The Difficult Airway Society has recommended an equipment list and provides guidelines for a standardised setup:

https://das.uk.com/content/difficult_airway_trolley

These include a bag-mask ventilation, facemasks, oropharyngeal airways, nasopharyngeal airways, supraglottic devices (laryngeal mask airway (LMA), intubating LMA), endotracheal tubes, a flexible intubating fibrescope with adjuncts, bougies, airway exchange catheter, alternative laryngoscopes (e.g. straight blade), video-laryngoscopes, surgical cricothyroidotomy set, cannula cricothyroidotomy set with ventilation equipment.

Patients who obstruct when supine will be more difficult to mask ventilate and are likely to require airway adjuncts such as oropharyngeal airways, nasopharyngeal airways or a LMA.

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There is a growing body of evidence that advocates optimal intubating conditions with use of muscle paralysis during laryngoscopy and forms part of many difficult airway guidelines. Having said this, in paediatric patients with critical airway pathology, maintaining spontaneous ventilation and attempting laryngoscopy under deep anaesthesia may be the favoured technique.

Airway techniques described include the use of a fibre optic scope, LMA, retrograde wires, Glidescope, Shikani scope, Airtraq, Air-Q scope to accompany the traditional laryngoscope. Combination techniques have also been described such as passing a fibre optic scope through an LMA or in combination with a video-laryngoscope.

Extubation should be performed awake, and consideration given as to whether or not a nasopharyngeal airway (NPA) should be inserted prior to extubation to minimise the risk of postoperative airway obstruction.

Particular preparation for transfusion or administration of blood products

Velocardiofacial syndrome patients may have T-cell immune deficiency and require irradiated blood.

Particular preparation for anticoagulation

Prophylactic tranexamic acid is commonly administered in craniofacial surgery and in some centres for cleft surgery according to local protocol.

Particular precautions for positioning, transportation and mobilisation

Additional care with transfer and handling for patients with PRS, in particular with Stickler syndrome who exhibit joint laxity and marfanoid features. Cervical spine instability has been described in patients with Stickler syndrome whilst rare in patients with PRS with associated musculoskeletal abnormalities which have led to significant cord damage.

Interactions of chronic disease and anaesthesia medications

None reported.

Anaesthetic procedure

Induction has been performed with both volatiles and intravenous propofol. Commonly the patient is spontaneously ventilating until intravenous access is secured and the chosen airway technique employed. Rigid laryngobronchoscopy is undertaken with the patient breathing and anaesthesia being maintained using a volatile anaesthetic or with intravenous propofol and a short-acting opioid. The larynx is topicalised with local anaesthetic to reduce the risk laryngospasm. Where the trachea is to be intubated, a muscle relaxant may be required or alternatively a deep plain or anaesthesia with the patient self-ventilating.

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Rocuronium or vecuronium is preferred when sugammadex is readily available for emergency reversal. Simple analgesics can be used and the choice of opioid is dependent on the extent of surgery and risk of postoperative airway compromise and apnoea. Alpha-2 agonists have been used as opiate sparing analgesics intraoperatively. Neuromuscular block monitoring is essential with full reversal prior to an awake extubation.

Particular or additional monitoring

Full monitoring including pulse oximetry, ECG, non-invasive blood pressure and capnography. Transcutaneous CO_2 can be utilised during rigid laryngobronchoscopy when end-tidal CO_2 cannot be monitored. Capnomasks have been used in the recovery room to aid breathing assessments by recovery nursing staff. Apnoea monitoring should be available for postoperative patients at risk of apnoeas.

Possible complications

Airway obstruction can occur in the preoperative period and can be managed by placing a NPA and by placing the patient prone, which allows displacement of the tongue. Airway obstruction can also occur during induction of anaesthesia and airway adjuncts should be readily available.

Laryngoscopy and tracheal intubation may be very difficult. Intubation techniques should be utilised according to local expert practice. A paediatric otolaryngologist should be available to perform rigid bronchoscopy and ventilation or to perform an emergency surgical tracheostomy should it be required.

The presence of gastro-oesophageal reflux disease may increase the risk of aspiration. Patients should be fully fasted, on prophylactic gastric protection and be extubated awake.

Postoperative care

Patients undergoing tongue lip adhesion or mandibular distraction osteogenesis may require sedation and paralysis on the PICU according to local practice

A nasopharyngeal airway should be considered for patients who are extubated to reduce the risk of postoperative airway obstruction.

Patients who required non-invasive ventilation prior to surgery for obstructive sleep apnoea may need their ventilation to be restarted in the recovery area of the operating room.

Apnoea monitoring should be available for patients with opiate sensitivity or who have a history of obstructive or central apnoea.

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Disease-related acute problems and effect on anaesthesia and recovery

DAS and the Association of Paediatric Anaesthetists have developed algorithms, which provide guidance for anaesthetising patients with difficult airways and in whom oxygenation, ventilation and intubation are difficult.

1) Paediatric Difficult Airway Guidelines:
https://das.uk.com/files/APA1-DiffMaskVent-FINAL.pdf

2) Paediatric Difficult Mask Ventilation Guidelines:
https://das.uk.com/files/APA2-UnantDiffTracInt-FINAL.pdf

3) Paediatric Unanticipated Difficult Intubation Guidelines:
https://das.uk.com/files/APA3-CICV-FINAL.pdf

4) Paediatric 'Can't Intubate Can't Ventilate':
https://das.uk.com/files/APA3-CICV-FINAL.pdf

Ambulatory anaesthesia

No published case reports.

Obstetrical anaesthesia

No published case reports.

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