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Transverse myelitis

Treacher-Collins syndrome

orphan**anesthesia**

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

SUPPLEMENT NR. 14 | 2022

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 OrphanAnesthesia 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.

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 recommendations 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.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

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orphananesthesia

Anaesthesia recommendations for Treacher-Collins syndrome

Disease name: Treacher-Collins syndrome

ICD 10: Q75.4

Synonyms: Mandibulofacial dysostosis, Franceschetti-Zwahlen-Klein syndrome

Disease summary: Treacher Collins (TCS) syndrome is a rare disorder of craniofacial development with an incidence of approximately 1:50,000 live births resulting from mutations in the TCOF1 gene [1]. Although Thomson [2] and Berry [3] were probably the first to describe the signs now associated with this syndrome, it was Dr Edward Treacher Collins, an English surgeon and ophthalmologist, who first characterised the condition in 1900 [4,5]. It exhibits autosomal dominant inheritance with variable penetrance. About 60 % of the cases arise from fresh mutations in TCOF 1 gene with no previous family history [1]. It is a disorder of neural crest cell proliferation, involving the first and second branchial arches. It is thus bilateral, symmetrical and restricted to the head and neck region of the body. Clinical features and severity are variable.

Features relevant to anaesthesia and intubation include bony hypoplasia involving maxillary, zygomatic, and mandibular bones, small oral aperture, high arched palate and severe temporomandibular joint abnormalities. Other findings include predominant hypoplasia of soft tissues observed in the malar bone, inferior orbital rim and cheek, antimongoloid obliquity of the palpebral fissures, a coloboma of the outer third of the lower eyelids with absence of eyelashes, external ear abnormalities including anotia with atresia of the external auditory canal, and anomalies of the ossicular chain. Facial retrognathia or micrognathia is also commonly associated. Cleft palate is present in up to 35 % of patients and an additional 30–40 % have congenital palatopharyngeal incompetence. Chronic respiratory deficiency, obstructive sleep apnoea (OSA) and sudden death are other features. Uncommon findings are spinal or cardiac anomalies. People with TCS usually have normal intelligence. Anaesthesia in patients presenting for surgery is a challenge in view of difficult mask ventilation and difficult intubation by conventional means [6,7]. This was first described by Ross way back in 1963 [8].

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

Airway surgery: mandibular distraction, tracheostomy.

Craniofacial: cleft palate repair, orbitozygomatic surgeries (including bone grafting), orthognathic surgery of maxilla (e.g. Le Fort I osteotomy) and/or mandible [9].

ENT: tympanostomy tube insertion, otoplasty.

Type of anaesthesia

Anticipation of a difficult airway is the key anaesthetic consideration (see below). There are no known contraindications to specific anaesthetic drugs or techniques beyond this consideration.

However, in adult patients, it may be preferable to employ regional anaesthesia techniques where feasible to avoid the need to manage the difficult airway. Regional anaesthesia/analgesia may also reduce the risk of airway obstruction post-operatively by reducing opioid requirements.

Necessary additional preoperative testing (beside standard care)

There may be coexisting obstructive sleep apnoea (sleep study may be warranted in individual cases).

Particular preparation for airway management

Always prepare for an anticipated difficult airway as this is the most frequent problem encountered during anaesthesia.

Initial face mask ventilation, with or without oropharyngeal airway after induction of anaesthesia, has been described as difficult in a third of the patient population [6]. A large proportion of children with TCS require intubation techniques other than conventional direct laryngoscopy.

Direct laryngoscopy becomes more difficult with increasing age [6]. Therefore preparation for an anticipated difficult airway should always be undertaken for a patient with TCS even if previous airway management and intubation was uneventful. Additionally, in contrast to Pierre Robin sequence, patients with TCS who have had surgical correction with mandibular distraction may still be difficult to intubate [10].

Preparation for an anticipated difficult airway includes:

- Previous history of general anaesthesia and difficult intubation
- Clinical examination of the airway including assessment of mouth opening, temporomandibular joint movement, and dental implantation
- Availability in the operating room of equipment necessary for difficult intubation and skilled assistance. This includes a variety of alternative airway devices and techniques / plans as described in difficult airway algorithms [11,12] such as: stylet,

bougie, video laryngoscope, fibre-optic intubation, two anaesthetist technique, paraglossal straight blade technique [13]. The laryngeal mask airway (LMA) [6] or second generation supraglottic airway devices such as the i-gel [14] have been successfully used to provide an adequate airway in TCS when intubation has been difficult. Equipment and experienced staff for the scenario of a “can’t intubate, can’t oxygenate” (CICO) situation needs to be available and in theatre [11,12,15].

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Eye protection might be warranted during craniofacial or ENT procedures – e.g., lubrication and lid closure.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Avoid sedative premedication where possible. Excessive use of opioids in the post-operative period should be avoided because of the risk of airway obstruction. As far as possible, regional analgesia techniques and opioid-sparing agents should be used.

Always prepare for an anticipated difficult airway (see the “Particular preparation for airway management” section). Anaesthesia technique for an anticipated difficult airway usually involves maintenance of spontaneous ventilation as far as possible. Inhalational anaesthesia or remifentanyl infusion or dexmedetomidine infusion may provide suitable conditions for intubation whilst maintaining spontaneous ventilation. Airway patency with manual bag mask ventilation should be confirmed before considering administration of muscle relaxant (an exception to this may be the occurrence of laryngospasm).

The patient should be extubated only when fully awake and keeping all precautions of repeat intubation at hand [16].

Particular or additional monitoring

The nature of the surgery will dictate the degree of monitoring.

Exercise great care to ensure endotracheal tube is well secured (adequate fixation with consideration to the operative field) as dislodgement intraoperatively has been reported [6]. ETCO₂ monitoring is mandatory to confirm intubation and to detect dislodgement of the airway device.

Postoperatively, it is important to monitor for signs of airway obstruction.

Possible complications

Airway instrumentation in the setting of a difficult airway may result in airway complications including dental trauma, lacerations to lips or gums, post-extubation oedema and stridor.

Endotracheal tube dislodgement intraoperatively has been described [6].

Gastric rupture has been described in a CICO scenario in a child [17].

Postoperative care

Postoperatively, it is important to monitor for signs of airway obstruction (including stridor), particularly in patients with OSA. CPAP may be needed in the postoperative period for oxygenation.

The appropriate choice of post-operative ward setting is dependent upon the surgery type and the patient's airway. High dependency or intensive care unit (ICU) may be appropriate. If not admitted to ICU, monitoring of SpO₂ is mandatory in the recovery room with a clinical focus on early detection of respiratory problems.

Disease-related acute problems and effect on anaesthesia and recovery

The most likely emergency scenario to be encountered under anaesthesia with Treacher-Collins syndrome is the "can't intubate, can't oxygenate" (CICO) scenario.

Ambulatory anaesthesia

Case-by-case determination based on of the impact of surgery and anaesthesia on the patient's airway.

Obstetrical anaesthesia

Early referral and regional anaesthesia techniques are preferable where feasible.

In urgent situations of failed intubation, the laryngeal mask airway (LMA) [18] or second-generation supraglottic airway devices such as the i-gel [14] have been reported to provide an adequate airway including in an emergency Caesarean section [18].

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