Aktiv Druck & Verlag GmbH | ISSN 0170 - 5334 I 02330

www.ai-online.info

61. Jahrgang | März 2020

A&I

ANÄSTHESIOLOGIE & INTENSIVMEDIZIN

Offizielles Organ: Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V. (DGAI)
Berufsverband Deutscher Anästhesisten e.V. (BDA)
Deutsche Akademie für Anästhesiologische Fortbildung e.V. (DAAF)
Organ: Deutsche Interdisziplinäre Vereinigung für Intensiv- und Notfallmedizin e.V. (DIVI)

Camp(t)omelic dysplasia

Fabry disease

orphanaesthesia

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

SUPPLEMENT NR. 4 | 2020

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 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:

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

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orphananesthesia

Anaesthesia recommendations for

Camp(t)omelic dysplasia

Disease name: Camp(t)omelic dysplasia

ICD 10: Q87.1

Synonyms: Camp(t)omelic dwarfism, Camp(t)omelic syndrome

Disease summary: Camp(t)omelic dysplasia (CD) is an autosomal dominantly inherited disorder of the SOX9 gene (17p24.3-q25.1), caused by de novo mutation or defective chromosomal recombination [1,2,3]. The SOX9 gene encodes a transcription factor that affects chondrogenesis, testicular development and determines phenotypic sex characteristics [4,5]. Pathological findings can be grouped in osseous and non-osseous disorders. Osseous features are the naming bowed femora and tibia (campomel = bent limb; camptomel bowed limb), short stature, vertebral abnormalities with cervical instability and possible cord compression (paraplegia), only eleven rib pairs, facial dysmorphia (Pierre Robin sequence, short neck), cleft palate, progressive scoliosis, club feet and dislocatable hips. Non-osseous features include laryngotracheobronchomalacia with respiratory compromise, ambiguous genitalia with sex reversal (female external genitalia with 46 XY karyotype), congenital heart disease (ventricular septal defect, VSD), hypotonia, kidney malformation (hydronephrosis) and neurological disorders including hydrocephalus and hearing impairment [6,7]. No clinical feature is obligatory. When femora and tibia are not affected, it is termed as acamp(t)omelic camp(t)omelic dysplasia (ACD). The estimated prevalence ranges from 1:10,000 to <1:1,000,000 [8,9]. Because of the rare occurrence of CD, an exact prevalence is uncertain. Currently, only symptomatic therapy is available. Most affected patients die in the neonatal period due to respiratory insufficiency [10]. Operative treatment is performed if individuals reach infancy.

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

▶ **Citation:** Albermann M, Prottengeier J: Camp(t)omelic dysplasia. Anästh Intensivmed 2020;61:S67–S73. DOI: 10.19224/ai2020.S67

Typical surgery

Routine operative treatment of

- cleft palate
- club foot
- hip luxation
- cervical vertebral instability
- kyphoscoliosis
- removal of gonads in sex reversal
- arthroscopic surgery.

Type of anaesthesia

So far, no drug intolerance associated with CD has been reported. One available case report describes malignant hyperthermia in a child with CD during intensive medical care [11]. No trigger was determined. There is no defined increased risk of malignant hyperthermia in these patients.

Generally, regional anaesthesia techniques are feasible. Because of the osseous disorders, difficult conditions or even failure have to be expected. Anatomical landmarks may be unreliable, but ultrasound guidance may be beneficial. Spinal or epidural anaesthesia can be technically difficult and must be individually considered in the setting of pre-existing neuronal damage.

Because of possible laryngotracheomalacia, caution is advised in the setting of sedation without a secured airway, i.e. during radiological diagnostics.

Necessary additional pre-operative testing (beside standard care)

Since CD is associated with VSD, it is necessary to evaluate and quantify the cardiac condition pre-operatively. A sizeable septal defect has significant consequences for the anaesthetic plan.

Furthermore, it is important to know if the cervical spine is unstable and needs additional stabilisation during anaesthetic interventions, the operation itself and in the recovery room afterwards. If the spinal cord is affected, it will be advisable to document the neurological status in order to recognise deterioration post-operatively.

In case of hydronephrosis, renal function should be established (creatinine, urea, diuresis) and drugs adjusted accordingly.

Particular preparation for airway management

Due to the defective chondrogenesis in CD patients, a narrow supra- and infraglottic space along with a hypoplasia of the trachea-bronchial cartilage might exist. Micrognathia, cleft palate and a limited cervical mobility may cause difficulties for intubation and tracheomalacia might impair ventilation. This complexity can make a direct laryngoscopy a challenge in the

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worst cases. Therefore, you should be prepared for a “cannot intubate conventionally” situation and should have tools available for a difficult airway, with a backup “cannot ventilate conventionally” plan in your mind. Frequently, only a tube smaller than calculated from height and weight can be inserted. A cuffed tube is recommended, as re-intubations with uncuffed tubes for size fitting is undesirable in these circumstances.

Particular preparation for transfusion or administration of blood products

Not reported so far.

Particular preparation for anticoagulation

Not reported so far.

Particular precautions for positioning, transportation and mobilisation

Because of possible cervical instability and potential cord compression, you have to be particularly careful when the head is positioned. A shoulder roll that places the external auditory meatus in line with the clavicle can be beneficial in patients with craniothoracic disproportionality. In cases where there is a high index of suspicion of instability, e.g. based either on imaging or diagnosed symptomatic neurological deficits, a secure fixation will be recommended. Pre-existing contractures and deformities may need special attention during positioning.

Interactions of chronic disease and anaesthesia medications

Not reported so far.

Anaesthetic procedure

Premedicants should be used with individualised caution given airway concerns.

Standard induction of anaesthesia may be performed if the following conditions are fulfilled: adequate mouth opening, no higher facial dysplasia, no cervical instability – all suggestive of “can ventilate”. After successful mask ventilation, conventional laryngoscopy may be performed initially. If unfeasible, the ASA difficult airway algorithm should be applied. A videolaryngoscope should be applied in conventional intubation, usually with a smaller tube than age or weight would suggest. Normally, intubation is successful this way. Otherwise, a laryngeal mask should be taken into consideration with a possible device change, e.g. via LMA Fastrach or bronchoscopy. If intubation is still impossible and supraglottic devices are deemed ineffective or inappropriate for the given surgery, the patient should emerge from anaesthesia. In this case an awake fibre-optic intubation should be performed (see ASA difficult airway algorithm).

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Should any of the above-mentioned hints of “cannot ventilate” be present (cervical instability, inadequate mouth opening, higher facial dysplasia), then awake fibre-optic intubation should be the method of choice, if feasible. In the case of a high index of suspicion for cord compromise, manual in-line stabilisation should be utilised throughout the intubation process.

Particular or additional monitoring

If relevant VSD exists, haemodynamic monitoring should be expanded by contiguous blood pressure measurement and application of catecholamines should be prepared if necessary.

Possible complications

The worst complication associated with CD is ventilatory compromise from either a “cannot intubate, cannot ventilate” situation or post-extubation ventilatory failure. Both instances can lead to hypoxic brain injury, negative pressure pulmonary oedema and death. Permanent or temporary spinal cord injury can result from undiagnosed or underestimated cervical instability in these patients.

Post-operative care

Extubations in deep sedation are discouraged in these patients, given the possibility of laryngotracheomalacia. Patients should be fully alert prior to extubation. Diagnosed or suspected, obstructive or central sleep apnoea must be taken into consideration relative to the length of post-operative monitoring. For patients who are on CPAP at home, the use in the post-operative period, e. g. in the recovery room, may be beneficial.

Disease-related acute problems and effect on anaesthesia and recovery

Remember: You may encounter a difficult airway and need equipment to handle such a scenario, a surgical airway option should also be available.

Ambulatory anaesthesia

Because of the possibility of a difficult airway in patients with CD, it cannot be recommended to perform general anaesthesia under ambulatory conditions without a second set of hands, means to provide a prolonged recovery phase or advanced airway equipment.

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Obstetrical anaesthesia

The rate of airway difficulties in the parturient is known to be increased per se. In the rare coincidence of pregnancy and campomelic dysplasia, a difficult airway and respiratory failure have to be anticipated.

Two alternatives seem feasible in principle and have to be evaluated individually:

1) Elective intubation and C-section well prior to expected date of delivery.
2) Neuraxial/regional anaesthesia for an elective C-section, so as to avoid the intricacies of intubation in these patients.

It is recommended that women with ACD give birth in a tertiary hospital only.

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- Little People of America, Inc. (USA): http://www.lpaonline.org/

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Date last modified: June 2017

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Disclosure The authors have no financial or other competing interest to disclose. This recommendation was unfunded.

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Disclosure The reviewers have no financial or other competing interest to disclose.

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Herausgeber

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Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V.
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Schriftleitung
Präsident/in der Herausgeberverbände

Gesamtschriftleiter/Editor-in-Chief:
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Verlag & Druckerei

Aktiv Druck & Verlag GmbH
An der Lohwiese 36 |
97500 Ebelsbach | Deutschland
www.aktiv-druck.de

Geschäftsführung
Wolfgang Schröder | Jan Schröder | Nadja Schwarz
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E-Mail: info@aktiv-druck.de

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Titelbild
Gestaltung: Klaus Steigner
Paumgartnerstraße 28 | 90429 Nürnberg
E-Mail: mazyblue@klaus-steigner.de
www.klaus-steigner.de

Erscheinungsweise 2020

Der 61. Jahrgang erscheint jeweils zum Monatsanfang, Heft 7/8 als Doppelausgabe.

Bezugspreise (inkl. Versandkosten):

- **Einzelhefte** 30,- €
- **Jahresabonnement:**
  Europa (ohne Schweiz) (inkl. 7 % MwSt.) 258,- €
  Schweiz 266,- €
  Rest der Welt 241,- €

  Mitarbeiter aus Pflege, Labor, Studenten und Auszubildende (bei Vorlage eines entsprechenden Nachweises)
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