orphanaesthesia

Anaesthesia recommendations for patients suffering from

Camp(t)omelic dysplasia

Disease name: Camp(t)omelic dysplasia

ICD 10: Q87.1

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

Certain phenotypes are named: acamp(t)omelic camp(t)omelic dysplasia

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 **laryngo-tracheo-broncho-malacia 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 neonatal period due to respiratory insufficiency (10). Operative treatment is performed if individuals reach infancy.

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

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 adviced in the setting of sedation without a secured airway, i.e. during radiological diagnostics.

Necessary additional diagnostic procedures (preoperative)

Since CD is associated with VSD, it is necessary to evaluate and quantify the cardiac condition preoperatively. A sizeable septal defect has significant consequences on the anesthetic plan.

Further it is important to know, if the cervical spine is unstable and needs additional stabilization during anesthetic interventions, the operation itself and in the recovery room afterwards. When the spinal cord is affected, it is advisable to document the neurological status in order to recognize deterioration postoperatively.

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 infra-glottic space along with a hypoplasia of the trachea-bronchial cartilage can be present. Micrognathia, cleft palate and limited cervical mobility may cause difficulties for intubation and tracheomalacia can impair ventillation. This complexity can make a direct laryngoscopy a challenge in the 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 smaller tube than calculated from size 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 precautions for positioning, transport or mobilization

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 cranio-thoracic disproportionality. In cases where there is a high index of suspicion for instability such as via imaging or symptomatic neuro-deficits, secure fixation is recommended. Pre-existing contractures and deformities may need special attention during positioning.

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

Not reported so far.

Anaesthesiologic procedure

Premedicants should be used with individualized caution given airway concerns.

Standard induction of anaesthesia may be performed if the following are given: 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 video-laryngoscope should be applied for conventional intubation, usually with a smaller tube than age or weight would suggest. Normally, in this way intubation is successful. Otherwise, a laryngeal mask should be taken in consideration with a possible device change, e. g. via LMA Fastrach or bronchoscopy. When 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 fiber-optic intubation should be performed. (see ASA difficult airway algorithm)

Should any of the above-mentioned hints of "cannot ventilate" be present (cervical instability, inadequate mouth opening, higher facial dysplasia), then awake fiber-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 stabilization should be utilized throughout the intubation process.

Possible complications

The worst possible 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 under-recognized cervical instability in these patients

Postoperative 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 as to the length of postoperative monitoring. For patients on CPAP at home, use in the postoperative period may be beneficial.

Information about emergency-like situations / Differential diagnostics

Remember: You may encounter a difficult airway and equipment to handle such a scenario, including a surgical airway option, should 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.

Obstetrical anaesthesia

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

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

1) Elective intubation and C-Section well prior to expected date of delivery.

2) Neuraxial/Regional Anesthesia 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.

Literature and internet-links

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Internet-Links:

International Skeletal Dysplasia Society: http://www.skeldys.org/

European Skeletal Dysplasia Network: http://www.esdn.org/

Restricted Growth Association (UK): http://www.restrictedgrowth.co.uk/

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

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