orphanaesthesia

Anaesthesia recommendations for

Achondroplasia

Disease name: Achondroplasia

ICD 10: Q77.4

Synonyms: Chondrodysplasia, chondrodystrophia fetalis

Disease summary: Achondroplasia is the most frequent of more than 100 described types of skeletal dysplasia which lead to dwarfism. The incidence is approx. 0.5-1.5 in 10,000 newborns [1]. Spontaneous mutations cause up to 80% of the diseases. Hereditary propagation takes place in terms of autosomal-dominant transmission. Females are more often affected than males [13,19,22]. Genetically speaking, this is the mutation of the fibroblast growth factor receptor 3 gene (FGFR3) [2]. This mutation results in an inhibition of cartilage proliferation and a disorder of enchondral ossification. As a consequence, premature ossification of epiphyseal cartilage is observed. Clinically speaking, the following symptoms are characteristic: disproportionate dwarfism, a relatively large head, midfacial hypoplasia, deformations of the spine, leg axis deviation, „trident hand“. As a primary or secondary consequence, other organ systems can be affected [1,14]. As a result, also anaesthesiological particularities have to be observed.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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Typical surgery

Otorhinolaryngology: tonsillectomy, adenotomy, grommet (tympanostomy tube); oral and maxillofacial surgery: dysgnathia (tightly aligned teeth); neurosurgery: ventricular shunt, craniectomy (foramen magnum stenosis); spinal surgery: spinal canal stenosis, kyphoscoliosis, (paediatric-)orthopaedics: malposition of the extremities, lengthening; bariatric surgery.

Type of anaesthesia

A general recommendation regarding the ideal anaesthetic regime cannot be given, as both general and regional anaesthesia present potential problems [2,14,19]. Therefore, individual decisions are necessary.

General anaesthesia: typical described problems are:

Excessive anxiety [2,8,12,37]; difficult intravenous access (10-50%) [2,9]; difficult mask ventilation, difficult intubation [2,4,10,11,18,19,21,23]; risk for cervico-medullary compression or rather spinal cord ischemia (reported sudden death events, above all in children < 4 years) [3,6,31]; eight-fold higher obesity rate potentiating the effects of the existing problems [14]; increased incidence of sleep apnea (obstructive and/or central) – rarely secondary pulmonary hypertension, restrictive lung diseases already at an early age [1,23,25-29]; chronic respiratory infections [1,28]; ten-fold increased cardiovascular risk – with a maximum between 25 and 35 years of age [30,35], tendency toward hypersalivation. In early childhood, a nasopharyngeal muscular hypotonia can become a problem [25], as well as gastro-oesophageal reflux [28].

There are descriptions of unproblematic general anaesthesia [9,12,27].

Despite the altered respiratory system, general anaesthesia is frequently regarded as the method of choice [1,10]. Due to the anatomical changes in the spine and the craniocervical junction as well as due to an increased incidence of hydrocephalus, neuraxial anaesthetics are relatively contraindicated.

Neuraxial regional anaesthesia: this type of regional procedure is considered to be technically difficult (narrow spinal canal/stenoses, reduced epidural space, kyphoscolioses, vertebral body deformities [10,23]). The conus medullaris often is positioned lower than usual [37]. In some cases, epidural anaesthesia was carried out successfully [19-22]. There are reports about accidental dural punctures, difficulties in advancing the catheter [1], increased risk of venous puncture [19], irregular or unpredictable (unpredictably high) spread of anaesthesia [19,21]. Epidural anaesthesia should be preferred because of the possibility of titration [13]. In most cases, injection of local anaesthetic into the caudal canal is easier in the case of paediatric patients.

Also spinal anaesthesia has been applied successfully [16-18]. Inadequate quality of neuraxial analgesia [16], punctio sicca and the risk of high spinal anaesthesia are conceivable [14]. Regarding both procedures, the use of opiates has been described [14, 18], however, clear dosage recommendations are lacking.

Furthermore, successful CSE with low spinal dose with the possibility to increase the height of the block using the epidural catheter is described. Ultrasound guidance can be very helpful to identify vertebral interspaces and to determine the distance to the ligamentum flavum [34,46].

Peripheral regional anaesthesia: possible. Needle placement can be complicated.

Conscious sedation / awake anaesthesia: There are no reports about severe occurrences. Caution is advised in the case of pre-existing sleep apnea syndrome [28].

Necessary additional pre-operative testing (beside standard care)

The preoperative diagnostic assessment depends on the respective symptoms of the patient and is arranged on the basis of a detailed anamnesis and physical examination. Particular attention has to be paid to signs indicating a difficult intubation (small mouth opening, big tongue, limited cervical vertebrae extension, tonsillar hyperplasia, instability of the cervical spine) [4,13,14].

Besides the usual preoperative care, the following pathognomonic clinical symptoms would indicate that a refined diagnostic assessment would make sense [1,14,25,28,30]:

- Chronic/current infections (otitis media):
  otorhinolaryngology consultation

- Severe scolioses/Rib cage deformities:
  lung function, echocardiography, blood gas analysis, thoracic radiography

Annotation: conventional pulmonary function tests are indexed to height and are unreliable. An equation to estimate vital capacity and long volumes of patients with achondroplasia exists [45].

- Neurological symptoms (hydrocephalus, cervicomedullary compression, spinal canal stenoses):
  neurology consultation, CT, MRI

- Sleep apnea:
  otorhinolaryngology consultation, sleep laboratory, blood gas analysis

- Cardiopulmonary problems (restrictive lung diseases, pulmonary hypertension, cor pulmonale, heart diseases):
  lung function, electrocardiography, echocardiography, blood gas analysis, thoracic radiography, if necessary further examinations.

Annotation: Cardiac output is based on body surface area formulae that rely on height which lead to a lower predicted value of the cardiac output requirements. Probably, the Boyd formula provides the most realistic cardiac index [44].

Particularly before a planned regional anaesthesia procedure, pre-existing neurological abnormalities have to be documented [19]. Frequently, achondroplasia patients show neurological abnormalities [6]. In infancy and childhood, but above all in adulthood, they often suffer from chronic pain due to skeletal changes [3,6].

Particular preparation for airway management

For anatomical reasons, problems may occur when face mask ventilation or intubation are performed [2,4,10,11,18,19,21,23]. The big tongue and pronounced adenoid vegetations may lead to complications. However, a frequent reason for intubation difficulties is also a limited flexibility of the atlanto-occipital joint [10,11,27]. On the other hand, due to the risk of a foramen magnum stenosis and cervical instability (risk of medullary compression), hyperextension of the cervical spine during intubation should strictly be avoided [9-12,19,23,31,43]. This is emphasized by a case report about a non-traumatic cervical spinal cord infarction which led to quadriplegia in a child – even without preceding manipulation of the cervical spine [33].

In case of a clinically expected difficult intubation it is recommended to have the airway devices ready which are normally used in the respective clinic.

Awake fiberoptic intubation (N.B. mid-facial hypoplasia) is considered the preferred method but often not feasible by the pronounced anxiety in these patients [2,8,12,37]. A GlideScope is not an option for an awake intubation. Using this device is not always possible due to macroglossia and the probably limited mouth opening [4,13,39]. In case of impossibility of an awake fiberoptic-guided intubation the procedure should be performed while spontaneous breathing is maintained. Difficulties can occur even when using LMA devices [36].

Despite pre-oxygenation patients desaturate very fast [37]. Patients further may suffer from an unusually collapsible larynx, trachea and/or bronchi [40]. They may be difficult to ventilate. Weaning from ventilator also may be delayed [43].

In the reports on unproblematic intubations in patients with achondroplasia, a small and short trachea is mentioned. The following rule applies: the size of the tube should be chosen according to the body weight (not as usual according to the age) [2,4,8,9,12,27,40,41]. Mechanical ventilation should be adjusted to 6 cc/kg ideal body weight for height. In case of indication of a tube thoracostomy smaller intercostal spaces and anatomical variations should be anticipated [43].

Due to the fact that regional procedures do not offer a safe alternative, also in these cases the airway management should be planned very well before the operation.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

There are no reports on a particular thrombosis prophylaxis in case of achondroplasia. In general, after these patients have entered puberty an adequate thrombosis prophylaxis has to be taken into consideration, particularly if they have to undergo an extended period of immobilization and lower extremity surgery.

One case report describes a severe and lethal intraoperative fat embolism as a consequence of a lower extremity intervention [32].

Particular precautions for positioning, transportation and mobilisation

Greatest care is required in order not to risk damages caused by patient positioning in case of anatomical particularities (above all spine and extremities). Frequently, these patients have joint contractures. Some of them are unable to lie either in a prone of supine position [36].

Case reports on damages caused by positioning do exist (e.g. two cases of brachial plexus palsy [9], one case of visual loss after prone positioning during spine surgery [24]).

As compared to the size of the body, the head is relatively big and due to the relatively increased body surface area, there is a good chance of a significant drop in body temperature. Especially in the case of children, early thermal management has to be remembered.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Premedication: Whereas some authors refrain from sedation medication when a difficult intubation has to be expected or when the patient is diagnosed with a sleep apnea syndrome [4], other authors describe that anxiolysis is helpful to these patients who are often very anxious [2]. For patients with severe adiposity, ranitidine and metoclopramide or sodium citrate are frequently administered in terms of aspiration prophylaxis [4,14,19].

Anaesthesia: when venous conditions are difficult, inhalational induction of anaesthesia can be considered. Due to excessively lax skin, soft tissue and joint flexion deformities the surgical exposure of a vein can be required [36]. The subclavian and jugular access can be challenging as well [43].

Regarding the use of anaesthetic medication calculated according to the body weight, no particularities are reported [2,12]. However, in case of a slim achondroplastic patient a dosage in this manner probably can lead to under-dosing [37]. Patients with severe hypersalivation can be given a vagolytic drug – in most cases, secretion aspiration is sufficient.

One should avoid negative inotropic medication [1].

Regarding other drugs which are generally used preoperatively, there are no absolute contraindications.

There are no signs indicating an increased tendency toward malignant hyperthermia.

Particular or additional monitoring

Monitoring should be oriented towards the patient's pre-existing, organ-specific diseases. In most cases, a routine monitoring corresponding to the surgical intervention is described [14]. The choice of the appropriate blood pressure cuff can present a challenge.

In the case of pre-existing cardio-respiratory problems, invasive blood pressure measurement is recommended [19].

Possible complications

- Difficult airway management (including smaller tube size)
- Hypersalivation
- Frequent respiratory tract infections
- Risk of cervico-medullary compression or spinal cord ischemia
- Difficult regional anaesthesia with partly unpredictably high spread
- Increased obesity rate
- Tendency towards sleep apnea syndromes (obstructive and/or central)
- Increased cardiovascular risk
- High risk of intraoperative damage caused by positioning.

Post-operative care

Primarily, postoperative care is based upon the intervention and the patient's pre-existing conditions. Even without anaesthesia, the tendency towards central and obstructive sleep apnea is held responsible for the increased mortality rate, above all in early childhood [3,6,30,31,42]. The risk for postoperative respiratory complications is increased [37]. For this reason, particularly in the case of children it is recommended to carry out pulsoxymetric monitoring after surgery [25] – this applies above all when opioids are administered [19]. Moreover, an extended stay in the anaesthetic recovery room should be scheduled. A stay in intensive care is not mandatory, but has proven its value in bariatric surgery [4]

Disease-related acute problems and effect on anaesthesia and recovery

Typical differential diagnostics refer above all to the neurological aspects of the disease:

- Potential neurological failures after regional anaesthesia (DD of disease-related neurological symptoms versus nerve damage caused by regional anaesthesia)
- Possibly increased risk of an ascending neuraxial blockade versus sleep apnea due to a central cause.
- Postoperative hypopnea / apnea (DD of central sleep apnea corresponding to the underlying disease versus residual opiate effect).
- 25,6% of the patients suffer from chronical pain in the lower limbs. This is often associated with a depression [47] (DD postoperative pain versus chronical pain).

Ambulatory anaesthesia

The hitherto existing literature does not provide any recommendations regarding outpatient procedures in patients with achondroplasia. As a rule, anaesthesia on an outpatient basis should be avoided due to the above mentioned possible postoperative problems. This applies above all to paediatric patients. Especially in the first year of life the risk of sudden death is significantly higher than in the general population even without anesthesia [38].

Obstetrical anaesthesia

Many articles concerning neuraxial procedures in case of achondroplasia deal with anaesthesia in Caesarean section. Due to a disproportion between the infant's head and the mother's pelvis, pregnant women with achondroplasia have a high rate of scheduled Caesarean sections [19,23]. Most articles indicate the anaesthesiological dilemma of a potentially difficult respiratory system and a possibly difficult regional anaesthesia [13-16,21]. Close to the spinal cord, preference should rather be given to epidural anaesthesia [13] because of the superior titratability. In case of emergency, also spinal anaesthesia has been carried out with success [14]. With respect to these pregnant women, general anaesthesia has to be planned particularly well. This procedure is favoured by some authors [1,10,41]. Pre-existing alterations of the respiratory system become even more difficult to handle. Moreover a reduction of FRC has to be encountered – clearly exceeding the reduction which is considered to be normal in pregnant women – as a consequence of a thoracic hypoplasia and a possibly restrictive lung function, resulting in intrapulmonary shunts. This means that in addition to an altered respiratory system, these women have a very limited pulmonary reserve, implying a great risk of hypoxia [23]. Phenotypically speaking, these patients appear in the 16th week of gestation like other women in the 30th week of gestation [19]. This might also involve an (earlier) increased risk of aspiration.

There are no definite recommendations regarding anaesthetic procedures during pregnancy. The decision for a certain anaesthetic procedure has to be taken in each individual case after a detailed risk-benefit analysis.

TAP blocks are suitable for postoperative analgesia after caesarean section to reduce morphine consumption in patients with achondroplasia [37]

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