Anaesthesia for Paediatric Patients With Achondroplastic Short Stature

Dr Andrew O'Donoghue[1†], Dr James Isherwood[2], Dr Bríd McGrath[3]

[1]SpR in Anaesthesia and Critical Care, Children's Health Ireland at Crumlin, Dublin, Ireland
[2]SpR in Anaesthesia, Sheffield Children's Hospital, England
[3]Consultant in Anaesthesia, National Orthopaedic Hospital, Cappagh, Dublin, Ireland

Edited by: Dr Kate Wilson, Consultant in Anaesthesia, Sheffield Children's Hospital, England

[†]Corresponding author e-mail: andrew.odonoghue@umail.ucc.ie

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KEY POINTS

- Achondroplasia is the most common cause of pathological short stature.
- The preanaesthetic assessment clinic is the ideal location for a holistic approach to preoperative preparation.
- Obesity and sleep-disordered breathing are very common in children with achondroplasia.
- There is a high prevalence of spinal stenosis in children with achondroplasia, increasing the risk of spinal cord compression during manipulation.

INTRODUCTION

There are multiple causes of short stature including skeletal dysplasias, mucopolysaccharidoses, and idiopathic short stature. Of these, achondroplasia (Latin, 'without bone growth'), which is a disorder of bone growth, is the most common pathological cause. Disordered change from cartilage to bone during growth results in rhizomelic shortening of limbs (by affecting the proximal long bone, hip, and shoulder), macrocephaly, and characteristic facial features. Mental abilities are usually unaffected.

AETIOLOGY

Achondroplasia occurs due to a heterozygous mutation in the fibroblast growth factor receptor 3 gene, located on the short arm of chromosome 4. Its inheritance is autosomal dominant. Most cases (80%) are not inherited but occur as a result of a de novo gene alteration. It is the most common cause of short stature at 1:26 000 to 1:28 000 live births. There are large variations in prevalence in different populations. For example, prevalence is as high as 1:4100 in Africa and the middle East and as low as 1:28 500 in Europe and South America.

REVIEW OF SYSTEMS

Airway

Airway abnormalities are common and include small mouth opening, malocclusion, macroglossia, and limited cervical vertebrae extension with instability. Sleep-disordered breathing occurs in children with achondroplasia due to several predisposing factors including hypoplasia of the facial bones, relative adenotonsillar hypertrophy, and skull base dysplasia. Stenosis of the vertebral canal at the cervical level may result in myelopathy and central apnoea.[1] Patients may have an unusually collapsible larynx, trachea, and/or bronchi.[2]

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Breathing

Smaller chest size and mild deformities including pectus excavatum and lateral indentation are common and can result in smaller effective lung volumes.[3]

Children with achondroplasia suffer from recurrent ear infections, with high rates of otitis media and conductive hearing loss. This is due to shortened Eustachian tubes. It has been reported that approximately 40% of people with achondroplasia have functionally relevant hearing loss, sometimes delaying language development.[2]

Cardiovascular System

The heart and other internal organs are usually functionally preserved in childhood. There is an increased ischaemic cardiovascular risk in the adult population, but this does not present until after adolescence.[4] Intravenous access can be challenging due to deeper skin folds and shortened limbs.

Neurological System

A neurological deficit will be present in 20% to 50% of children with achondroplasia. Narrowing of the cranio-cervical junction may cause brainstem compression as well as neurological symptoms and signs in the upper limbs. There is 7.5% mortality in the first year of life due to apnoeas, but careful evaluation and management can reduce this to 0.3%.[2] Hydrocephalus and hypotonia are also commonly observed in infancy.

Gastrointestinal System

Increased prevalence of obesity,[5] with an increased risk of gastro-oesophageal reflux. Interestingly there is a *decreased* risk of diabetes mellitus.[6]

Musculoskeletal System

Abnormal joint range of movement is common and can result in joint contractures. These contractures can make positioning challenging. Spinal abnormalities include lumbar lordosis and thoracolumbar kyphosis in 80% to 99% of patients. The lower limbs exhibit varus deformity in 80% to 99% of patients, with lateral bowing, internal tibial torsion, and dynamic instability of the knee.

COMMON PROCEDURES

Common procedures performed on patients with achondroplasia include the following:

- Otorhinolaryngological procedures
  - Adenotonsillectomy
  - Myringotomy or tympanoplasty tube (grommets)
- Neurosurgical procedures
  - Ventriculo-peritoneal shunt
  - Suboccipital (cranio-cervical) decompression
- Orthopaedic procedures
  - Spinal surgery (for kyphosis, lordosis, scoliosis)
  - Genu varus correction
  - Limb lengthening
  - Laminectomies for spinal stenosis

PERIOPERATIVE MANAGEMENT

Preoperative Management

Airway assessment is particularly important, looking for features mentioned earlier. Presence of snoring and risk factors for sleep apnoea should be discussed and polysomnography considered for those considered high risk.

Assessment of lung function to look for restrictive lung disease secondary to chest and spinal deformities may be useful and becomes more relevant as the patient gets older. Conventional pulmonary function tests are indexed to height and are unreliable, but an equation to estimate vital capacity and lung volumes of patients with achondroplasia exists.[7]

Cardiac function should be assessed closely, looking for signs of pulmonary hypertension, particularly in those with significant restrictive lung disease. Electrocardiography and echocardiography may be helpful, especially prior to thoracic spinal surgeries. Cardiac output monitors derive cardiac index using formulae based on body surface area, which is calculated from height. This will lead to overprediction of the patient's cardiac index. The Boyd formula has previously been shown to provide the most realistic value for cardiac index in these patients.[8]

A thorough examination of the neurological and musculoskeletal systems is important given the prevalence of hydrocephalus, joint laxity, hypotonia, and lower limb claudication. The frequency of axial deformities should be kept in mind and magnetic resonance imaging or computed tomography of both lumbar and cervical spine should be considered to assess the presence of stenosis or cervical instability. If computed tomography or magnetic resonance imaging are not available, lateral flexion and extension views on plain film x-ray will help to demonstrate spinal stability.[9] Subtle spondylolisthesis may indicate the presence of stenosis.[10]

Intraoperative Management

Same-day admission on morning of surgery, if possible, minimises anxiety and time away from home. Premedication is helpful in minimising anxiety and future fears around general anaesthesia and this can be important in the context of multistage operations. Clear fluids can be taken orally up to 1 hour preoperatively to minimise unnecessary prolonged fasting and fluid depletion and to minimise difficulty gaining intravenous access.

Intravenous induction is preferable if practical, as it allows rapid administration of emergency drugs when necessary and separates airway concerns from administration of anaesthesia. A plan for airway management should be based on preoperative assessment at the preanaesthetic assessment clinic and should include appropriate equipment and alternative plans for a difficult airway. Use of a head ring and shoulder roll can help to stabilise the cervical spine and provide the best possible first attempts for intubation. Endotracheal tube size is generally that expected for *age,* as the head and laryngeal development correlate with this. Supraglottic airway devices, however, may need to be smaller than expected due to a small mouth and large tongue. Mechanical ventilation should be set initially to 6 mL/kg ideal body weight for height and adjusted as appropriate.

Special attention to detail should be paid to positioning the patient for surgery. There is a risk of atlantoaxial subluxation during airway manoeuvres and transfer. Avoid undue tension of the joints, particularly at the cervical spine and shoulders with well-placed, supportive padding. Raising the knees reduces strain on the lumbar spine when supine. Given the relatively high surface area to weight ratio, temperature control should be established early and monitored throughout.

Drug doses may be calculated as usual according to ideal, total, or lean body weight. There are no absolute contraindications, but agents that may interfere with spinal cord monitoring or exacerbate apnoeas should be used with caution.

A multimodal approach to perioperative analgesia is effective. Typically, this might include the following:

- Simple analgesics (paracetamol, nonsteroidal anti-inflammatory drugs)
- Short-acting opiates
- Neuraxial analgesia, if not contraindicated; may be technically challenging
  - Epidural catheter (eg, levobupivacaine dosed by weight and spread required)
  - Caudal epidural (eg, levobupivacaine dosed by weight and spread required)
- Ketamine infusion 0.05 to 0.2 mg/kg/h
- Dexmedetomidine infusion 0.3 to 1 μg/kg/h

Postoperative analgesia should be prescribed according to the procedure and local guidelines. Avoidance of long-acting opiate medication is desirable in the context of higher prevalence of sleep-disordered breathing but may be unavoidable when appropriate neuraxial or regional techniques are contraindicated. Gabapentin may be considered if there is abnormal or neuropathic pain and diazepam if there is muscle spasm.

At the end of the procedure preoxygenation and careful positioning is necessary prior to extubation to avoid any sudden or rapid changes in neck positioning in case of laryngospasm and apnoea.

Otorhinolaryngological Procedures

Involve the surgical team in planning and positioning of the patient; this is particularly important in shared-airway procedures. It is important to avoid hyperextension during manipulation of the head and airway. It is preferable to keep the patient breathing spontaneously intraoperatively, if possible, to reduce the risk of rapid desaturation and obstruction.

Neurosurgical Procedures

The use of cerebrospinal fluid (CSF) diversion procedures is now rare following the realization of the natural history of CSF space enlargement in these patients.[11] Overflexion of the neck, even for short times, has been shown to cause increased intracranial pressure due to venous outflow obstruction and blockage of CSF outflow,[12] therefore careful positioning must be agreed upon between the neurosurgeon and the anaesthetist to ensure minimal compromise.

Craniocervical decompression procedures can cause severe postoperative pain, nausea, and vomiting, which may be difficult to manage. This can last for up to 10 days and is especially likely if the dura has been opened. Careful preoperative counselling is essential, along with multimodal analgesia and regular antiemetics postoperatively.

Orthopaedic Procedures

Many of the orthopaedic procedures typical for this patient population, particularly spinal surgery, can result in profound blood loss. Careful monitoring of haemorrhage is essential and cell salvage or transfusion is often required. Preloading with crystalloid to lower haematocrit and red cell loss is well described and can be effective. Early treatment is important not just to reduce haemodynamic instability but to support spinal cord monitoring. If the operative site does not allow for exsanguination and tourniquet, loading dose of the antifibrinolytic agent tranexamic acid is advisable, with an infusion in place for longer procedures, dosed by weight.

A clinical picture of sudden transient hypotension, tachycardia, pyrexia, and hypercapnia is commonly seen with torniquet release. After 30 to 60 minutes of tourniquet time, muscle tissue begins to break down creating an acidotic, hypoxic, hypercapnic, hyperkalaemic, and lactataemic inflammatory soup, which is returned into systemic circulation following release of the tourniquet. Vasopressors are sometimes required to counteract the peripheral vasodilation and central effects of this mixture. An increase in minute ventilation may be required if end-tidal CO_2 increase is substantial.

Spinal Cord Monitoring

If lumbar stenosis is present, then regular monitoring of somatosensory and motor evoked potentials while under general anaesthesia is useful during spinal surgery. Total intravenous anaesthetic such as propofol/remifentanil infusion is necessary to facilitate this, avoiding neuromuscular block if monitoring motor evoked potentials. Volatile agents supress motor cortex excitability resulting in diminished amplitudes and total intravenous anaesthetic has been shown to mitigate this.[13] While somatosensory evoked potentials are less susceptible to the dose-dependent suppression of potentials by inhalational agents, other factors such as blood pressure, anaemia, and temperature influence latency and amplitude of responses.[14]

Postoperative Management

Admission to the high-dependency unit may be necessary for management of analgesic and ventilation requirements postoperatively depending on local policy and patient risk factors.

It is important to encourage return to oral intake as soon as possible and mobilise as soon as is allowable from the surgical perspective to minimise postoperative length of stay.

SUMMARY

Given its high prevalence, it is likely that the anaesthetist will encounter patients with achondroplasia during their career. Most procedures are corrective and will occur in the paediatric population.

Preoperative planning is the key to a safe and holistic approach to these patients; it provides an appropriate setting for both assessment and counselling of patient and parent or guardian. Expectations and outlook can be managed and optimised.

Obesity and sleep-disordered breathing are prevalent in this population and avoidance of long-acting opiate analgesia is advantageous if possible.

Monitoring of somatosensory evoked potential and motor evoked potential during spinal surgery is essential due to prevalence of spinal stenosis. Total intravenous anaesthetic is required as inhalational agents supress cortical activity and thus amplitude of motor potentials.

Diligent management of haemodynamic status and intraoperative anaemia is also essential as these influence somatosensory potentials. We have experienced pronounced instability following the release of a tourniquet and advise vigilance.

A high-dependency unit should be considered for postoperative care depending on the procedure, analgesia requirements, and presence/extent of sleep-disordered breathing.

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