

Anesthetic Management of a Patient with Achondroplasia Undergoing Adenotonsillectomy: A Case Report

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Abstract

Background: There are several situations that can create a challenge to every anesthesiologist; one of them is genetic disorders such as achondroplastic dwarfism. Achondroplastic dwarfism is the most common form of skeletal dysplasia, affecting about one in 20,000 newborns. It is an autosomal disorder caused by a mutation of the fibroblastic growth factor receptor-3 gene.

Case Presentation: A 7-year-old boy diagnosed with achondroplasia was admitted for adenotonsillectomy. He had several classical symptoms and signs of upper airway and cardiac involvement. In this case report, we describe the anesthetic management of this patient, while reviewing the difficulties encountered by the anesthesia team perioperatively.

Conclusions: Difficulties with airway management and physiology of this disease can create significant challenges to the anesthesiologist. So, we should keep in mind that achondroplasia can be a complicated situation and we should be prepared to manage it.

Conflicts of Interest: The Authors declare no conflicts of interest.

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Background

There are several situations that can create a challenge to every anesthesiologist; one of them is genetic disorders such as achondroplastic dwarfism (1). Achondroplastic dwarfism is the most common form of skeletal dysplasia, affecting about one in 20,000 newborns (2). It is an autosomal disorder caused by a mutation of the fibroblastic growth factor receptor-3 gene (3). This mutation results in decreased endochondral ossification and defects in craniofacial and vertebral bones (4). So careful perioperative evaluation and preparation in the operating room are required.

In this case report, we discuss the anesthetic management of a patient with achondroplasia referred for adenotonsillectomy.

Case Presentation

A 7-year-old boy, with achondroplasia, presented with the history of excessive

nasopharyngeal secretion and bilateral rhonchi in the lungs, who was referred for adenotonsillectomy due to adenotonsillar hyperplasia.

The patient had been treated for obstructive sleep apnea (OSA), asthma, gastroesophageal reflux, and hypertrophic cardiomyopathy. Furthermore, he had been admitted to PICU 3 times due to aspiration pneumonia and was intubated during those periods. He had undergone circumcision by local anesthesia six years ago.

The prescribed drugs for his conditions were omeprazole, inhalational salbutamol and fluticasone, as well as oral sildenafil. Physical examination revealed macrocephaly, small midface with flattened nasal bridge, small oral orifice, malalignment of the teeth, chest wall deformity (pectus excavatum), severe kyphosis, short stature (proportionally small upper and lower limbs), finger anomaly (trident finger), as

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well as speech and language problems with normal intelligence. He stood 85 cm tall and weighed 17 kg (BMI: 23 kg/m²). Laboratory parameters including hematological and biochemical profiles were normal. In perioperative evaluation, a cardiologist and pulmonologist were consulted to optimize the perioperative condition of the patient's underlying diseases. Echocardiography revealed ejection fraction of 48%, mild mitral and tricuspid regurgitation, and pulmonary artery pressure of 35 mmHg. The patient was monitored with ECG, pulse oximetry and noninvasive blood pressure entering the operating room, indicating blood pressure of 120/80 mmHg and pulse rate of 120, while the arterial saturation of oxygen during breathing oxygen via facemask was 100%. Inhalational induction was performed using 8% sevoflurane to reach the acceptable depth of anesthesia and then intravenous line was inserted. Next, 0.2 mg of atropine, 1 mg of midazolam, 50 mcg of fentanyl, 20 mg of lidocaine and 10 mg of atracurium were administered and the patient was intubated under video laryngoscopy using a cuffed spiral ETT (≠4). Isoflurane was the maintaining hypnotic agent throughout the surgery (1.2-1.5 %).

Pressure controlled ventilation mode was selected to reach the optimal volume and pressure of the respiratory system considering the underlying condition (PEEP: 3, peak pressure: 18, RR: 18, FiO₂:50 % with air). The procedure lasted for 30 minutes and the neuromuscular block reversed with titrated neostigmine (0.05 mg/kg) and atropine (0.02 mg/kg). Following a successful and uncomplicated extubation, the patient was transferred to the ICU.

Discussion

Numerous studies have reported OSA and breathing abnormalities in achondroplasia (5). Guilleminault et al. revealed that both craniofacial features and tonsillar hypertrophy correlate with OSA in this population (6).

Previous studies have shown the effectiveness of adenotonsillectomy in the treatment of OSA in the normal population (7). However, its effectiveness in children with achondroplasia is less certain (8). Although the success rate is lower than in the general pediatric population, long-term follow-up has suggested that OSA can be cured by adenotonsillectomy in many cases of this specific group (8).

Anesthetic risk in achondroplasia include difficulty with laryngeal visualization, problems in mask ventilation due to facial anomalies, small caliber of the airway, short neck, upper airway muscular hypotonia, risk of cervicomedullary compression due to neck hyper-extension or uncontrolled head movement and difficulties in ventilation because of restrictive lung disease (9, 10). In this case, we did not experience any events through intubation process, although we had planned several remedies considering the probability of any of the above difficulties. Indirect visualization of all oropharyngeal landmarks was possible using a videolaryngoscope. However, stridor occurred while sevoflurane was being administered via anesthesia facemask despite maintaining enough ventilation monitored by capnography. Another complication that is associated with achondroplasia is hydrocephalus due to intracranial venous hypertension secondary to occipital bone abnormalities (11).

For relieving neurological symptoms, suboccipital craniectomy for foramen magnum stenosis, cervical fusion for cervical instability and ventriculoperitoneal shunt for hydrocephalus can be done (12). Our patient did not have this problem. Cardiopulmonary abnormality that is associated with achondroplasia include congenital and valvular heart disease, cardiomyopathy, coronary artery disease, and pulmonary hypertension (11). This case had cardiomyopathy and pulmonary hypertension.

Due to skin and soft tissue laxity in achondroplasia, venous access may be difficult

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(13). We used the inhalation method of induction as this method is an option for an anticipated difficult airway and after adequate depth of anesthesia, we inserted secured intravenous access.

In adult patients with achondroplasia, fiberoptic bronchoscopy is a good choice but in children they are not cooperative. Meanwhile, special care should be done in the manipulation of neck. Note that because of cervicomedullary compression and cervical myelopathy that can cause paralysis and death.

Another concern is sleep apnea manifestations postoperatively. In our case, we decided to observe the child closely in our post anesthesia care unit (PACU) for an extended period of 4 hours while keeping CPAP (continuous positive airway pressure) in mind as adjuvant

therapy in case of symptomatic obstruction. However, lateral positioning and oxygen via nasal cannula were all he needed. Nevertheless, delayed obstructive symptoms may happen, even late as 24 hours, which is why “inpatient” surgery is recommended for diagnosed sleep apnea undergoing adenotonsillectomy. We also hospitalized our patient for the next 24 hours with proper monitoring.

Conclusions

Difficulties with airway management and physiology of this disease can create significant challenges to the anesthesiologist. So, we should keep in mind that achondroplasia can be a complicated situation and were should be prepared to manage it.



Figure 1. Midface anomalies and short stature of achondroplasia.



Figure 2. Trident fingers of achondroplasia.



Figure 3. Short neck and skeletal deformities of trunk result in difficulty in airway management, as well as restrictive lung disease leading to pulmonary hypertension.

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Conflicts of Interest

The Authors declare no conflicts of interest.

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