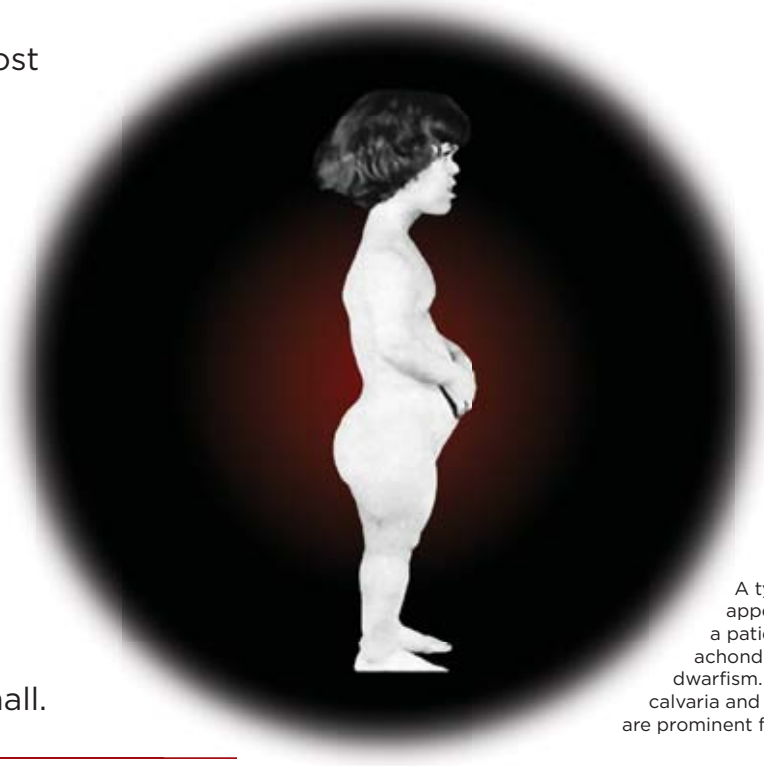
Achondroplasia:
Implications and Management Strategies in Anesthesia

Joan E. Spiegel, MD
Assistant Professor
Beth Israel Deaconess Medical Center
Harvard Medical School
Boston, Massachusetts
Editorial Advisory Board Member, *Anesthesiology News*

Matthew Hellman, MD
Clinical Fellow in Anesthesiology
Beth Israel Deaconess Medical Center
Boston, Massachusetts

The authors reported no relevant financial disclosures.

Achondroplasia is the most common cause of dwarfism. As a genetic disorder of skeletal dysplasia, it literally translates to "without cartilage formation." Affected patients fail to achieve a height of 148 cm by adulthood, and have a typical appearance of shortened limbs and a large head in comparison with body size. Midgets, by contrast, are proportionately small.



A typical appearance of a patient with achondroplastic dwarfism. Large calvaria and mandible are prominent features.

Achondroplasia is observed in approximately 1 to 1.5 of every 10,000 births as a result of a spontaneous fibroblast growth factor receptor gene mutation. Therefore, most achondroplastic patients will have parents of normal height. Achondroplasia is also transmitted in an autosomal dominant fashion. As a result of inhibition of cartilage formation, there is premature ossification in the epiphyseal growth plates with concurrent restriction of growth. Clinically, patients will have deformations of the spine and airway, and hypoplasia of structures of the head. The functioning of many organ systems can be affected by achondroplasia, but intelligence and life expectancy are usually normal. The safe delivery of an anesthetic requires an understanding of this disorder and the particular pathophysiology of the patient undergoing surgery.

Anatomy in Dwarfism

Major anatomic differences can create difficulties in the anesthetic management of achondroplastic dwarfs (Table). Airway abnormalities include narrowed nasal passages from mucopolysaccharide deposition, tracheal narrowing, sternal prominence, pharyngeal and maxillary hypoplasia, and thickening of pharyngeal and laryngeal structures. Laryngomalacia can spontaneously cause stridor, whereas subglottic stenosis has been reported to necessitate tracheostomy. Patients may have a large tongue, tonsils, and adenoids along with a stiff temporomandibular joint. An estimated 40% of patients with achondroplasia have obstructive sleep apnea, even in childhood.

Spinal deformations in achondroplasia include a short neck, odontoid hypoplasia, thoracolumbar kyphosis, pelvic narrowing, lordosis, and moderate to severe spinal stenosis. The narrowing of the spinal column can result in cauda equina syndrome, nerve root compression, thoracolumbar spinal cord compression, or, rarely, high cervical cord compression secondary to stenosis of the foramen magnum.

Quadriplegia resulted in a patient with dwarfism and severe kyphosis after surgery, and in an infant with achondroplasia with foramen magnum stenosis secondary to normal range of motion of the neck resulting in atlantoaxial subluxation. The concave shape of the occipital bone may prevent optimal positioning for intubation due to entrapment of the atlas. Additionally, high cervical stenosis or foramen magnum stenosis can cause central sleep apnea. Respiratory function may be impaired from having a small rib cage, obesity, and spinal deformations.

Surgical Optimization

Achondroplastic dwarfs must be carefully assessed before undergoing anesthesia, ideally in a preoperative testing center with sufficient time for any workup, if needed. As previously mentioned, contributions to airway difficulties include anatomic differences in both airway structure and stability.

Thoracolumbar anatomic differences such as severe scoliosis and rib cage deformities resulting in restrictive lung diseases, pulmonary hypertension, cor pulmonale, and heart disease may warrant echocardiography, pulmonary function tests, chest radiography, and a baseline blood gas analysis. Patients with severe sleep apnea and/or chronic respiratory illnesses may require consultation with an otolaryngologist to identify airway anomalies.

Flexion–extension lateral cervical spine radiographs as well as a view of the odontoid process should be standard due to the risk for atlantoaxial subluxation secondary to odontoid hypoplasia. MRI or CT scanning

Table. Considerations for Anesthetic Management of Achondroplastic Dwarfs

Common Problems	Considerations for General or Regional Anesthesia
Anxiety	IV placement/difficult awake FOI
Anatomy Large head Large tongue Midfacial hypoplasia Obesity Rib hypoplasia Short neck Small mouth Tonsillar hyperplasia	Difficult head positioning Difficult laryngoscopy Difficult mask ventilation/difficult seal
Cor pulmonale Pulmonary abnormalities Pulmonary hypertension Restrictive and obstructive lung disease	Postoperative respiratory difficulties Respiratory difficulties/ventilation
Cervical instability Foramen magnum stenosis Neurologic/orthopedic Scoliosis Spinal stenosis	Caution with positioning (extension) for intubation and surgery to avoid paralysis and plexopathies Dosing for regional anesthesia unclear Epidural anesthesia may be preferred over spinal anesthesia Potential high spinal
Miscellaneous	Smaller airway adjuncts required

FOI, fiber-optic intubation

should be considered if radiographs fail to provide adequate imaging before surgery, or if there is concern for high cervical stenosis or foramen magnum stenosis. All neurologic symptoms necessitate further consultation and workup before elective surgery.

Airway Management

Previous airway management records should be obtained when possible. Pharyngeal and maxillary hypoplasia along with narrow nasal passages and an enlarged tongue can cause problems with routine laryngoscopy. Thus, awake FOI is probably the safest method of securing the airway, but direct laryngoscopy has been reportedly successful. There are reports of minimal problems with masking, ventilating, and intubating these patients when appropriately sized—usually smaller—airway adjuncts are used.

The concern for atlantoaxial instability and alterations in the base of the skull may suggest the use of in-line stabilization with video laryngoscopy. Awake intubation with a GlideScope (Verathon) is not recommended, as these patients suffer from high levels of anxiety, making sedation and compliance difficult. Administering an anticholinergic drug before intubation may aid in the identification of airway structures because hypersalivation is frequently encountered. Laryngeal mask airways are still very much a part of the difficult airway algorithm; however, pediatric-sized LMAs should be readily available.

Ventilatory difficulty with LMAs may be encountered if underlying pulmonary disease is present, or if a distorted airway prevents an optimal fit. Careful attention should be paid to respiratory rate and tidal volume, as lung anatomy and function may be significantly reduced.

Regional Anesthesia

Neuraxial regional anesthesia in achondroplastic dwarfs may prove difficult, impossible, or perhaps dangerous—even for the experienced provider. Patients with achondroplasia are known to have narrowed and stenosed spinal canals, decreased distances between lumbar pedicles, kyphoscoliosis, osteophyte formation, engorged epidural veins, and a reduced epidural space. There may be difficulty in advancing catheters, unpredictable and patchy or high spread of anesthesia, and increased risk for venous and dural puncture.

Epidural anesthesia is preferred over spinal anesthesia, as careful titration is desirable. Unintentional dural puncture may be difficult to detect in the setting of severe spinal stenosis because of low or absent cerebrospinal fluid.

Case Report

A 51-year-old woman with achondroplasia presented for noncosmetic nasal septoplasty. Anesthetic history revealed multiple difficult intubations and laryngeal mask airway (LMA) placements. She was successfully intubated in 2007 for cervical fusion surgery via awake nasal fiber-optic intubation (FOI) after failed oral FOI. She refused awake FOI for this surgery.

On physical examination, she was a well-appearing, attractive woman with achondroplastic features. Height and weight were 48 inches and 88 pounds, respectively. Airway exam revealed a Mallampati III view, marginal mouth opening at 2 to 3 cm, adequate mandibular prognathism, good dental health, and a normal-sized tongue. Her neck was short, and its range of motion was severely limited in flexion and extension. Films revealed an absence of the odontoid process and fusion of anterior and posterior C2-4 and C6-7.

After a discussion with her otolaryngologist, a 20-g IV line was easily placed. She was given 1 mg of IV midazolam and 0.2 mg of glycopyrrolate. Standard monitors were applied in the operating room, and the patient was preoxygenated for approximately 3 minutes. She was given 100 mcg of fentanyl with good effect, and 150 mg of propofol was administered in divided doses to achieve apnea. Mask ventilation was possible but somewhat difficult, even with an oral airway. A pediatric fiber-optic scope was placed to view the airway before attempted LMA insertion, but visualization of the vocal cords was unsuccessful due to distortion from surrounding tissue. The head and neck surgeon was also unable to identify anatomy. An LMA ProSeal 3 was placed and initial ventilation was possible, but after a few minutes, the end tidal CO_2 waveform was not uniform. The LMA was removed and mask ventilation was adequate, although more difficult.

A fiber-optic scope was reinserted, but technical difficulties with the equipment made the procedure unsuccessful and it was aborted. A decision was made to awaken the patient. However, mask ventilation continued to be difficult, so succinylcholine and increased sevoflurane concentrations were given. Despite this, barely 30- to 60-cc tidal volumes were obtained on ventilation, and 2 providers were required to maintain air movement. Abdominal distention was significant, which further limited lung excursion. After 15 to 20 minutes of careful positive-pressure ventilation, the patient emerged from anesthesia and recovered without incident. She ultimately underwent her procedure under local anesthesia without complications.

Clear dosage recommendations are lacking for neuraxial procedures in patients with achondroplasia. Peripheral nerve blocks can be performed following careful documentation of preexisting abnormalities. Chronic pain syndromes are encountered more commonly in achondroplasia as a result of bony malformations.

Obstetric Considerations

Pregnancy is an especially high-risk condition for achondroplastic dwarfs. Thoracic structural abnormalities with potential restrictive lung function cause a further reduction in the functional residual capacity normally seen in parturients, resulting in exaggerated shunts and ventilation/perfusion mismatch. This limited pulmonary reserve, coupled with the even greater airway management difficulty in a parturient with achondroplasia, warrants early scheduled cesarean delivery to avoid emergent airway intervention.

Obstetric patients with achondroplasia have a very high rate of elective cesarean deliveries due to the likelihood of a cephalopelvic disproportion in those with a contracted pelvis, a common coexisting condition. A carefully titrated epidural is the preferred anesthetic for these patients. However, any neuraxial choice can be complicated by high-level, patchy, or incomplete neuraxial spread of the anesthetic.

There are a handful of cases in the literature documenting the safe administration of spinal anesthesia for cesarean delivery. In one case, a standard amount of medication (12 mg of hyperbaric bupivacaine, 25 mcg of fentanyl, and 0.3 mg of morphine) was used without creating too high a level.

Conclusion

Achondroplastic dwarfism presents many anesthetic challenges, and deaths have been reported despite careful preparation. The anatomic variations and their effects on organ systems and airway management can make these patients unexpectedly difficult to manage. The prevalence of achondroplasia makes it likely that most anesthesia providers will encounter one of these patients at least once. An understanding of the pathophysiology of these patients, careful preparation, and communication of the anesthetic plan with the surgical team are essential components of perioperative management.

Additional Reading

Berkowitz ID, Raja SN, Bender KS, et al. Dwarfs: pathophysiology and anesthetic implications. *Anesthesiology*. 1990;73(4):739-759.

Cevik B, Colakoglu S. Anesthetic management of achondroplastic dwarf undergoing cesarean section—a case report. *Middle East J Anaesthesiol.* 2010:20(6):907-910.

Jain A, Jain K, Makkar JK, et al. Anaesthetic management of an achondroplastic dwarf undergoing radical nephrectomy—a case report. *South Afr J Anaesth Analg.* 2010;16(2):77-79.

Krishnan BS, Eipe N, Korula G. Anaesthetic management of a patient with achondroplasia. *Paediatr Anaesth*. 2003;13(6):547-549.

Mayhew JF, Katz J, Miner M, et al. Anaesthesia for the achondroplastic dwarf. *Can Anaesth Soc J.* 1986;33(2):216-221.

Oppitz F, Speulda E. Anesthesia recommendations for patients suffering from achondroplasia. www.orpha.net/data/patho/Pro/en/Achondroplasia_EN.pdf. Accessed June 30, 2015.

Mitra S, Dey N, Gomber KK. Emergency cesarean section in a patient with achondroplasia: an anesthetic dilemma. *J Anesth Clin Pharmacol*. 2007;23(3):315-318.

Tetzlaff J, Benedetto P. Skin and bone disorders. In: Fleisher L, ed. *Anesthesia and Uncommon Diseases, 6th Edition.* Philadelphia, PA: Saunders; 2012.