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Case Report

Anesthesia Management in Achondroplasia: A Case Report

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ABSTRACT

Achondroplasia is the result of a mutation in the gene encoding the type 3 receptor for a fibroblast growth factor. This abnormality results in malformation endochondral ossification. Achondroplasia is characterized by disproportionately short stature, lumbar lordosis, large head, midface hypoplasia, short hands, and normal cognitive development.

Our report is about a 28-year-old patient with achondroplasia who underwent surgery for correction of kyphosis due to spinal canal stenosis. In this present case we study management anesthesia.

Achondroplasia is the most common form of dwarfism. Dwarfism is defined as a failure to achieve a height of 148 cm in adulthood. This condition is the result of a mutation in the gene encoding the type 3 receptor for a fibroblast growth factor (FGFR3). This defect result in abnormal endochondral ossification. The majority of cases (90%) are the result of a sporadic mutation. If the condition is inherited, it is topically through autosomal dominant transmission [1].

More than 95% of patients have the same point mutation in the gene for fibroblast growth factor receptor 3 (FGFR3), and more than 80% of these are new mutations. The mutation, which causes a gain of FGFR3 function, affects many tissues, most strikingly the cartilaginous growth plate in the growing skeleton, leading to a variety of manifestations and complications. Most of the serious complications can be modified favorably or prevented by anticipation and early treatment. Possible future treatment includes chemical inhibition of receptor signaling, antibody blockade of receptor activation, and alteration of pathways that modulate the downstream propagation of FGFR3 signals [2].

Achondroplasia is characterized by disproportionately short stature, lumbar lordosis, large head, midface hypoplasia, short hands, and normal cognitive development. Its incidence is estimated at 1 in 10,000 to 1 in 30,000. Although it is an autosomal dominant condition, the majority of cases occur as a result of a de novo genetic mutation. Patients with achondroplasia may present for orthopedic surgery as children or adults for correction of associated abnormalities such as tibial bowing and spinal stenosis. The primary anesthetic challenge in patients with achondroplasia is airway management. Midface hypoplasia with a pharynx that is small in proportion to the tonsils, adenoids, and tongue makes these patients prone to upper airway obstruction and may hinder direct laryngoscopy. A flat nasal bridge and a large mandible may make it difficult to obtain an adequate seal for mask ventilation. Hyperextension of the neck should be avoided due to the possibility of foramen magnum stenosis. Video laryngoscopy or fiberoptic intubation should be considered for these patients, and a range of endotracheal tube sizes should be on hand, as many patients require a smaller than what would be expected based on age. Other anesthetic considerations in patients with achondroplasia include the possibility of difficult neuraxial anesthesia due to spinal deformity or stenosis and cardiopulmonary sequelae such as restrictive lung disease, central and obstructive sleep apnea, and resultant pulmonary hypertension. Preoperative echocardiogram to assess for pulmonary hypertension should be considered prior to major surgery [3].

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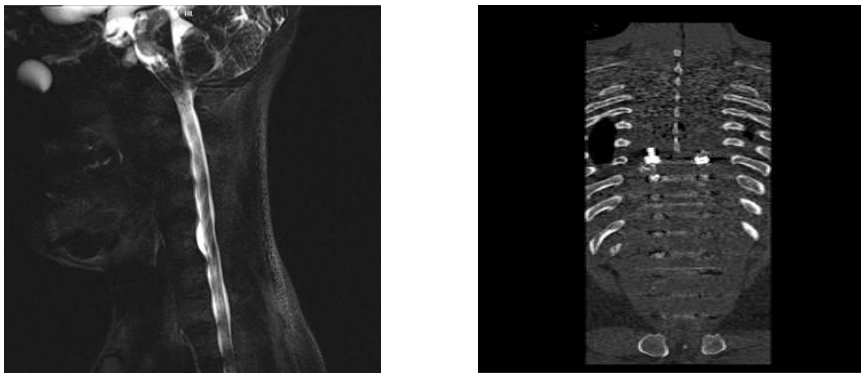
Case Report

The patient was a 28-year-old man who was a candidate for PSF8 level surgery due to spinal canal stenosis and to correct the deformity of thoracic lumbar kyphosis (Figure 1). The patient was a candidate for surgery due to chronic pain and foot droop. He had two other brothers, both of whom had achondroplasia. The patient's parents also had achondroplasia. The patient was 139 cm tall and weighed 45 kg. Mental growth was completely normal. He had a short neck, large mandible, and thoracic kyphosis and lumbar lordosis. The patient reported normal activity and had no history of sleep apnea, heart, or respiratory disease. X-ray examination of the spinal canal stenosis is quite evident. Laboratory findings were normal. In cardiac counseling, echocardiography was performed which was reported to be normal. At the time of admission, the conscious and awake patient had BP = 120/60 mmHg, HR = 92/min, SPO2 = 95%. After preparing the difficult airway equipment and an endotracheal tube of different sizes, the patient was ready for anesthesia. After pre-oxygenation, the patient was ready for induction of anesthesia. 0.5 mg of midazolam, 100µg of fentanyl, 40 mg of lidocaine, 100 mg of propofol, and 40 mg of succinylcholine were injected to facilitate endotracheal intubation. The patient was intubated with cuff tube No. 6 under video laryngoscopy without neck extension. The endotracheal tube was confirmed by hearing and capnography and after confirming the location of the endotracheal tube, it was fixed with a bandage. Peripheral vein catheter (16 G) and arterial line catheter were installed to monitor blood pressure. Pulse oximetry, capnography, ECG, and NIBP were also placed. Appropriate chest position and head ring were placed for the prone position. During anesthesia surgery with isoflurane (1Mac) and atracurium injection at regular intervals was continued if the return of muscle relaxation was confirmed. The operation lasted 6 hours, during which the patient had 1 liter of bleeding. In total, he received 1 liter of fluid (1.2 normal saline, 1.2 ringer) per unit cell pack. At the end of the surgery, he had 250 ccs of urine. ABG was taken 3 times, which was normal. At the end of the surgery, the patient was reversed with 2 mg of neostigmine and 1 mg of atropine. After full consciousness, the patient was extubated and transferred to the ICU with spontaneous breathing and fully conscious and awake after 1 hour of recovery.

Figure 1- A: spinal canal stenosis B: Corrected deformity of thoracic lumbar kyphosis



A **B**

Discussion

Achondroplasia is a skeletal dysplasia that presents with limb shortening and short stature. Plain X-ray images show shortened tubular bones with increased diameter, broad and irregular metaphyses of the long bones that exhibit cupping, shortened femoral necks, fibula bones that are longer than tibia bones, narrowing of the distance between the lumbar pedicles, posterior scalloping of the lumbar vertebral bodies, narrowing of the sciatic notch, a horizontal acetabular roof, shortening if the base of the skull, and facial hypoplasia. Platyspondyly sometimes observed in the neonatal period. The diagnosis is made when the symptoms and bone X-ray images are consistent. An enlarged head circumference (above the 97 percentile), shortening if the femoral bone (below the 5 percentile), and trident hand are observed in the fetal period [4]. The characteristic clinical symptoms and radiological findings for achondroplasia are observed starting from the neonatal period, although approximately 20% of patients are not diagnosed at this time [5]. Foramen magnum narrowing, ventricular enlargement, sleep apnea, upper airway stenosis, otitis media, a narrow thorax, spinal canal stenosis, spinal kyphosis, malformation of the lower extremities, and joint hyperextensibility are also observed with achondroplasia [4-6].

When achondroplasia is clinically suspected, radiographs from a skeletal survey can generate a differential diagnosis and confirm the definitive diagnosis. Occasionally, the diagnosis can be established by prenatal testing. when the family history is positive and the parents desire a prenatal diagnosis, chorionic

villus sampling at 11 to 13 weeks of gestation or amniocentesis after 15 weeks of gestation can be performed. These tests allow the use of polymerase chain reaction and restriction enzyme fragment to detect the FGFR3 G380R point mutation [7].

Impairment from achondroplasia is not limited to the musculoskeletal system. Hydrocephalus may occur during the newborn and infantile period. Because children with achondroplasia have relatively large heads and frontal bossing, diagnosis may be difficult. Diagnosis is made by closely monitoring the head circumference and motor development and by comparing those measurements with standard achondroplastic data. Ventricular shunting is required in approximately 5% of patients [8].

Otolaryngol problems also are prevalent: 90% of patients with achondroplasia experience otitis media before age two years and a half requires placement of tympanostomy tubes.23 Relative adenotonsillar hypertrophy secondary to midface hypoplasia can cause obstructive sleep apnea. Otitis media and adenotonsillar hypertrophy may result in conductive hearing loss that can impair speech development. These problems may lead to disabilities in communication and learning [8].

The cause of the developmental delay is not clear, although foramen magnum stenosis must be ruled out. Cognitive development is normal. Maintaining ideal body weight is a continuous challenge for many individuals with achondroplasia, and obesity is more common than in the general population. Sports participation, especially swimming and biking, should be encouraged. The child should avoid gymnastics and collision sports because of the potential for neurologic complications secondary to cervical stenosis [9].

Patients with achondroplasia are very healthy compared with patients with other dysplasias, but mortality rates in all age groups are higher than those in the general population because of sudden death in young infants, central nervous system and respiratory problems in older children, and cardiovascular problems in young adults [10].

Anesthetic Management

Perform a preoperative assessment and carefully assess the airway. These patients are at increased risk of upper airway obstruction and difficult intubation. Obesity is a common finding. Obstructive sleep apnea (OSA) is present in up to 40% of patients, including children. A recurrent ear infection may lead to chronic serous otitis media and hearing impairment [11].

Respiratory compromise can be from a combination of factors, including reduced chest circumference, upper airway obstruction, and cervivomedullary compression. Pulmonary hypertension may also occur. There is an increased risk of sudden death due to central respiratory failure. Consider electrocardiogram (ECG), echocardiography, and chest X-ray to evaluate Cardiopulmonary status [11].

Common procedures for these patients include tonsillectomy, adenoidectomy, limb lengthening, cervicomedullary decompression, fusion, and ventriculoperitoneal shunt. Monitor somatosensory evoked potentials for spine case. Choose the anesthetic technique that is best for the surgical procedure; both general and regional anesthesia have been used successfully. The most important consideration is airway management. Consider awake fiberoptic intubation. use a smaller -sized endotracheal tube. Base drug dosages on the patient's weight. Neuraxial anesthesia (both spinal and epidural) has been described but may be technically difficult. Anesthesia for cesarean section has been described with both neuraxial and general anesthesia techniques [12].

Extubate the patient when fully awake. Use postoperative opioids with caution because these patients are at risk for airway obstruction. Consider postoperative inpatient monitoring. Prolonged postoperative ventilation may be necessary [12].

Conclusion

Achondroplasia is the most common skeletal dysplasia. Manifestations are seen in the spine, the upper and lower extremities, and the otolaryngol system. Foramen magnum stenosis may become symptomatic in the first 2 years of life and require decompression. Although thoracolumbar kyphosis typically resolves, surgery may be necessary for spinal stenosis. Genu varum may require corrective osteotomy. The indications and outcomes of limb lengthening for patients with achondroplasia remain controversial. Because the manifestations appear in multiple locations, pediatricians and orthopedic surgeons from multiple specialties need to take part in providing care for the patient with achondroplasia.

These patients are usually referred for shunt surgery, spinal stenosis, laminectomy, limb deformity, tympanostomy, tonsillectomy, and cesarean section. Choice of anesthesia including IV sedation and MAC where there is a risk of airway obstruction. Regional, peripheral nerve block, and neuneuraxial methods can be used. Patients may be intubated while awake in the event of a difficult airway. Postoperative care is important, including awake extubation because of opioid sensitivity.

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