Anesthesia for a patient of acromesomelic dysplasia with associated hydrocephalus, Arnold Chiari malformation and syringomyelia

Rudrashish Haldar, Prakhar Gyanesh, Sukhen Samanta

Department of Anaesthesiology, Sanjay Gandhi Post Graduate Institute of Medical Sciences, 1Department of Critical Care Medicine, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow, Uttar Pradesh, India

Abstract

Acromesomelic dysplasias are autosomal recessive osteochondrodysplasias. Acromesomelic dysplasia Maroteaux-type (AMDM), also known as St Helena dysplasia, is of two types: The classical and the mild variety. About 50 cases of AMDM have been reported till date, most of them being the classical variety. There is scarcity of literature on anesthesia for such patients. We are reporting a case of general anesthetic management of AMDM, associated with hydrocephalus, Arnold Chiari malformation type-1 and syringomyelia. The patient was a 10-year-old short-statured boy who presented with symptomatic thoracic kyphoscoliosis, gibbus deformity and back pain. On examination, there was no neurological deficit. Radiology revealed thoracic kyphoscoliosis, mild ventriculomegaly and upper cervical syringomyelia. The patient underwent posterior fossa decompression in the prone position under general anesthesia. We will discuss the anesthetic considerations for such patients and review the pertinent literature.

Key words: Acromesomelic dysplasia, arnold chiari malformation, osteochondrodysplasia, short stature, syringomyelia

Introduction

Acromesomelic dysplasia Maroteaux-type (AMDM), first described by P. Maroteaux in 1971, is a rare autosomal recessive osteochondrodysplasia. Abnormalities such as low thoracic kyphosis, narrow thoracolumbar interpedicular distance, gibbus deformity, oval vertebral bodies, lumbar lordosis, increased joint laxity, limited elbow extension, short, broad phalanges, and loose skin over the fingers are present. Facial appearance and intelligence are however normal.[1] The AMDM gene has been mapped to human chromosome 9p13-q12.[2]

AMDM is the rarest of all acromesomelic dysplasias, with nearly 40-50 reported cases.[3] We report a case of acromesomelic dwarfism associated with kyphoscoliosis, syringomyelia, hydrocephalus and tonsillar descent with Arnold Chiari malformation type-1 undergoing posterior fossa decompression under general anesthesia.

Case Report

A 10-year-old male patient (30 kg, 122 cm) presented with short stature and a stooping posture due to pronounced kyphoscoliosis [Figure 1] along with back pain for the past three years. History of snoring, breath holding, cyanosis, paradoxical chest movement, restless sleep, daytime somnolence and family history of dwarfism were absent. He was cooperative, had normal mental function and intellectual development without any neurological deficit. Airway examination revealed adequate mouth opening, Mallampati class-II and unrestricted neck extension.

Pulmonary functional tests and laboratory values were within regular limits. Chest and cervical region X-ray appeared normal, without subluxation on flexion and extension. Thoracic kyphosis was present with a Cobb’s angle of 35° [Figure 2]. Magnetic resonance imaging of brain revealed hydrocephalus,
ventriculomegaly, a short posterior fossa, tonsillar herniation and a 3-mm syrinx. Posterior fossa decompression by C1, C2 laminectomy with removal of a foramen magnum rim was planned.

On the night before surgery the patient was instructed to follow standard ASA fasting guidelines. Anticipating difficult venous cannulation due to the coarse and loose skin [Figure 3], the AV300 Vein Viewing System was used to identify a vein in the dorsum of the left hand, which was marked.

On the day of surgery, the patient was brought to the operation theatre in a calm state and 5-lead ECG, non-invasive blood pressure and pulse oximetry were connected. A 20-G intravenous cannula was inserted into the previously marked site after application of EMLA cream.

The severe kyphoscoliosis and gibbus deformity prevented the patient from assuming a supine position for intubation [Figure 4]. We modified the position by placing a thick pillow that supported the occiput and compensated the gibbus deformity [Figure 5]. Keeping the difficult airway cart ready the child was sedated with intravenous midazolam (1 mg), fentanyl (60 µg) and a bolus dose of propofol (60 mg). After confirming the ability to mask ventilate and glottic visualisation following a gentle check laryngoscopy, vecuronium (3 mg) was administered to facilitate intubation. Endotracheal tube size prediction in such patients is difficult. Age formula predicted an endotracheal tube size of 6.5 mm, but we could intubate the patient with a 5.5-mm cuffed, flexometalllic endotracheal tube. The operative site being near the brainstem, the left radial artery was cannulated for invasive blood pressure monitoring and an oesophageal temperature probe was inserted. Anesthesia was maintained with an air-oxygen mixture (1:1) and isoflurane, and intermittent boluses of vecuronium and fentanyl as needed.

After preloading with 200 ml of normal saline, the patient was positioned prone on pillows keeping the abdomen free for excursions and the pressure points were padded. Surgery was completed in about 2 h. Postoperatively the patient was turned supine and placed on a thick pillow in a position similar
to intubation. The neuromuscular block was antagonised and the trachea was extubated after complete awakening. The patient was shifted to the ICU for monitoring. Injection of paracetamol (500 mg intravenously six-hourly) provided postoperative pain relief. After an uneventful postoperative period, the patient was discharged after 3 days with advice for spinal extension brace, physiotherapy and follow-up.

Discussion

Acromesomelic dysplasia is an extremely rare skeletal disorder characterised by shortening of the middle and distal segments of the limbs.\[4\] This disproportion occurs at birth, becoming obvious during the first years of life, with diagnosis at around 3 years.\[3\] Literature is scarce regarding appropriate peri-operative management of patients with AMDM. Berkowitz et al.,\[4\] published a review of the pathophysiology of different types of dwarfs and their anesthetic implications. Although AMDM was not specifically mentioned, we followed their guidelines in the anesthetic management of this child.

Our concerns included the severe kyphosis and the resulting pathophysiological changes, an anticipated difficult airway, hydrocephalus, syringomyelia and surgery in prone position near vital brain structures. Kyphosis presented with back pain but was not associated with the changes in the pulmonary function tests. Murray et al.,\[6\] reported similar findings in their study of the natural history of Scheuermann kyphosis. Kyphosis, however, posed significant problems in securing the airway, which were resolved by modification of the intubation position.

Chiari malformation and hydrocephalus demanded stringent intracranial pressure management. Anesthetising a dwarf with raised intracranial pressure necessitates balancing the different anesthetic priorities and techniques. Inhalational induction may cause hypercarbia and increase in intracranial pressure. Conversely, intravenous induction, in the presence of a difficult airway, may cause apnoea or airway loss, which may be catastrophic. Our objective was to secure the airway with minimum manipulation and avoiding hypercarbia. Positioning using a thick pillow, confirmation of mask ventilation, gentle check laryngoscopy for glottic visualisation and use of intravenous drugs helped us attain this goal. Huang et al.,\[7\] suggested succinylcholine as the first choice in difficult airway cases. Due to hydrocephalus and mask ventilation feasibility, we avoided using it.

AMDM patients suffer from limited elbow extension and joint laxity.\[1\] Prone positioning requires carefully coordinated movements and paddling of the pressure points. Pulmonary hypertension is the most common cardiovascular complication in dwarfs.\[1\] Even though its presence could not be confirmed, we avoided nitrous oxide and used air with isoflurane instead.

To conclude, osteochondrodysplasias represent varied clinical syndromes of which AMDM is an extremely rare form presenting as an anesthetic challenge. In spite of an apparently normal airway, the thoracic kyphoscoliosis and gibbus deformity made conventional laryngoscopy and intubation difficult in our patient. A simple and innovative modification of the intubation position aided in airway management and prevented increase in intracranial pressure.

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