INTRODUCTION

Infantile Systemic Hyalinosis (ISH) is a rare autosomal recessive disease that normally presents itself within the first few months of life. It is painful, disfiguring and progressive in nature. Recent genetic breakthrough has identified the gene that encodes for capillary morphogenesis protein 2 (CMG2) as the affected site of mutation in these patients causing fibroblasts to deposit hyaline. Characteristically, patients suffer from painful thickened and nodular skin, hypertrophic gingiva, hypotonia, and profound limitation of body movement secondary to the diffuse deposition of hyaline. Additional concerns include osteoporosis, contractures of multiple joints, malnutrition and growth failure. Death usually results from recurrent pulmonary infections and diarrhea. These joint contractures may make airway management challenging as a result of cervical spine (c-spine) and TMJ movement limitations. Though ISH patients are intellectually normal, this case of a 10-month-old infant suffering from ISH presents with multiple contractures and subsequent limited movement of all limbs, micrognathia with limited mouth opening and head movement secondary to temporomandibular joint contracture.

ANESTHETIC MANAGEMENT

The anesthetic management for a patient with ISH is largely centered around airway management and positioning. Options for airway management in a patient with ISH are dependent on the degree of involvement of head and neck joints in regards to limitation of movement. Other options that have been described in the literature for management of a patient with ISH includes nasal airway use with concomitant oral fiberoptic intubation, nasotracheal intubation when oral intubation is impossible, and use of an LMA if mouth opening allows and if it is appropriate for the surgical procedure. Inhalation induction is considered safest with maintenance of spontaneous respirations. Positioning is also a point of important consideration for ISH patients. The contractures necessitate appropriate padding and bolstering in order to avoid neuropathy and pressure points. As the disease progresses, this becomes more critical as the patient becomes more contracted, less flexible and in considerable pain.

CASE

A 10-month-old 5.3 kg male presented for insertion of a tunneled venous catheter for delivery of antibiotics. He was failure to thrive and chronic diarrhea and returned to the PICU for further management.

DISCUSSION

A satisfactory outcome in this challenging case depends on the perioperative team’s ability to consider and be proficiently knowledgeable in the following:

1. The importance of airway assessment and identification of potentially difficult pediatric airway prior to operating room
2. The necessity of preoxygenation to increase time prior to desaturation
3. Avoidance of nitrous oxide during induction in order to maintain spontaneous respirations
4. The maintenance of spontaneous respirations when dealing with a known difficult airway syndrome
5. Back up airway devices made readily available and consideration of immediate use of advanced airway equipment
6. Appropriate positioning to avoid severe pressure points in these patients
7. Awareness of the likelihood of electrolyte abnormalities secondary to failure to thrive and chronic diarrhea
8. Communication between the surgeon and the anesthesiologist is vital

REFERENCES