Freeman-Sheldon Syndrome and General Anesthesia for Mandibular Osteotomy

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INTRODUCTION

Freeman-Sheldon syndrome (FSS) is a rare multiple congenital contracture syndrome. It is also a non-progressive or slowly progressive myopathy affecting the facial, limb, and respiratory muscles. Less than 100 cases have been reported to date. Its pathognomic sign is the “whistling face” due to microstomia and contractures of the facial muscles. Other facial features include down-slanting palpebral fissures, prominent nasolabial folds, high arched palate, a long philtrum, mandibular hypoplasia, and H- or Y-shaped creases in the chin. Patients with FSS also have contractures of the limbs resulting in club feet, camptodactyly of the hands, and severe scoliosis. As a result of these features, anesthetic concerns include difficult airway, difficult IV placement, risk of post-operative pulmonary complications, and reported increased risk of malignant hyperthermia (MH).

CASE REPORT

BL is a 3 week old, 3.3 kg female presenting for direct laryngoscopy, bronchoscopy, and bilateral mandibular osteotomy. Manifestations of Freeman-Sheldon syndrome include microstomia, arthrogryposis, microtia, bilateral ptosis. Severe OSA was diagnosed by polysomnography with an AHI of 43.7.

The anesthesia machine was prepared in the usual fashion for malignant hyperthermia precautions. The patient was sedated for laryngoscopy with propofol and dexmedetomidine, maintaining spontaneous respirations. The ENT surgeon made multiple attempts at laryngoscopy with a Hollinger laryngoscope and flexible and rigid endoscopes, noting an anterior glottic opening and an epiglottis that was difficult to lift. She was easy to mask ventilate throughout and oxygen saturation remained above 95%. The patient was intubated on a blind attempt with a 3.0 cuffed ETT, then maintained on propofol and remifentanil infusions for the remainder of the procedure.

She remained intubated electively due to the difficult intubation, severe OSA, and ongoing mandibular distraction. She remained intubated for 16 days until mandibular distraction was complete. After extubation, she intermittently required nasal CPAP and supplemental O2. BL returned to the OR on POD #65 for ptosis repair, G-tube placement, and repeat laryngoscopy and bronchoscopy. She was discharged on POD #72 on 0.5L O2 by nasal cannula.

Figure 1. Two patients with Freeman-Sheldon syndrome with the characteristic “whistling face” and arthrogryposis. From Stevenson et al, 2006.

CONCLUSION

Difficult intubation is a known risk in patients with FSS due to a very small mouth opening, mandibular hypoplasia, limited neck mobility, and muscle contractures. BL was difficult to intubate for all of these reasons. She was intubated once again at 2 months with difficulty by ENT with a Parsons laryngoscope, once with difficulty at 5 months with a grade 3 view by direct laryngoscopy, and then easily at 6 months with a grade 2 view by direct laryngoscopy.

 Patients with FSS may be at increased risk of MH, but this has not been proven. In one series, 3 of 19 patients who had general anesthesia developed MH and two more developed unexplained hyperpyrexia. There have been two reports of masseter muscle spasm after halothane and succinylcholine, one relieved by dantrolene, the other by termination of halothane. There is one case report of muscle rigidity and elevated CK after halothane and succinylcholine. It is thought that FSS may be an unconventional myopathy, which would explain a predisposition to MH. Though the risk of MH is unclear, it seems prudent to use a non-triggering anesthetic in these patients. It should be noted that this patient underwent four more anesthetics at our institution, three non-triggering and one with sevoflurane with no evidence of MH.

There is an increased risk of postoperative pulmonary complications such as aspiration, obstruction, pneumonia, and respiratory failure given intercostal myopathy, obstructive sleep apnea, and abnormal respiratory mechanics. These patients should be monitored in an ICU setting after anesthesia.

REFERENCES