Anesthesia for Freeman-Sheldon syndrome using a laryngeal mask airway

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Purpose: To present a case of Freeman-Sheldon syndrome (FSS) with a previously unreported technique of anesthetic management, consisting of a malignant hyperthermia free anesthetic and laryngeal mask airway.

Clinical features: Freeman-Sheldon syndrome (also known as whistling face syndrome, Windmill-Vane-Hand syndrome, cranio-carpo-tarsal dysplasia and distal arthrogryposis type 2) is a rare congenital disorder defined by facial and skeletal abnormalities. The three basic abnormalities are microstomia with pouting lips, camptodactyly with ulnar deviation of the fingers and talipes equinovarus. Patients with FSS frequently present for surgical correction of musculoskeletal or facial abnormalities. There are several anesthetic challenges including difficult airway, intravenous cannulation and regional technique. They may be at increased risk for malignant hyperthermia and postoperative pulmonary complications. We present a case of a two-year-old child with FSS undergoing elective unilateral inguinal hernia repair. A non-triggering anesthetic technique was used, consisting of 2 mg·kg⁻¹ propofol followed by a continuous infusion, nitrous oxide 50%/oxygen, and 3 µg·kg⁻¹ fentanyl. Intraoperative and postoperative analgesia was provided by an ilioinguinal nerve block with 10 ml bupivacaine 0.25% with epinephrine 1:200,000. The airway was maintained with a #2 laryngeal mask airway. The anesthetic was uneventful and there were no signs or symptoms of malignant hyperthermia. The patient was discharged home later the same day in good health.

Conclusion: The use of a laryngeal mask airway and non-triggering anesthetic technique should be considered as options for anesthetic management in patients with FSS for short procedures that do not require neuromuscular blockade.

Objectif : Présenter un cas de syndrome de Freeman-Sheldon (SFS) et une technique d'anesthésie inédite avec l'utilisation d'un anesthésique libre d'effet déclencheur d'hyperthermie maligne, et d'un masque laryngé.

Éléments cliniques : Le syndrome de Freeman-Sheldon (ou syndrome du siffleur, dystrophie ou dysplasie cranio-carpo- tarsienne) représente un ensemble rare de malformations congénitales intéressant la face et le squelette. Les trois principales anomalies sont la microstomie avec lèvres éversées, la camptodactylie avec déviation des doigts du côté cubital et les pieds bots varus équins. Ces anomalies musculo-squelettiques ou faciales exigent souvent une correction chirurgicale. L'anesthésie peut alors présenter certains obstacles: intubation, canulation intraveineuse et technique régionale difficiles. Les patients peuvent être à risque élevé d'hyperthermie maligne et de complications pulmonaires postopératoires. Nous présentons ici le cas d'un enfant de deux ans admis pour la réparation élective d'une hernie inguinale unilatérale. L'anesthésie sans effet déclencheur d'hyperthermie comprenait 2 mg·kg⁻¹ de propofol suivis d'une perfusion continue, d'un mélange de protoxyde d'azote et d'oxygène à 50 % et de 3 µg·kg⁻¹ de fentanyl. L'analgésie peropératoire et postopératoire a été assurée par un blocage du nerf ilio-inguinal avec 10 ml de bupivacaine 0,25 % et de l'épinéphrine 1:200 000. Le dégagement des voies aériennes a été maintenu avec un masque laryngé n° 2. L'anesthésie a été sans incident et sans signe ou symptôme d'hyperthermie maligne. L'enfant est retourné chez lui le même jour, en bonne santé.

Conclusion : L'utilisation du masque laryngé et d'une technique anesthésique sans effet déclencheur d'hyperthermie est un choix dont il faut tenir compte pour l'anesthésie de patients atteints du SFS lors d'interventions courtes qui ne nécessitent pas de blocage neuromusculaire.

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FREEMAN-SHELDON syndrome (FSS), first described in 1938,¹ is a rare congenital disorder defined by facial and skeletal abnormalities. It is also known as whistling face syndrome,² Windmill-Vane-Hand syndrome³ and cranio-carpo-tarsal dysplasia. Recently, FSS has been reclassified as distal arthrogryposis type 2.⁴ Many of the features of FSS may be due to an underlying myopathy.⁵

The syndrome is characterized by three basic abnormalities: microstomia with pouting lips, camptodactyly with ulnar deviation of the fingers and talipes equinovarus. Expression of these and other associated features is often variable. Other facial characteristics include flat "mask-like" facies, vertical skin folds in the jaw, microglossia, micrognathia, and a high arched palate. They may be prone to airway obstruction and have a nasal quality to their speech. Feeding problems may result from microstomia and difficulties with swallowing. The eyes are deeply set with hypertelorism, blepharophimosis and epicanthal folds. Strabismus may be present. The nose and nostrils are

small with a long philtrum. Radiological imaging of the skull may demonstrate a steep anterior cerebral fossa. Bilateral ulnar deviation and contracture of fingers 2-5 at the metacarpophalangeal joints with adduction of the thumbs define the "windmill vane" position of the hands. There is an association of undescended testes and inguinal herniae. Intercostal myopathy and kyphoscoliosis (often presenting later in life) may result in restrictive lung disease. Congenital cardiac disease is not increased in FSS. Intelligence, general health and life expectancy are usually normal.

The syndrome occurs sporadically, but autosomal dominant and recessive inheritance patterns have been described. Males and females are affected in equal numbers. It is usually recognized at birth, but has been diagnosed prenatally by ultrasound.⁶ Chromosomal analysis is normal. Electromyography and muscle biopsy may support the diagnosis. At present, there is no genetic testing specific for FSS. Recently, a variant of FSS (distal arthrogryposis type 2B) has been mapped to 11p15.5-pter.⁷

FIGURE 1 Frontal photograph of a patient with FSS showing characteristic facial features.

FIGURE 2 Lateral photograph of a patient with FSS.

Children with FSS frequently present for surgical correction of musculoskeletal or facial abnormalities. FSS provides several challenges for the anesthesiologist including difficult airway, intravenous cannulation and regional technique. They may be at increased risk for malignant hyperthermia and postoperative pulmonary complications. We present a case of FSS with a previously unreported technique of anesthetic management, consisting of a malignant hyperthermia free anesthetic and laryngeal mask airway.

Case History

A two-year-old male child weighing 13.9 kg with the diagnosis of Freeman-Sheldon syndrome was scheduled for elective left inguinal hernia repair (Figures 1,2). Detailed family history revealed that the patient, his brother, his mother, his maternal grandmother and her two brothers, one of her nieces and her father were all affected with FSS (Diagram). The pedigree is consistent with an autosomal dominant mode of inheritance. Prenatal diagnosis had been confirmed with ultrasound. Chromosomal analysis was normal. Overall, he had normal growth and development.

He had many of the classical features including microstomia with pursed lips and camptodactyly with ulnar deviation of the fingers. His face was mask-like with a prominent brow, hypertelorism, deeply recessed eyes and a small nose. Airway examination revealed severely limited mouth opening of approximately 1 cm, micrognathia, microglossia, normal dentition with slightly limited neck flexion and extension. The spine was straight. He had not undergone any previous anesthetics. He was receiving no medications and had no allergies.

A malignant hyperthermia free anesthetic machine and equipment were prepared. A fiberoptic bronchoscope was readily available. Standard monitors including non-invasive blood pressure cuff, ECG, pulse oximetry and axillary temperature were applied to the patient. Venous access was not difficult and a 22-G intravenous catheter was secured in the right hand with the patient awake. After preoxygenation with oxygen 100% for three minutes, anesthesia was induced with 2 mg·kg⁻¹ propofol. Spontaneous ventilation was easily maintained by mask. A #2.5 laryngeal mask airway (LMA) was too large to be placed past the patient's narrow oral aperture. A completely deflated #2.0 LMA was positioned and the cuff inflated without difficulty. Spontaneous ventilation was maintained with nitrous oxide 50%/oxygen and a propofol infusion of 120 µg·kg⁻¹·min⁻¹ titrated to clinical endpoints of heart rate and blood pressure. Fentanyl was titrated in 1 µg·kg⁻¹ boluses to a total of 3 µg·kg⁻¹. Prior to incision, an ilioinguinal nerve block

DIAGRAM The family pedigree consistent with an autosomal dominant mode of inheritance. (■ = affected)

was performed using 10 ml bupivacaine 0.25% with 1:200,000 epinephrine. Surgery was uneventful and lasted approximately 30 min. There were no signs or symptoms of malignant hyperthermia. At the end of the case, the propofol infusion and nitrous oxide were discontinued and the patient breathed oxygen 100%. The laryngeal mask was removed in the operating room when the patient demonstrated spontaneous eye opening and purposeful movement. The patient was transferred to the recovery room and discharged home later the same day in good health.

Discussion

Patients with FSS provide several anesthetic challenges. The most obvious concern for the anesthesiologist is the difficult airway. This may result from severe microstomia, micrognathia, small nasal passages, kyphoscoliosis and upper airway obstruction. The rami of the mandible may be hypoplastic and the malar bones small. Diffuse fibrosis of the orbicularis oris muscle and dermal fibrous bands along the vermillion border of the lower lip may be responsible for the contracted oral aperture. Neuromuscular blockade may have very limited effect on these fibrous changes.⁸

Several case reports have outlined approaches to airway management in FSS. All cases have emphasized either difficult or failed intubation in these patients. The first to report anesthetic difficulties was a case of FSS in a father and son.¹⁰ Intubation was described as impossible in the child, and achieved with great difficulty in the father. The case presented by Tateishi *et al.*¹¹ ascribed the difficulty of intubation to "limitation in elevating the rigid epiglottis". Duggar *et al.*⁸ reported an unsuccessful attempt at awake, blind nasotracheal intubation in a seven-month-old child with FSS. Awake orotracheal intubation was achieved with difficulty. Laishley *et al.*¹² presented three cases in which intubation was achieved

with considerable difficulty following inhalational induction with halothane. Vas *et al.*¹³ reported a case of FSS combined with cervical kyphoscoliosis that was induced with a combination of thiopental, halothane/ether, paralyzed with succinylcholine and the trachea was intubated with difficulty. Both blind¹⁴ and fiberoptic¹⁵ intubation through a laryngeal mask airway has been successful in patients with FSS.

The LMA has been used in children with difficult airways (Pierre Robin, Treacher Collins,¹⁶ Goldenhar,¹⁷ cri-du-chat, cleft palate¹⁸). Several variations on the original advice of Brain have been suggested for insertion including partial inflation and rotation. Given the minimal mouth opening of patients with FSS, complete deflation and midline insertion of the LMA may be the most intuitive approach to successful placement. This technique was used in our patient. Securing the airway with a fiberoptic guided intubation through the LMA was considered, but it was decided that ventilation was easily maintained, muscle relaxation was not required and this was a minor, short procedure in an otherwise well child. With this in mind, the LMA will not protect against laryngospasm, aspiration or airway obstruction. Maintenance of the airway by mask was considered, but we felt that the LMA would provide a more secure airway. The routine use of the LMA in children undergoing elective groin surgery has been well described.¹⁹

The underlying myopathy of FSS may predispose patients to malignant hyperthermia. Tachycardia and hyperpyrexia (38.7°C) was reported in one patient following administration of halothane.¹² This was attributed to the use of atropine, poor infantile temperature regulation and a traumatic intubation and not to malignant hyperthermia. Arterial blood gas analysis was normal. Sobrado *et al.*²⁰ described a case of generalized muscle rigidity in a two-year-old patient with FSS after anesthetic induction with halothane and succinylcholine. Severe muscular rigidity that compromised ventilation occurred after 10 min. Tracheal intubation was impossible due to masseter muscle rigidity. Successful treatment required dantrolene. Jones *et al.*²¹ presented two patients with FSS that developed muscle rigidity following halothane anesthesia. One patient required dantrolene, bicarbonate and supplemental oxygen to relieve muscle rigidity and treat the metabolic acidosis. In the other case, discontinuing halothane was sufficient to treat the masseter spasm. Serum creatine phosphokinase peaked at 9,215 IU·l⁻¹ and 1,193 IU·l⁻¹, respectively, the following day. The authors concluded, "halothane and succinylcholine should not automatically assumed to be safe in patients with FSS".

Although clinically suspicious for MH, none of these cases underwent muscle biopsy. The combination of halothane and succinylcholine has been known to trigger masseter muscle rigidity in up to 1% of children.²²

Although controversial, we chose to minimize the risk and proceed with a non-triggering anesthetic technique consisting of propofol, nitrous oxide, fentanyl and regional nerve block. Intravenous cannulation may be difficult in patients with FSS due to thickened subcutaneous tissue and frequent extremity surgery. Intravenous access was not difficult in our case. If this had proven to be a problem, we would have considered sedation with 0.5 mg·kg⁻¹ midazolam *po* or inhalation of nitrous oxide 50% in oxygen suitable to secure an *iv*. We felt that an inhalational induction with a potentially MH triggering agent would not have been a suitable alternative to *iv* induction.

Patients with FSS may be at increased risk for postoperative pulmonary complications. Pneumonia, empyema, respiratory insufficiency and recurrent respiratory infections have been reported.^{1,5,8} These may result from intercostal myopathy and loss of lung volume. Upper airway obstruction severe enough to require neonatal tracheostomy on day 17 of life has been described.⁹

An ilioinguinal block was used because of surgical preference. Caudal anesthesia is another option for intraoperative and postoperative pain relief and has been described in patients with FSS.^{8,15} A pure regional technique would not have been possible in our case due to the young age of the patient and difficulty with cooperation. Coexisting kyphoscoliosis, distorted anatomy and an increased incidence of spina bifida occulta may complicate regional techniques.¹³

Freeman Sheldon syndrome is a rare disorder that provides many anesthetic challenges. Affected individuals generally have a normal life span and may present for a variety of surgical interventions requiring anesthetic management. We presented a case of FSS managed with a non-triggering anesthetic technique consisting of propofol, fentanyl and nitrous oxide. Intraoperative and postoperative pain relief was with an ilioinguinal nerve block. The airway was managed with a laryngeal mask airway. These options should be considered in the anesthetic management of patients with FSS who are scheduled for short procedures that do not require neuromuscular blockade.

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