Freeman Sheldon Syndrome (FSS)

Freeman Sheldon syndrome (FSS) is a rare congenital myopathy and dysplasia characterised by multiple contractures of face, chest and limbs. The musculoskeletal and soft tissue deformities require frequent orthopaedic, ocular and plastic reconstructive surgeries for improving the quality of life. The combination of difficult airway, poor venous access and possible susceptibility to malignant hyperthermia make anaesthetic management in these infants extremely challenging.

We report a case of a 2 year old child with Freeman Sheldon Syndrome presenting for bilateral inguinal herniotomy. A non trigerring anaesthetic technique consisting of propofol, fentanyl and N2O was used. The anticipated difficult airway was secured with laryngeal mask airway. Intraoperative and postoperative analgesia was provided with bilateral ilio inguinal nerve block. The patient was discharged home later the same day in good health.

Introduction

Freeman-Sheldon syndrome is a rare non progressive or slowly progressive myopathy with dysplasia involving the facial, limbs and respiratory muscles. The syndrome was first described in 1938 by Ernest Freeman, a British orthopaedic surgeon, and Joseph Sheldon, a British physician.. Electromyography and muscle biopsy may support the diagnosis. It is usually recognized at birth, but has been diagnosed prenatally by ultrasound in a patient with a strong family history. Chromosomal analysis is normal. The dysmorphic features are attributed to the underlying myopathy which may show varying degrees of weakness, increased tone or fibrosis. The syndrome is characterized by three basic abnormalities: microstomia with pouting lips, camptodactyly with ulnar deviation of the fingers and foot abnormalitie (talipes equino varus). Fibrotic contractures of the facial musculature and other soft tissues cause the patient to have a mask like facies with circumoral fibrosis and microstomia (whistling face).

Bilateral ulnar deviation and contracture of 2-5 fingers at the metacarpophalayngeal joints with adduction of the thumb define the ‘windmill vaale’ position of the hands. FSS is also known as the craniocarpotarsal syndrome in view of the body parts affected; or “whistling face syndrome” describing the typical pursed mouth. Windmill-Vane-Hand syndrome and distal arthrogryposis type 2A have also been used alternatively. Systemic nature of this condition often poses a challenge to anaesthesiologists in airway management, intravenous access, regional anaesthesia and body positioning. Other anaesthetic issues include a possible risk of malignant hyperthermia, postoperative pulmonary complications and an unpredictable response to neuromuscular blocking agents.

Case report

A 2 year old male child weighing 10 kgs presented with bilateral inguinal hernia. His face was notably expressionless and his mouth was puckered as if whistling. Airway assessment revealed a limited mouth opening, interincisor distance of 1.5 cm and mallampatti grade IV indicative of anticipated difficult intubation by direct laryngoscopy. The thyromental distance and neck movements were normal. His alae nasi were hypoplastic and a dimple on chin was noted.

He had deep set eyes with hypertelorism and low set ears. Wrists showed ulnar deviation and flexion contractures of fingers. The skin over his wrists was smooth and no veins were visible. The child had a modest degree of developmental delay. There was no history of seizures. Examination of the spine revealed mild scoliosis involving the lower thoracic and lumbar spine. He had not undergone any previous anaesthesia exposures. Past medical history was unremarkable. There was no relevant family history. Patient was scheduled for an elective bilateral herniotomy.

He was referred to the child developmental clinic and was diagnosed as a case of Freeman Sheldon Syndrome. Preoperative haematological and biochemical investigations were normal. He was ASA physical status II on preoperative clinical assessment. General anaesthesia supplemented with regional anaesthesia was planned. Written informed consent was obtained from the parents of the child after explaining the risks involved. Before the conduct of anaesthesia, written informed consent was also obtained for the publication of the selected photographs of the child in scientific journals.

Appropriate size airway, laryngoscope blade, endotracheal tube, emergency tracheostomy tray were kept ready for emergency airway management. A fibreoptic bronchoscope (FOB) was readily available. A malignant hyperthermia free anaesthesia machine was prepared. EMLA cream under an occlusive dressing was applied on a suitable leg vein 1 hour before intravenous cannulation. Baseline vitals were recorded with non invasive monitors like ECG, pulse oximeter, nonIABP, axillary thermistor. O2 was insufflated by a face mask held above the face of child with 50% O2 N2O mixture. 24 G IV cannula was secured over the Eutectic mixture of local anaesthetic (EMLA) applied limb.

Anaesthesia was induced with intravenous injections of 2mcg/kg fentanyl and 2mg/kg propofol. We chose a proseal laryngeal mask airway for airway management to preserve spontaneous ventilation. The appropriate depth of anaesthesia was confirmed by jaw relaxation, loss of eyelash reflex and loss of verbal contact before attempting to insert the PLMA. There was no improvement in the mouth opening even after jaw relaxation due to congenital anatomical anomaly. A lubricated completely deflated PLMA size 2 was easily inserted inside the pharynx. The PLMA cuff was inflated till the OSP of 50 cm H2O. Correct placement of PLMA was confirmed with capnography and chest auscultation.

Regional anaesthesia was provided with USG guided bilateral ilioinguinal nerve block using a high frequency linear probe. 2 ml of 0.25% bupivacaine was used on each side for nerve block. 150 mg paracetamol was given intravenously. Anaesthesia was maintained with 50% N2O in 02 and propofol infusion @ 3mg/kg/hr on PCV. Muscle relaxants were avoided. On completion of surgery, N20 and propofol infusion were stopped. Intraoperative vitals including ETCO2 and temperature remained stable and there were no signs and symptoms of malignant hyperthermia. Surgery was uneventful and lasted 1.5 hrs. At the end of surgery, LMA was removed when the patient was awake with return of reflexes. The postoperative period till the hospital discharge was uneventful.

Discussion

FSS is a congenital myopathy manifesting as fibrotic contractures of facial, respiratory and limb musculature. A triad of physical features has been depicted - microstomia with pouting of lips,

camptodactyly, with ulnar deviation of fingers and talipes equinovarus. These patients pose several anaesthetic challenges. The most obvious concern for the anaesthesiologist is the difficult airway. This may result from severe microstomia, micrognathia, limited neck mobility, small nasal passages, kyphoscoliosis and upper airway obstruction. The microstomia and pursed lips are thought to be due to diffuse fibrosis within the orbicularis oris muscle and a fibrous band along the vermillion border of the lower lip. There is often an associated high arched palate and mandibular hypoplasia. Muscle contractures also limit neck mobility. The combination of all these features make laryngoscopy and intubation extremely challenging. Neuromuscular blockade may have a very little effect on these fibrous changes.

However microglossia and a high arched palate allows proper placement of LMA. Apart from the whistling facial expression, the face is virtually expressionless. Facial contractures tend to push the developing lower incisors lingually and a mound of soft tissue results in vertical furrows on the chin. This characteristic skin dimple may be either H or Y shaped. Hypertelorism with deep set eyes and short downslanting palpebral fissures are common and may be associated with strabismus, mild ptosis or exotropia. The ears may be low set and there may be a hearing deficit. The cartilage of the nose is underdeveloped and hence the nose is small with hypoplastic upturned and notched alae nasi (nasal coloboma). The philtrum is usually long.

The tongue may be small and the limited movement of soft palate may cause a nasal speech. Feeding problems may result from microstomia and difficulties with swallowing. The pharyngeal muscles may also be affected posing a risk of upper airway obstruction, gastroesophageal reflux and aspiration. They are at increased risk of postoperative pulmonary complications (POPC) like pneumonia, empyema, respiratory insufficiency and recurrent respiratory infections. These may result from intercostals myopathy, loss of lung volume, abnormal respiratory mechanics (scoliosis, pectus excavatum, rigid immobile thoracic cage). Sleep apnea and cor pulmonale due to chronic airway obstruction have also been reported.

The underlying myopathy of FSS may predispose these patients to malignant hyperthermia. A suggested association between FSS and malignant hyperthermia is not supported by review of literature, though abnormal responses to suxamethonium and volatile agents are well described. So a nontriggering anaesthetic technique without using halogenated inhalation agents and suxamethonium was used. They are at an increased risk of postoperative pulmonary complications due to unpredictable responses to neuromuscular blocking agents. Also neuromuscular blockers were avoided to avoid the unpredictable muscle paralysis and possible persistent postoperative muscle weakness contributing to POPC. (Thus the perioperative anaesthetic management in FSS can be uneventful with complete avoidance of halogenated inhalational anaesthetic agents, suxamethonium and neuromuscular blocking agents as observed in our patient.)

So our technique of securing airway with LMA and maintaining spontaneous ventilation with a nontriggering anaesthetic technique is well suited for these patients. But at the same time, appropriate size airway, laryngoscope blade, ETT, FOB and emergency tracheostomy should be kept ready for emergency airway management. Postoperative complications like respiratory insufficiency, upper airway obstruction can be decreased by minimal use of systemic sedatives and opioids along with successful regional anaesthesia. Hence peripheral nerve locator and USG guided blocks are useful for regional anaesthesia. So an ilioinguinal nerve block was used in our patient. Caudal anaesthesia would have been another option for intraoperative and postoperative pain relief.

However contracture and limited joint movement can make access to peripheral nerves difficult. Spinal anesthesia and combined spinal epidural anaesthesia have been describes foe lower limb surgery in older children with FSS to avoid potential airway complications from anticipated difficult tracheal intubation. However scoliosis, vertebral anomalies and spina bifida occulta may be considered a contradiction to central blockade. Deformities of hands and feet combined with thickening of subcutaneous tissues and frequent extremity surgery can result in difficult venous access, although this was not a problem in our patient. Even central venous access may be difficult in view of limited neck movement of the short neck. The generalised myopathy associated with FSS has been implicated in the development in the development of scoliosis (often presenting later in life), short stature, pectus excavatum and lower limb contractures. Many of the features of FSS persist and evolve with development. There is an association with undescended testis and inguinal hernia. Congenital cardiac disease is not increased in FSS. Intelligence, general health and life expectancy are usually normal.

Conclusion

Every syndrome complex has a unique cluster of abnormalities. FSS also has such manifestations which are particularly challenging from anaesthesiologists perspective. The difficult airway, intravenous cannulation, regional anaesthesia and body positioning pose a challenge to the anaesthesiologist. Also the susceptibility to malignant hyperthermia and POPC pose additional challenges. The use of LMA and a non triggering anaesthesia technique should be considered as options for anaesthetic management in patients with FSS for short procedures that do not require muscle relaxation.

New, interesting and rare cases can be reported. They should be unique, describing a great diagnostic or therapeutic challenge and providing a learning point for the readers. Cases with clinical significance or implications will be given priority. These communications could be of up to 1000 words (excluding Abstract and references) and should have the following headings: Abstract (unstructured), Key-words, Introduction, Case report, Discussion, Reference, Tables and Legends in that order.