Anaesthesia and Freeman Sheldon Syndrome

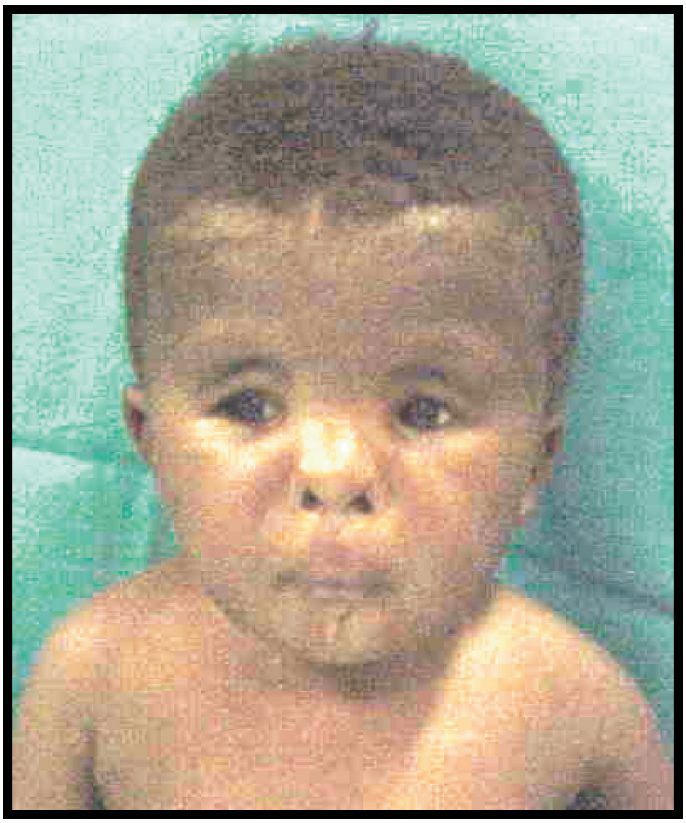
A Bösenberg, *MBChB, DA(SA) FFA (SA)*
Department of Anaesthesia, University of Cape Town

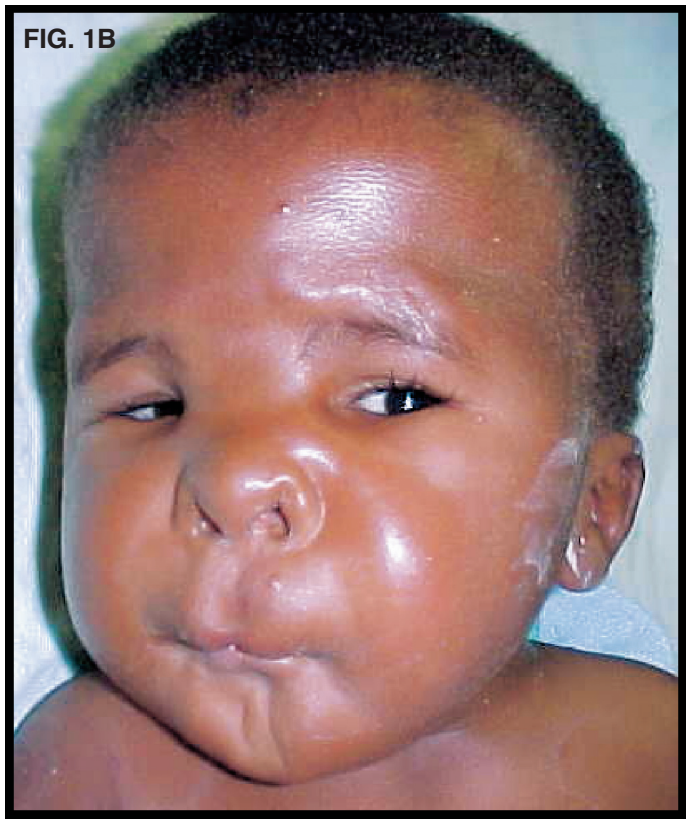
Synopsis of patients

Case 1

An 8 month old male, weighing 8.2kg, presented on three separate occasions for correction of bilateral talipes equinovarus and flexion contracture of his wrist. His face was notably expressionless and his mouth puckered as if whistling. His mouth, which was 20mm in width, was limited to 15mm opening while awake. (Fig.1a and b) Neck movement was limited in flexion and extension. Mild ptosis, hypoplastic alae nasi and a dimple on the chin were noted. The skin over his wrists was smooth and no veins were visible. His wrists, elbows and ankles were stiff. He had cryptorchidism. Chest was clear. Blood results were unremarkable and his haemoglobin was 10gm.

Case 1: Figure 1a as an 8th month old male and 1b: as a 22 month old, who required surgery on three occasions for congenital talipes equinovarus correction and contracture of the right wrist. The facial features are prominent even at a young age (1a)





Case 2

A 22 month old Mauritian girl, weighing 11.2kg, presented for correction of a craniofacial defect (brachycephaly). She showed similar facial features and her lips were puckered as if whistling. She also had limited mouth opening and a chin dimple(Fig 2). The neck appeared short and movement was limited, particularly in extension. The chest was an abnormal shape but clear on auscultation. There was no history of sei-

Correspondence :
Prof A Bosenberg
email: bosie@cormack.uct.ac.za

zures and her intelligence was normal. Her wrists, which showed ulnar deviation, and her ankle joints were stiff. Her muscle tone was generally increased. Flexion contractures of the fingers were also present.

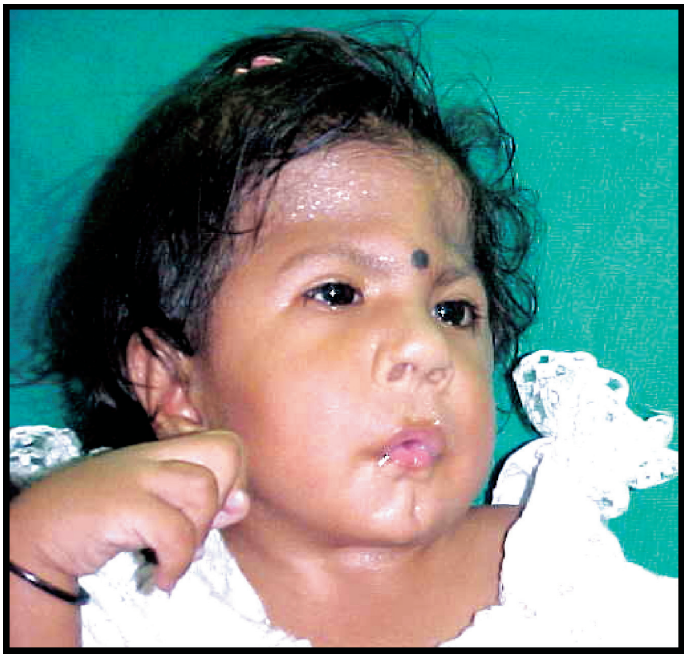


Figure 2: Case 2: A 22 month old female, who required craniofacial surgery, demonstrating the typical facial features and flexion deformity of her right wrist

Freeman Sheldon syndrome

Freeman Sheldon syndrome is rare nonprogressive or slowly progressive myopathy where the facial, limb and respiratory muscles are primarily affected. The majority occur sporadically but autosomal dominant and recessive inheritance patterns have been described. Males and females are equally affected. The exact gene involved has not been identified and there is no prenatal diagnostic test available at present. Electromyography and muscle biopsy may support the diagnosis.

The syndrome occurs worldwide and was first described in 1938 by Ernest Freeman, a British orthopaedic surgeon, and Joseph Sheldon, a British physician.[1] The triad of physical features has been depicted in art including a pre-Columbian vase and in ceremonial objects and folklore of the aboriginal peoples of the Canadian Northwest. These features include fibrotic contractures of the facial muscles giving rise to the mask like whistling facial expression; ulnar deviation of 2nd to 5th digits with adduction contractures of the thumb ("windmill vane position"); and foot deformities (talipes equino varus). These limb deformities are similar to those seen in arthrogryposis multiplex congenita.

Freeman Sheldon syndrome is also known as the craniocarpotarsal syndrome in view of the body parts affected; or "whistling face syndrome" describing the typical pursed mouth.[2] Windmill-Vane-Hand syndrome and distal arthrogryposis type 2A have also been used.

The generalised myopathy is implicated in the development of scoliosis, short stature, pectus excavatum, intercostal myopathy, inguinal hernia and undescended testis.[5,6] Development may be delayed but intelligence is usually normal (sufficient to become a paediatric anaesthesiologist at University of Washington!! -personal communication). Many of these features persist into adulthood unless surgical correction has been undertaken.

Apart from the 'whistling mouth' other dysmorphic features are expressed in varying degrees and are recognisable at birth. (Figs 1a,b and 2) Prenatal diagnosis has even been made by ultrasound in a family with a strong family history.[4] The dysmorphic features are attributed to the underlying myopathy which may show varying degrees of weakness, increased tone or fibrosis. Apart from the whistling facial expression, the face is virtually expressionless. Hyperteleorism, with deep set eyes and short downslanting palpebral fissures are common,[5] and may be associated with strabismus, mild ptosis or exotropia.[5] The ears may be low set and there may be a hearing deficit. The cartilage of the nose is under developed and hence the nose is often small with hypoplastic upturned and notched alae nasi (nasal coloboma). The philtrum is usually long.

Airway

The microstomia and pursed lips are thought to be due to diffuse fibrosis within the orbicularis oris muscle and a fibrous band along the vermilion border of the lower lip.[6] 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. There is often an associated high arched palate and mandibular hypoplasia. Muscle contractures may also limit neck mobility. The combination of all these features make direct laryngoscopy and intubation extremely challenging.[3, 6-17] Muscle relaxants have little or no effect on the circumoral architecture[6,17] neither does surgery to the mouth necessarily improve subsequent intubating conditions.[9]

The tongue may be small and the limited movement of the soft palate may cause nasal speech. Feeding problems may result from microstomia and difficulties with swallowing.[9,17] The pharyngeal muscles may also be affected posing a risk of upper airway obstruction,[12,13] gastroesophageal reflux and aspiration.

These patients are therefore at risk of pneumonia not only on the basis of intercostal myopathy, abnormal respiratory mechanics (scoliosis; pectus excavatum; rigid, immobile thoracic cage)[9] but also aspiration. Sleep apnoea has been described.[14] Cor pulmonale may develop on the basis of chronic upper airway obstruction, sleep apnoea and/or recurrent chest infections.[9] Postoperative pneumonia[1] and death following airway obstruction 13 days postoperatively[11] have been reported.

Regional anaesthesia has been recommended by some authors since the use of opiates may contribute to the risk of

postoperative airway obstruction, sleep apnoea and pneumonia. However contractures and limited joint movement can make access to peripheral nerves difficult. The use of a nerve stimulator is recommended. Scoliosis, vertebral anomalies and spina bifida occulta[16] may be considered a contraindication to central blockade.

Risk of malignant hyperthermia. There is a possible link to malignant hyperthermia although this seems somewhat tenuous. Two authors reported masseter muscle spasm,[7,15] and one generalised muscle rigidity with elevation of creatinine phosphokinase,[7] following exposure to halothane or suxamethonium[7,15] The masseter spasm was relieved by dantrolene[7,15] or termination of halothane.[7] Intraoperative pyrexia has been reported in another but this was not considered to be MH related.[9] (Table 1). This inconsistent response may reflect the genetic heterogeneity of the syndrome.

Halothane and sevoflurane have been used with impunity in most cases reported in the literature, but it seems these patients should be considered "potentially susceptible"[6] in view of the three cases described earlier. Patients should be monitored closely and should switch to a non triggering anaesthetic when indicated. Some authors advocate a non triggering anaesthetic ab initio.[7,15] However this is not as simple as it sounds because the "arthrogryposis-like" deformities of the hand and feet, combined with smooth featureless skin overlying these joints, make venous access extremely difficult. Even central venous access may be difficult in view of the limited movement of the short neck.

Table 1.
Reported cases of Freeman Sheldon syndrome in the anaesthetic literature outlining the anaesthetic given and the complications that occurred.

Author	No	Surgery	Anaesthesia	Complications	MH	Airway
Freeman		Ortho		Pneumonia		
Laishley	3 (9)	Various	Halo,N20, Thio Sux Halo	Nil	Temp (1) No	Awake Stylet
Munro	1	Clubfoot	Halo, caudal	Nil	No	LMA; FO
Duggar	1	Clubfoot	Halo, caudal	Nil	No	Awake
Mayhew	1	Squint	Halo,	Nil	No	Topical
Yamamoto	1	Cosmetic	Sevo	Nil	No	FO
Namiki	1	Clubfoot	Sevo	Nil	No	Direct
Okawa	1	Cosmetic	Sevo	Died postop		Nasal FO
Bosenberg	2 (6)	Ortho(3) Cosmetic	Propofol, caudal Propofol, block	Nil	No trigger No trigger	Bougie Bougie
Cruickshank	1 (R)	Ing hernia	Propofol, block	Nil	No	LMA
Jones	1 1 (5)	Cosmetic	Halo, sux Halo	Elevated CPK Elavated CPK	MS MR - Dant	Difficult oral
Sobrado	1	Diaphrag	Halo Sux		MS - Dant	Failed

MH = malignant hyperthermia MS = masseter spasm MR = muscle rigidity
LMA = laryngeal mask airway. Dant = dantrolene FO = fibreoptic scope

Summary

Freeman Sheldon syndrome is a rare progressive myopathic disorder affecting the face, chest and limbs. Multiple deformities require frequent cosmetic and orthopaedic surgical interventions. The combination of difficult airway, poor venous access and possible susceptibility to malignant hyperthermia make the anaesthetic management in these infants extremely challenging. They are prone to upper airway obstruction and postoperative respiratory complications.

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