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CORRESPONDENCE AND COMMUNICATION

Freeman-Sheldon syndrome: a functional and cosmetic correction of microstomia☆

Freeman-Sheldon syndrome (FSS) is a rare form of multiple congenital contracture syndrome and is the most severe form of distal arthrogryposis.[1,2] Described in 1938 by Freeman and Sheldon, it is also referred to as distal arthrogryposis type 2A,[1,2] craniocarpotarsal dysplasia or whistling face–windmill vane hand syndrome.[2] Its main form of inheritance is autosomal dominant, but it can also be autosomal recessive or X-linked[2–4] with neither sex nor ethnic preference.[2] Up until 1990 only 65 patients had been reported in literature.[2,4] The syndrome is limited to the musculoskeletal systems.[2] The primary deformities are the result of myopathic arthrogryposis.[3] Stevenson et al. published strict diagnostic criteria for FSS, which include: two or more major defects plus the following – microstomia, whistling face, prominent nasolabial creases and 'H-shaped' chin dimple[1,2] (Figure 1a). Major manifestations include: ulnar deviation of wrists and fingers; camptodactyly, hypoplastic and/or absent flexion creases, and/or overriding fingers at birth; talipes equinovarus (therapy resistant); calcaneovalgus, vertical talus, and/or metatarsus varus.[1,4] Other common facial findings include: flatness of the middle part of the face, microstomia, deep-set eyes, epicanthal folds, hypoplastic alae nasi, long philtrum and mask-like immobility[1,3,4] (Figure 1a).

In spite of these abnormalities the patients have normal mental development, and a normal life expectancy.[1,4]

A 1-year-old boy presented to us with difficulty in opening his mouth, difficulty in feeding, struggling to maintain oral hygiene and delayed speech. According to the Stevenson's criteria our patient qualified as having FSS. The intercommisural distance measured 18 mm, with stenosis of the oral sphincter.

The microstomia was treated first. Tracheal intubation was difficult, so fibre optic-assisted nasal intubation was performed after inhalation induction without the use of neuromuscular blocking agents.

The surgical procedure was as follows:

1. Marking of the neocommissure: perpendicular line dropped from midpupil $\pm$ 1 cm lateral to the commissure.

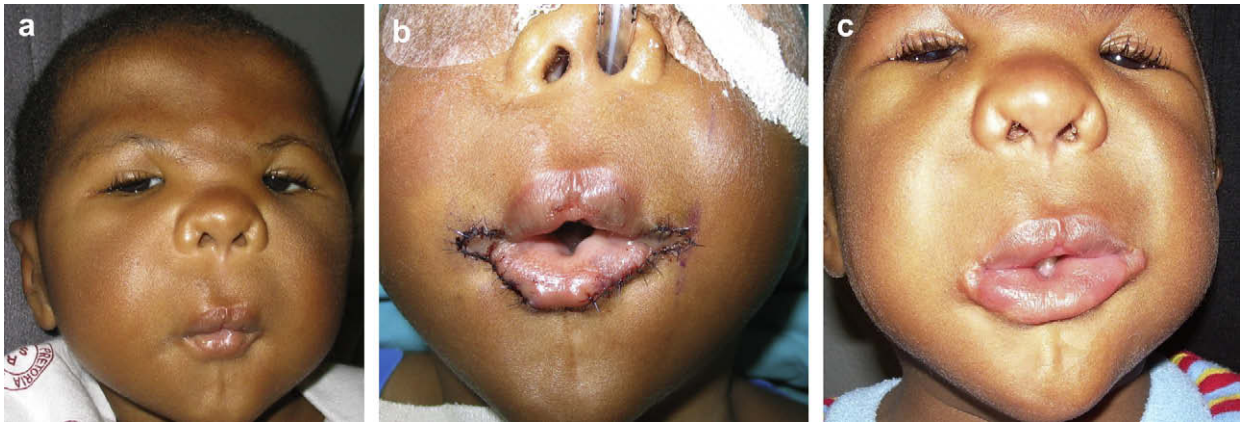

Figure 1 (a) Preoperative. (b) Intraoperative. (c) One year postoperative.

☆ This case study was presented at the Annual Congress of the Plastic and Reconstructive Surgeons of South Africa (APRSSA), 24 October 2007.

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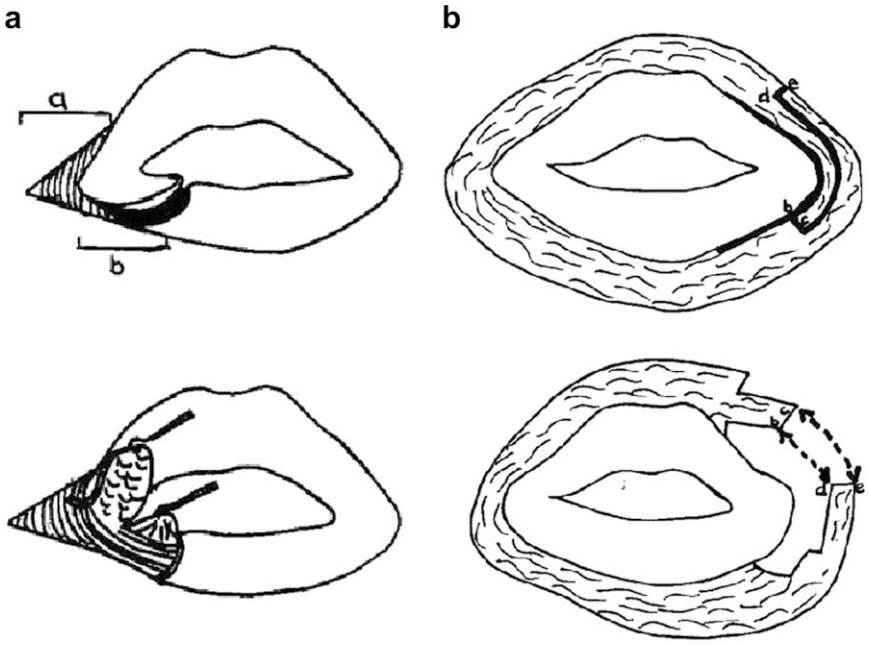

Figure 2 (a) Vermillion flap for neocommisure (a = b). (b) Lengthening of orbicularis oculi muscle.

2. Marking of the vermillion on the lower lip (1 cm from the commissure) for reconstruction of the upper lip vermillion.
3. Vermillion flaps raised (Figure 2a).
4. Exposure of the orbicularis oris muscle and section thereof according to the diagram (Figure 2b).
5. Suturing of points bc to de with absorbable suture (Figure 2b).
6. Lower lip vermillion used to reconstruct upper lip vermillion (Figure 2a).
7. Donor area on lower lip covered with buccal mucosa advancement flaps (Figure 2a).

Postoperatively no respiratory complications were experienced. The mother was encouraged to help the child with oral hygiene and feeding. The patient was discharged after four days with a satisfactory result.

Our goal was to widen the oral aperture in a functional and aesthetically acceptable manner without recurrence. Previous authors reconstructed the vermillion deficit of the upper and lower lip using mucosal advancement flaps.[5] We felt that the upper lip is the aesthetically more important part of the mouth, and because of its projection is more visible, hence using lower lip vermillion in correction of upper lip vermillion deficit. With this procedure we widened the intercommissural distance from 18 to 35 mm. With later follow up, there was no recurrence or contracture of the orbicularis oris muscle and an aesthetically pleasing result was obtained (Figure 1b, c).

References

1. Stevenson DA, Carey JC, Palumbos J, et al. Clinical characteristics and natural history of Freeman-Sheldon syndrome. *Pediatrics* March 2006;**117**:754–9.
2. Freeman-Sheldon syndrome [online] Available from: http://en.wikipedia.org/wiki/Freeman-Sheldon_syndrome [accessed 24.05.07].
3. Senen D, Adanali G, Tuncel A, et al. Whistling face syndrome associated with camptodactyly. *Ann Plast Surg* 2002;**48**:339–40.
4. Wiedemann HR, Kunze K, Dibbern H. *Atlas of Clinical Syndromes, a Visual Aid to Diagnosis*. 2nd ed. St Louis: Mosby Year Book; 1992. 46–47.
5. Ferreira LM, Minami E, Andrews J. Freeman-Sheldon syndrome: surgical correction of microstomia: a case report. *Br J Plast Surg* 1994;**47**:201–2.

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