CASE REPORT

Improving oral function and cosmesis in a case of Freeman-Sheldon syndrome

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SUMMARY

Children born with the rare congenital condition Freeman-Sheldon syndrome (FSS) have a characteristic facial appearance: microstomia, a long philtrum, 'H-shaped' chin abnormality and 'pinched lips' in addition to extra-facial features such as kyphoscoliosis and hand deformities. Such children often have problems with oral continence and difficulties with speech leading to both nutritional and psychosocial concerns. Prompt correction through surgery is therefore important in effectively managing the condition. We report the case of a 7-year-old girl who presented with the appearance of scarred lips with difficulties in closing her mouth, a speech impediment and oral incontinence. Using a muscle-sparing technique to reconstruct the upper and lower lips, better oral continence was achieved alongside markedly improved speech and an aesthetically pleasing result. Taken together, this case aptly illustrates the benefits of lip reconstruction in improving function and cosmesis in patients with FSS.

BACKGROUND

Freeman-Sheldon syndrome (FSS) is a rare genetic condition, in which children are born with multiple congenital contractures (MCCs),¹ resulting in impaired flexion/extension of certain areas of the body. The disorder itself was first described in 1938 as 'cranio-carpo-tarsal dystrophy'² by Freeman and Sheldon and later as 'whistling face syndrome' by Burian in 1963.³ The latter refers to the strikingly small oral orifice (microstomia), which gives the appearance of someone whistling.

Patients with FSS have characteristic facial and extra-facial features that mean it is well-known among other distal arthrogryposes. While FSS includes a range of abnormalities, poor lip closure and function can lead to significant functional problems with speech and eating/drinking alongside psychological and cosmetic issues. Early surgical correction can help to limit the damaging effects of sustained poor nutrition and also helps to minimise the psychosocial impact of the associated facial abnormalities.⁴

This case illustrates the functional and psychological impact of FSS on children, while describing a surgical technique to try and address the appearance of lip scarring and rectify poor lip closure.

CASE PRESENTATION

A 7-year-old girl was diagnosed with FSS shortly after birth, after review by clinical genetics. She exhibited various physical features of FSS including microstomia and bilateral clubfoot and had

previously undergone reconstructive surgery for the latter. She presented to our unit with the appearance of scarring of her upper and lower lips, together with oral incontinence and difficulties with speech. She did not have any dysphagia, vomiting or regurgitation. An informal assessment by the speech and language therapy team found she had problems in producing bilabial consonants leading to significant problems with intelligibility and communication. Importantly, this had led to significant teasing at school and concomitant parental concerns. There was no other relevant history; she had achieved normal developmental milestones.

On examination, she had significant attenuation of the orbicularis oris muscle in the central area of the lower lip, while the muscle was also much thinner than average in the upper lip. Consequent defective oral sphincter function led to difficulty in achieving oral continence and intelligible speech. This midline muscle defect gave rise to the characteristic 'whistling face' (figure 1). Furthermore, the patient had a number of skin dimples, depressions, unusual adhesions and accessory frenula in the perioral area. Apart from the features mentioned, examination was normal and she was alert and looked well.

TREATMENT

The patient underwent oral sphincter muscle reconstruction to improve continence and speech. The surgical procedure in a stepwise fashion involved:

1. Subcutaneous wide release of tight, accessory frenula, contracture bands and tethering of skin dimples in the perioral region to address the tendency to contracture of this condition.
2. The attenuated muscle in the central aspect of the lips (figure 2) was reconstructed by bringing together the orbicularis oris muscles in the midline to address the midline oral sphincter defect. At the end of the muscle reconstruction,

Figure 1 Cropped preoperative image of the patient depicting the characteristic 'whistling face'.

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Figure 2 Cropped intraoperative image illustrating the attenuated orbicularis oris muscle in the midline of the upper lip and the intraoral incisions used to bring the muscle together.

assessment of the oral sphincter was satisfactory, so additional, more invasive procedures to correct constriction, such as commissuroplasty, were not needed.

3. The mucosa of the buccal sulcus was elevated starting just beyond the sulcus reflection close to the gum on the inside of the lower lip to include advancement of the mucosa through multiple V-Y plasties (figure 3). More mucosal tissue was brought thereby into the internal lining of the lip in order to improve oral competence while preserving the oral opening and continuity of the vermilion border.

OUTCOME AND FOLLOW-UP

Postoperative recovery was uneventful. Following her 6-month clinic review (including further assessment by the speech and language therapists), she was discharged. Her parents reported a significant improvement in her speech, particularly with respect to bilabial consonants, which resulted in a significant improvement in intelligibility. At this stage, the patient herself reported normal oral continence, no tightness and a pleasing cosmetic outcome (figure 4).

DISCUSSION

FSS encompasses a wide spectrum of facial and extra-facial features. Stevenson and colleagues have outlined certain criteria which must be fulfilled in order for a diagnosis of FSS to be made. Two or more of the following manifestations must be present, in addition to certain facial features (small pinched mouth, H-shaped dimpling of the chin and prominent nasolabial folds) for FSS to be considered as a diagnosis: ulnar

Figure 3 Cropped intraoperative image modified with overdrawn lines illustrating where multiple V-Y plasties were performed to bring in more mucosal tissue into the free internal lining of the lower lip.

Figure 4 Cropped postoperative image illustrating the patient's ability to now fully close her lips.

deviation of the wrists and fingers, camptodactyly, hypoplastic and/or absent flexion creases and/or overriding fingers at birth, talipes equinovarus, calcaneovalgus deformities, vertical talus and/or metatarsus varus.¹

Owing to the midline attenuation of the orbicularis muscle complex, oral sphincter function is defective resulting in the characteristic 'whistling face abnormality' and posing both aesthetic and functional difficulties. These include psychosocial issues, poor oral continence—often leading to nutritional concerns—as well as speech difficulties. We used a muscle-sparing technique to reconstruct the oral sphincter defect and allow appropriate lip closure without the need for radical dissection or division of the orbicularis oris muscle. By avoiding external incisions, we were able to achieve not only a good functional outcome but also an excellent aesthetic result. We feel this is particularly important considering that psychosocial concerns, such as teasing, are often the reason for referral of FSS children to the plastic surgery service.

Preoperative and postoperative multidisciplinary team input is essential to ensure that the functional and aesthetic long-term outcomes from surgery are maintained including paediatric nutrition and oral health. In this patient, speech and language therapists played an important role in the highlighting and early diagnosis of specific speech defects and the surgical technique improved both the intelligibility of speech as well as the oral continence.

Learning points

- Prompt surgical correction of facial abnormalities in Freeman-Sheldon syndrome (FSS) is advisable to limit the potential damaging effects of sustained poor nutrition and to minimise the psychosocial impact of facial abnormalities both for the children themselves and their families.
- A muscle-sparing technique to reconstruct defective oral sphincter function allows appropriate lip closure without compromising the orbicularis oris muscle. Meticulous technique combined with incisions confined to the inside of the lips allows an excellent aesthetic as well as functional outcome to be achieved, particularly relevant when psychosocial aspects are the prime reason for presentation. Care must be taken to deal with any residual constriction of the oral sphincter if necessary, after muscle repair.
- Multidisciplinary team input is critical to the management of FSS children. In this case, speech and language therapy in particular was important in ensuring that speech defects were correctly diagnosed and highlighted early on as well as in helping guide the surgical technique employed to improve the intelligibility of speech.

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