Congenital Lower Lip Pits: A Familial Case of Van der Woude Syndrome

S. Karti, A. Chfiri, A. Jalal, A. El Harti, M. Diouri

ABSTRACT

Van der Woude syndrome or lip pits syndrome is a rare genetic autosomal dominant affection that represents the first cause of syndromic cleft lip and palate.

Lower lip pits associated or not with cleft lip or palate is characteristic in this syndrome.

The treatment of lip pits is surgical and can be very challenging since aesthetic good results can be hard to achieve.

We report a familial case of Van der Woude syndrome with lip pits as the only manifestation.

Keywords: Congenital, excision, lip pits, Van der Woude.

I. INTRODUCTION

Van der Woude syndrome is an inherited autosomal dominant syndrome [3] due to a mutation in chromosome band 1q32, more recently 1p34 [1], [4] in the gene encoding IRF6 (interferon regulator factor 6) [3], [5], [7], [9], lip pits on the vermilion of the lower lip is one of the manifestations of this syndrome, and can be isolated.

II. CASE REPORTS

A mother with her two boys a 7-year-old (Fig. 1) and 16 year-old (Fig. 2) came to our consultation, the two children presented depressions on both sides of lower lip present since birth.

Clinical examination showed bilateral symmetrical pits in the lower lip vermilion, with no saliva secretion. The physical examination didn’t find any abnormalities such as cleft lip or palate, or hypodontia. The mother also reported she has been operated for the same condition at the age of 17.

The lip pits were treated surgically by simple vertical wedge excision in both cases, and dissection of the pits. The results in both patient were aesthetically good.

Fig. 1. A 7-year-old boy with lower lip pits.
VWS is a rare syndrome affecting 1/60000 to 1/75000 births [1] autosomal dominant developmental malformation [1], [7], which associates multiple affections that can reflect variations in gene expression, like cleft lip or palate [6], [7], [8] or hypodontia [9], syngnata, high or low arched palate that are less frequent.

Extra oral affections such as congenital heart defects or limb anomalies are very rare [1].

Lip pits are the most distinctive trait of this syndrome [3], [10] seen in 88% of the cases, and can be isolated in many cases (64%) [5].

Severity of this affection can be variable even within the same family members [1], [10].

A good physical examination and history of patients with lips pits is very important, to detect associated anomalies and family history.

The diagnosis can be made if a first-degree relative (Parent, Child, sibling) of a diagnosed patient have either pits or cleft lip or palate.

Differential diagnosis can be made with popliteal Pterygium syndrome because of genetic proximity between the two syndromes [6]-[8] Pterygium syndrome can manifest with popliteal pterygia, genital abnormalities, and syndactyly associated to lower lip pits [8].

Lip pits usually manifest as bilateral paramedian symmetric depressions in the vermilion of the lower lip, it can also be unilateral in the center of the vermilion or only one pit left or right [1], [4], [5], they can be superficial or deep reaching the accessory salivary glands resulting in saliva draining.

Most commonly asymptomatic, lip pits can manifest with chronic inflammation, or infection in some cases [3].

Histologically we find muscular fibers surrounding salivary accessory glands [3].

Surgical treatment can be proposed for correcting the aesthetic deformity when it is causing emotional or social discomfort [1], [3], [9] or in case of recurrent inflammation or infection [10].

There is no precise protocol for lip pit excision, but different techniques have been described such as simple excision of the pits after catheterization which is the most commonly used [3], but the dissection of orbicularis oris in this technique can be very difficult due to the depth of the pits, and the results cannot be always satisfying what can end up in patients reoparation [3].

- Inverted T lip reduction [2], [4] can give good results.
- Routine horizontal monobloc excision with mucosal flaps reconstruction can be a good option for large pits and can help avoiding lower lip hypotonia [5].

The different techniques can be used depending on the severity and the depth of the lip pits.

IV. CONCLUSION

We presented a familial case of Van der Woude with bilateral lip pits as the only manifestation which are the most common feature found in this syndrome, and though this affection seems easy to treat surgically, it is usually very hard to achieve esthetically pleasing results.

REFERENCES

[1] Souissi A, Euch DE, Mokni M, Badri T, Dhahti AB. Congenital lower lip pits: A case report. Dermatology Online Journal. 2004; 10(2).
[2] Richardson S, Khandeparker RV. Management of lip pits in van der woude syndrome: a clinical classification with difficulty index. Journal of Oral and Maxillofacial Surgery. 2016; 74(9), 1849-e1.
[3] Abbo O, Vaysse F, Bieth E, Galinier P. Le syndrome de Van der Woude: une entité clinique méconnue. Annales de Chirurgie Plastique Esthétique. 2014; 59(1): 81-84. (French).
[4] Samargandi OA, Fakiha M. Isolated lower lip pits. *Natl J Maxillofac Surg*. 2014; 5: 247.

[5] Baghestani S, Sadeghi N, Yavarian M, Alghasi H. Lower lip pits in a patient with van der woude syndrome. *Journal of Craniofacial Surgery*. 2010; 21(5), 1380–1381.

[6] Bozkurt M, Kulaici Y, Zor F, Kapi E, Yucetas A. Reconstruction of the lower lip in Van der Woude syndrome. *Ann Plast Surg*. 2009; 62(4): 451–455.

[7] Vesna A. Van Der Woude syndrome. *J Dent Health Oral Disord Ther*. 2017; 7(2): 268-270.

[8] Etöz OA, Etöz A. Isolated Lower Lip Fistulas in Van der Woude Syndrome. *Journal of Craniofacial Surgery*. 2009; 20(5), 1612–1614.

[9] Krauel L, Parri FJ, Muñoz E, Sancho AM, Gean E, Morales L. Van der Woude Syndrome and Lower Lip Pits Treatment. *Journal of Oral and Maxillofacial Surgery*. 2008; 66(3), 589–592.

[10] Peralta-Mamani M, Terrero-Perez A, Dalben G, Rubira CMF, Honorio HM, Rubira-Bullen IF. Treatment of lower lip pits in Van der Woude syndrome: a systematic review. *International Journal of Oral and Maxillofacial Surgery*. 2018; 47(4): 421-427.