Surgical Treatment of Lip Pits In The Van Der Woude Syndrome

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Abstract

Van der Woude syndrome (VWS) is a congenital autosomal dominant disease caused by mutations in IRF6 gene. It is characterized by the presence of lower lip pits, cleft lip and/or cleft palate with hypodontia, bifid uvula, accessory salivary glands in labial pits and congenital heart defect commonly associated. Seventeen-year-old female patient, Van der Woude syndrome holder manifested by congenital lower lip pits, bifid uvula, complaining predominantly about aesthetic, asymptomatic and in good general health condition. Surgical correction of lip pits was done by semilunar incision in the lower lip. The incision and suture were done to enable better aesthetic results. In the postoperative follow-up there was good outcome and the patient was satisfied. Van der Woude syndrome is congenital but it may have its facial manifestations removed in order to provide social comfort to the patient. The patient must be oriented about the hereditary nature of the disease and the need of medical evaluation to detect the extent of the syndrome, particularly the cardiac abnormality risk, and undergo treatment if necessary.

Keywords: Van der woude syndrome; Lip; Syndrome cleft lip, Lower lip pit.

Introduction

Van der Woude Syndrome (VWS) is a congenital disease, inherited as an autosomal dominant disorder. The carrier has a 50% chance of transmitting the trait to any offspring and it is caused by mutations in the IRF6 gene. The diagnosis is clinical and it is based on typical signs such as lower lip pits, cleft palate, bifid uvula, among others [1,2].

The first report of labial pits was made by Demarquay in 1885 and it was reported again in 1900 by Epstein. However, only in 1954 Anne Van der Woude made an extensive study about the syndrome that eventually got his name [3-5].

VWS is characterized by the expression of lower lip pit, lip cleft and palate cleft that are present in 80% of patients (it is the only manifestation of the disease in 64% of cases), in addition it may also present hypodontia, hypoplasia, ankyloglossia, atresia of the palate, malocclusion, bifid uvula and accessory salivary

glands in the lip pits. As extra-oral manifestations, congenital heart defect, accessory nipple and Hirschsprung disease can be found. It has an incidence of 1 in 75,000 to 100,000 [1-4,6].

Therefore, the aim of this article is to describe the clinical findings of VWS through a report of a case in addition to reviewing the etiology, treatment of oral lesions as well as the management of the patient and family, taking into consideration the hereditary factor.

Case History

Seventeen-year-old female patient was evaluated with the complaint of pit-shaped injury located in semi lower lip mucosa, since birth, and there was no salivary secretion. During the intra oral examination, bifid uvula was noted, others mucous membranes showed normal aspects of coloration and texture and teeth were in excellent conditions of hygiene and conservation. It was asymptomatic and the patient complained predominantly about aesthetic. Good general health condition was noted and the medical history was unchanged (Figure 1A). After what was mentioned above, a presumptive diagnosis of VWS was made. During anamnesis a similar familiar history was not described, but the patient was referred to a clinical physician for cardiac evaluation, where a normal cardiac standard was found. After planning, surgical correction was carried out of the labial pits through navicular incision along the lower lip semi mucosa (Figure 1B), followed by excision. After controlling hemostasis, layered closure was performed (Figure 1C). Histopathological examination showed fragment of mucosa coated by parakeratinized stratified squamous epithelium. Lamina propria consists of dense connective tissue, exhibiting moderate mononuclear inflammatory infiltrate predominantly lymphocytic near the epithelium. In the sub mucosa region mature adipocytes were observed (Figure 2). Postoperative follow-up showed good cosmetic results and good patient satisfaction (Figure 1D). The patient was counseled regarding the hereditary characteristics of the syndrome and the need for monitoring and early approach in any cases of lips and palate clefts of the descendants.

Figure 1

Figure 2

Discussion

The VWS syndrome is the most common form of cleft lip occurring in 2% of all cases, and the lower lip pit is present in 80% of patients affected by the syndrome, being the only manifestation of the disease in 64% of cases [2,4,7]. However, it is a poorly documented disease with few cases described in the literature.

In a survey conducted by Lam et al. with 22 patients affected by VWS, 7 cases with cleft lip and bilateral cleft palate were found (32%), 7 patients with cleft lip and unilateral cleft palate (32%), 2 cases with isolated cleft palate (9%), 6 patients with submucosal cleft palate (27%) and no case of isolated cleft lip [7].

According to the literature, there is no consensus on the prevalence of VWS in terms of gender. Some studies report that both sexes are equally affected, including the original article of Woude A. (1954) that describes the pathology [4,5,7]. Other authors describe the prevalence in females and there are also some studies reporting the prevalence in males [6,8].

Deshmukh et al. (2014) describe the histopathology of VWS

as pits with a large depression in the central portion surrounded by elevated edges with stratified squamous epithelium in the margins and a thin central area, while most basal cells are vacuolated with displacement of the nucleus, which resemble immature epithelial cells [6].

According to Krause et al. (2008), the pits may have depth of 1 to 25mm reaching the orbicularis oris or communicate with minor salivary glands that drain aqueous fluid or saliva to the inside spontaneously or in response to stimulation, so the incomplete removal of fistula can result in cysts mucosa. In this case study, the patient had no salivary drainage [1,2,8].

VWS has varied expression can be manifested through various signs and symptoms mentioned above, but there are other syndromes that should be considered in the presumptive diagnosis: Popliteal Pterigia Syndrome (PPS), Hirschsprung's disease, digital or of acial syndrome type 1, ankyloblepharon threadlike adnatum [2,6].

Thus, as it is a syndrome caused by mutations in IRF6 gene manifested by changes in chromosome 1q32-q41 and 1p34 (VWS2), there is no cure, but there are treatment options for the signs and symptoms depending on the event, ranging from surgeries in cases of cleft lips and cleft palate through the closure of the clefts, bone grafts, implants, orthodontic and/or orthopedic treatment, dental prosthesis, cosmetic dentistry, in addition to have the cardiac abnormalities monitored and treated by a cardiologist [1-8].

Conclusion

VWS, as a congenital disease, has no cure, but it may have its maxillofacial manifestations attenuated by surgery in order to provide social comfort to the patient. It is also necessary a personal guidance regarding the hereditary characteristics of the syndrome and the need for cardiological evaluation and early approach in cases of cleft lips and cleft palate of the descendants. If the patient does not want the surgical removal of the labial pits, recommendations about the necessary precautions such as careful hygiene must be taken.

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