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Case Report

Van Der Woude Syndrome: Report of a Case

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Abstract

Van der Woude Syndrome (VWS) is a genetic disorder caused by a dominant mutant gene on an autosome which is associated with lower lip pits and clefts of the lip and/or cleft palate. Considerable heterogeneity regarding expression of the disorder constitutes a rare developmental malformation as congenital pits of the lower lip. Commonly associated characteristic features with lip pits are submucosal palatal clefts, velopharyngeal insufficiency and genitourinary or cardiovascular anomalies. The association of congenital pits with clefts is observed in about 50% of cases. They are usually located on either side of the midline of the vermilion portion of the lower lip, symmetrically and bilaterally. They may also present as unilaterally, medially or bilaterally asymmetrical structures. Surgical excision is the best-choice of treatment for lip pits. As the presence of salivary tissue in any residual tract increases the possibility of cyst formation, excision of lip pits is crucial in ensuring proper removal of the entire tract. In the present case scenario fistula repair was done using tongue flap and surgical excision of lip pits was performed using wedge incision. The purpose of the current case report is acquiring adequate knowledge about proper evaluation and treatment along with genetic counselling which will provide a proper guide for management of such cases.

Keywords: Lip Pits; Surgical Excision; Van der Woude Syndrome

Introduction

VWS is a rare genetic disorder, which is characterized by the association of congenital pits over lower lip with cleft lip and/or cleft palate. VWS is a developmental, congenital malformation with autosomal dominant inheritance. It is one of the commonly associated syndromes with orofacial clefts accounting for 1 - 2% individuals that have cleft lip and palate. The condition occurs in about 1:100,000 - 1:40,000 stillborn or live births. The syndrome is observed to be associated with submicroscopic deletion in chromosome 1q32-q41. Recently it has been proposed that mutation in the interferon regulatory factor-6 gene may also be responsible for VWS, but the exact mechanism is unclear [1].

In 1845, Demarquay first described about the lower lip pits as small pits on the vermilion border of the lips to large snout-like structures in the midline. These snout-like structures have been considered as accessory salivary glands, supernumerary labial cysts, labial fistulae, mucous glands or cysts, mucoceles, and dermoid cysts in earlier literature. In 1954, Anne Van der Woude first reported that there is a syndromic association of congenital pits of the lower lip with cleft lip and/or palate [2]. Thus, the term was coined as 'Van de Woude Syndrome' for such complex.

Depending on the location of lip pits, they may be divided into three types such as commissural, midline upper lip, and lower lip [3]. The present case exhibited the rarity of VWS presenting with bilateral paramedian congenital pits over lower lip. The rationale of the case was to address the surgical treatment with jeopardising the aesthetic and functional characteristics.

Case Report

A 17 year old female reported to the outpatient department of Oral and Maxillofacial Surgery of Sharad Pawar Dental College and Hospital with a chief complaint of defect in her upper jaw. The previous medical history of the patient revealed that she had undergone surgical correction of bilateral cleft lip at Government Hospital in Yavatmal at 2 years of age and surgical repair of wide bilateral cleft palate by two flap palatoplasty technique at Acharya Vinoba Bhave Rural Hospital, Sawangi at 7 years of age. The patient was the only child born of a nonconsanguineous marriage and had a negative family history of cleft lip/palate and lip pits. Patient was born after complete gestation of 9 months by vaginovertex delivery and all developmental milestones were achieved on time. The general examination of the patient was unremarkable.

Extraoral examination revealed scar over the upper lip suggestive of previous surgery and bilateral paramedian lower lip pits. The lower lip pits were asymptomatic and the patient did not report any discharge from the lip pits during meal time. Intraorally, surgical scar was observed over the hard and soft palate with presence of palatal fistulae over bilateral canine region. The dentition status included set of permanent teeth with missing maxillary central and lateral incisors. Outcomes of her cardiovascular and genitourinary evaluations were normal. After routine investigations the patient was subjected to surgery under general anaesthesia in which the fistula repair was carried out using anteriorly based tongue flap. In order to provide cosmetic change to patient's appearance, the lower lip pits were removed with the help of wedge incision. On histopathological examination, pits were observed to be lined with mucosal epithelium of the normal-appearing salivary gland lining. A focus of mild mucosal hyperplasia with mild inflammation and fibrosis was observed.

The patient's follow up continued for 6 months and there was no recurrence of lip pits or any associated cosmetic defects.

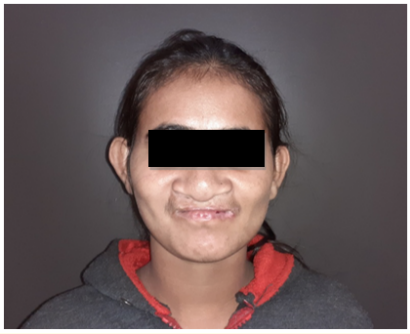

Figure 1: Preoperative frontal view.

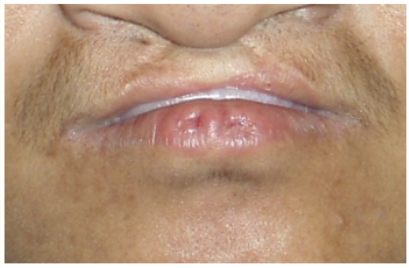

Figure 2: Paramedian lip pits over lower lip.

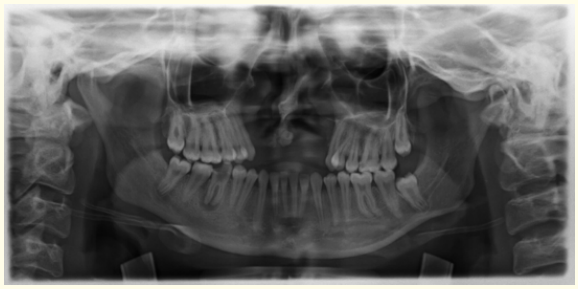

Figure 3: Radiograph suggestive of hypodontia.

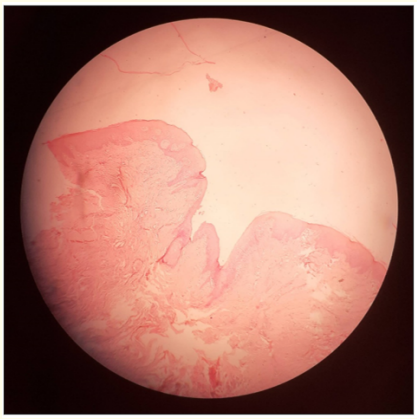

Figure 4: Histopathological picture of excised lip pits.

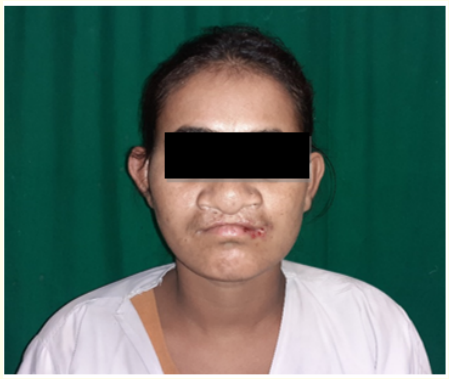

Figure 5: Postoperative frontal view.

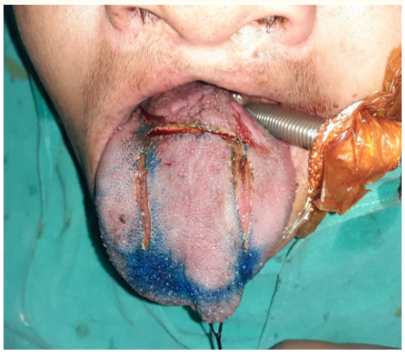

Figure 6: Tongue flap for fistula closure.

Discussion

Fusion of the mandibular arch and sulcus lateralis of lower lip takes place at around 5 weeks and that of frontonasal process with maxillary process at around 6 weeks of intrauterine life. It is postulated that a common event may disrupt fusion simultaneously at both the locations. This accounts for the strong association between lip pits and cleft lip and/or palate.

Out of all patients with VWS, around 10 to 81% are found with hypodontia. Other associated anomalies found in patients with lip pits include syndactyly of the hands, clubfoot, genitourinary abnormalities and cardiovascular anomalies [3].

Canals are formed by congenital pits and their length ranges from 1 to 25 mm. These canals generally extend into the orbicularis oris muscle. Accessory salivary glands commonly drain into these canals. The action of the orbicularis oris muscle helps the saliva to propel to the surface, which leads to mucous secretion from the pits [4].

While considering the surgical procedure, the simple excision technique may require more than two surgeries due to poor postoperative outcome. Hence, new surgical procedures have been advised for the removal of lower lip pits which include vertical wedge resection and Mutaf Goldstein technique. Vertical wedge resection technique reduces the lip volume, but preserves the white roll. The Mutaf-Goldstein technique shows split advancement by complete excision of middle part of lower lip. The inverted T - technique demonstrates entire removal of lip pits with deep incisions and joining of the vertical wedge borders in the midline [5].

The most common postoperative complication is the formation of mucocele and is often observed in individuals treated with simple excision technique. Pit recurrence may also be observed, and both require a second surgery [6].

Conclusion

VWS may be associated with certain underlying anomalies which may remain undiagnosed. A meticulous examination of lip pits may aid in the prompt diagnosis of such underlying anomalies. Surgical management of persisting lip pits improves the aesthetics along with functional characteristics. Even though excision of lip pit appears to be a simple procedure, it is difficult to obtain aesthetically appealing results in all cases. Thus, appropriate surgical technique and genetic counselling are of utmost importance.

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