Case Report

A Rare Case of Familal Van der Woude Syndrome

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Abstract

Van der Woude syndrome (VWS) is a rare autosomal-dominant craniofacial disorder characterized by the lower lip pits, cleft lip, and/or cleft palate. Other associated features of VWS include ankyloglossia, high-arched palate, limb anomalies, congenital heart defects, and bifid uvula. An early and proper diagnosis followed by a multidisciplinary approach is necessary to improve the esthetics as well as the self-esteem of affected individuals. This article presents a rare case report of familial VWS with lower lip pits and highlights its peculiar clinical presentation and management.

Keywords: Ankyloglossia, cleft palate, lip pits, Van der Woude syndrome

Introduction

Orofacial clefting (OFC), a common developmental genetic disorder, accounts for 1 in 2500 live births depending on geographic origin, racial, ethnic variation, and socioeconomic status. Van der Woude syndrome (VWS) is one such form of syndromic OFC constituting 2% of the cases.[1] VWS is inherited as an autosomal dominant disease caused by a mutation in a single gene encoding interferon regulatory factor 6.[2] The classical signs observed in VWS are congenital lower lip pits associated with the cleft lip, cleft palate, or both. The present case report highlights the features and esthetic management of a rare case of familial VWS.

Case Report

Two siblings aged 12 and 14 years reported with a chief complaint of unesthetic lip appearance. Past history revealed that both have undergone surgical correction for the cleft lip and palate. Family history revealed nonconsanguineous marriage and their elder brother had no such deformities. However, bilateral paramedian lower lip pits were noticed with their father [Figure 1a]. No abnormality detected on general physical examination, and they were mentally fit. On extraoral examination, gross deformity of the lower lip with a visible scar on the upper lip associated with flattened nose tip was noticed with the siblings. The elder sibling had a sinus like opening in the midline of the lower lip with little serous discharge [Figure 1b], whereas the younger had a dome-shaped elevation on the right half of lower lip with numerous mucosal folding and pits, exhibited a concave profile with the underdeveloped maxilla [Figure 1c].

Intraoral examination of elder sibling revealed an oronasal fistula, and abnormal architecture of soft palate (webbing) was observed with the absence of uvula [Figure 2a], whereas in younger sibling, a scar-like fold is present on the right side of the anterior hard palate [Figure 2b]. Both of them are in their mixed-dentition period associated with tongue-tie, lisping anterior segmental skeletal crossbite and posterior crossbite [Figure 2c-f].

Lateral cephalogram revealed maxillary hypoplasia leading to skeletal Class III in both siblings [Figure 3a and b]. Orthopantamograph revealed the congenital absence of 12, 22, 35, and 45 in elder sibling [Figure 4a] and congenital absence of 12, 35, and 45 in the other [Figure 4b]. Correlating the history and clinical findings, it was differentiated from popliteal pterygium syndrome (PPS) and orofacial–digital syndrome before coming to a final diagnosis of VWS.

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Received: 15-Apr-2019 **Revised:** 12-07-2019
Accepted: 04-Sep-2019 **Published:** 29-Nov-2019

Access this article online

Quick Response Code:

Website: www.ijpedor.org

DOI: 10.4103/ijpr.ijpr_7_19

How to cite this article: Vallabhaneni K, Muktineni S, Avula SS, Korikani A. A rare case of familal Van der Woude syndrome. Int J Pedod Rehabil 2019;4:76-9.

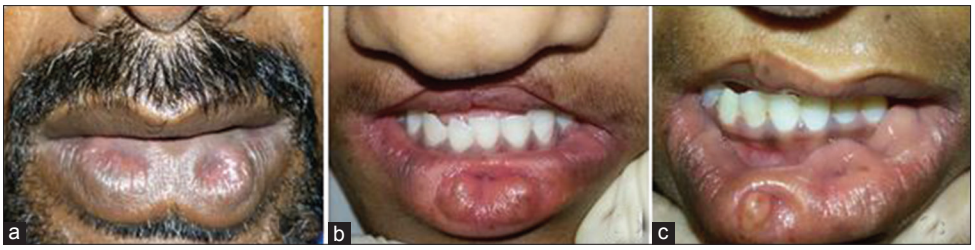

Figure 1: (a) Bilateral paramedian lower lip pits of the father. (b) Sinus like opening on the lower lip in elder sibling. (c) Dome-shaped elevation of the lower lip in the younger sibling.

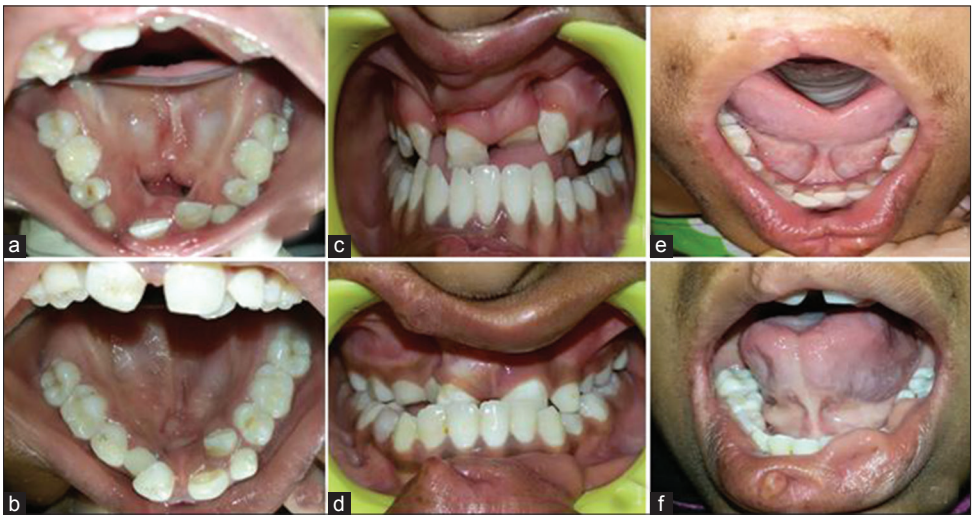

Figure 2: (a) Oronasal fistula and webbing of the soft palate in the elder sibling. (b) Scar-like fold on anterior hard palate in the younger sibling. (c and d) Anterior segmental and posterior crossbite in elder and younger sibling. (e and f) Tongue-tie in elder and younger sibling.

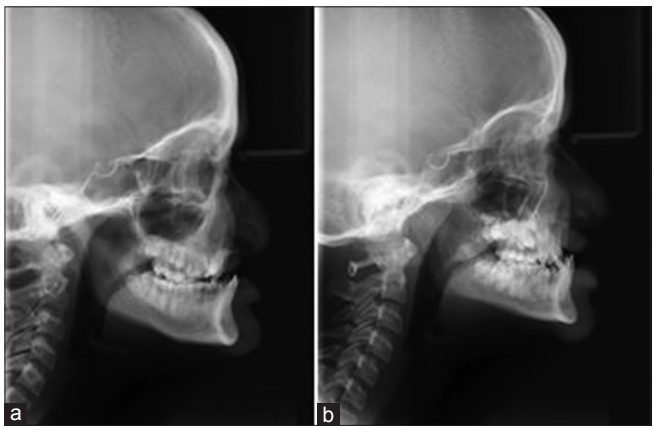

Figure 3: (a and b) Lateral cephalogram showing skeletal Class III in elder and younger sibling.

Considering the esthetic concern of the patients, surgical correction of the disfigured lower lips followed by the orthodontic intervention was planned. In addition, surgical intervention to close the oronasal communication (palatoplasty) for elder sibling and frenectomy for both siblings was considered. Inverted-T lip reduction technique in elder sibling and simple surgical excise of lower lip pits in younger sibling along with cheiloplasty under local anesthesia were followed. Postoperative healing was uneventful and 2 months follow-up showed acceptable esthetic and functional results regarding lip sensation, musculature, movement, and architecture in both the siblings [Figure 5a and b].

Discussion

VWS is a rare craniofacial syndrome, first reported in the literature by Demarquay in 1845. Later, Van der Woude in 1954, reviewed these features and established a relationship between lip pits and cleft lip or palate. During the embryonic life, the fusion of mandibular arch and lateral sulcus of the lower lip occurs at 5.5 weeks, whereas the fusion of the maxillary and nasofrontal processes occurs at 6 weeks. An incomplete reduction of the naturally occurring lateral sulcus, which normally occurs at the same time as the fusion of the lip and palate probably leads to the formation of lip pits. The entire lateral sulcus usually gets obliterated except the cephalic end, which deepens with growth. Simultaneously, edges of the furrow become more prominent and ultimately fuse together converting it into a tubular canal, opening at its upper end. The canal subsequently incorporates into deeper tissues of the lower lip, as this separates from the mandibular arch and remains as a congenital fistula.[3]

VWS can be seen in 2% of all cleft lip and palate cases with a prevalence rate of 1:35,000–1:100,000 live births. Lower lip pits, with or without cleft lip or palate, is a cardinal sign present

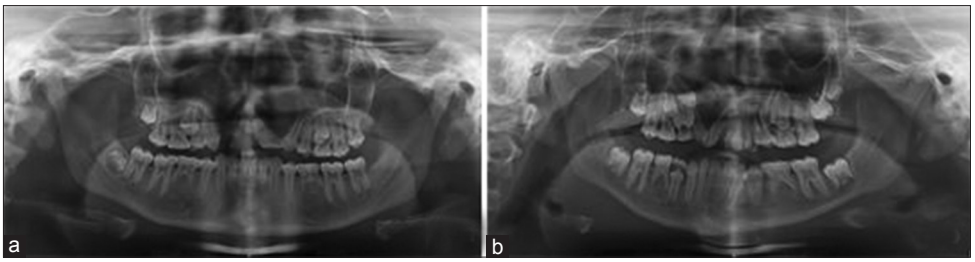

Figure 4: (a and b) Multiple congenital missing teeth in the elder and younger siblings.

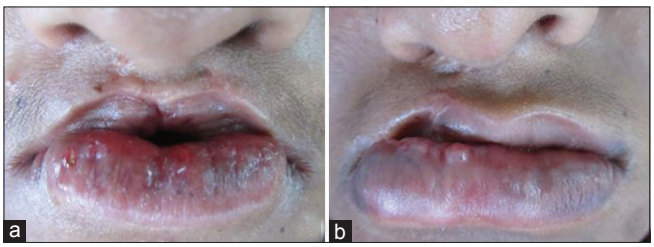

Figure 5: (a and b) Postoperative healing in elder and younger siblings.

in 90% of the cases with no significant gender difference regarding its prevalence.[4-6]

The classical type of lip pits is bilateral paramedian sinuses, symmetrically on either side of the midline of the lower lip. However, they can be unilaterally, medially, or bilaterally asymmetrical. A single median or paramedian lesion is considered as an incomplete expression of the trait. Lip pits are usually circular or oval but can also be transverse, slit-like, or sulci. The transverse mucosal ridges and the conical elevations (nipple-like) represent microforms of lower lip pits.[2] In the present case report, the male parent exhibited a classical type of lip pits, whereas the elder sibling exhibited a single median lip pit and the younger sibling exhibited multiple lip pits, with the most prominent one being a conical elevation on the right side (microform of lip pits).

The lip pits form canals are lined by labial mucosa, which extends inside the orbicularis oris muscle, with length ranging from 1 to 25 mm.[2] Here, the elder sibling exhibited a sinus tract of approximately 15 mm. Lower lip pits are usually asymptomatic, the only symptom might be the continuous or intermittent drainage of watery or salivary secretions[7,8] which was observed in this case.

Syndromes exhibiting lip pits other than VWS are PPS characterized by multiple defects of the extremities, face, mouth, and genitourinary system. Other malformations in PPS include webbing of the skin extending from the ischial tuberosities to the heels, cleft palate and/or lip, lower lip pit with salivary drainage, genital anomalies, and synechia or syngnathia. Orofacialdigital syndrome type 1 is an X-linked dominant trait, predominantly in males with striking orodental, facial, digital, renal, and central nervous system abnormalities. Orodental signs include cleft palate, bifid tongue, hypodontia, hypoplasia of nasal cartilages, hypertelorism, median cleft of the upper lip, and lip pits. Kabuki makeup syndrome manifestations include the dysmorphic face, postnatal growth retardation, skeletal abnormalities, mental retardation, and unusual dermatoglyphic patterns.[9] As the hallmark features of other syndromes were not manifested, it was confirmed as a classic case of familial VWS.

Management of VWS is mainly focused on the cosmetic correction of cleft and lip pits. Lip pits with mild symptoms may not require treatment unless the patient demands a cosmetic correction. However, persistent mucous secretions and painful recurrent inflammation require surgical intervention.[3] Ideally, the treatment for lip pits is complete excision of sinus tracts and optimal restoration of the orbicular ring for correction of lip hypotonicity, protrusion, and preserving its function. Owing to unsatisfactory results of the older modalities, alternative techniques such as vertical wedge resection, split-lip advancement technique and resection with alloderm graft implantation, and inverted-T lip reduction technique were introduced.[10] This technique showcases the advantages such as complete excision of sinus tracts, horizontal reduction of the lower lip, as well as restoration of the orbicular ring to correct hypotonia.[10] The inverted-T lip reduction technique was followed in elder sibling, whereas simple excision technique in a younger sibling for cosmetic correction [Figure 3a]. The cosmetic correction gave good results in elder sibling, whereas in younger sibling through the prominent snout-like projection of lower lip has reduced in size, and there was a sign of recurrence [Figure 3b]. The possible reason could be an incomplete excision of the sinus tract.

Conclusion

The identification of familial lip pits is crucial for genetic counseling and all parents with VWS should be cautioned that there is a 50% risk of having a child with a cleft lip and/or cleft palate.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

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