Van der Woude Syndrome: A Case Report

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Abstract

Congenital lip pits are developmental defects that occur on the paramedian portion of the vermilion border of the lower lip. They may be unilateral or bilateral and may occur as an isolated conditionor in association with cleft lip and or cleft palate. When the labial pits occur in association with cleftlip and/or palate the condition is referred to as Van der Woude Syndrome. In 1845, Demarquay(1945) first described the lower lip pit and postulated that the condition occurs due to the indentation of the central incisor. Later in 1954, Anne Van der Woude first reported the association of congenital pitsof the lower lip with cleft lip and palate. Congenital lower lip pits are present in 88% of all VWS patients, and in 65%–75% of the cases these are associated with cleft lip and palate. The Van der Woude syndrome is a rare condition the phenomenon of a cleft lip and cleft palate combined in the same pedigree makes it unique. The VDWS should be considered in the differential diagnosis of cleft lip and palate. The dentist may be the first person to diagnose this syndrome, and therefore,should be aware of its variable expressions. A proper treatment plan at the correct time will help to satisfy the psychological and esthetic needs of the patients. Further, genetic counseling is highly recommended.

Keywords: *Van der Woude Syndrome, Cleft lip, Cleft Palate, Labial Pits, Congenital anomalies of head and neck.*

Introduction

Congenital lip pits are developmental defects that occur on the paramedical portion of the vermilion border of the lower lip. They may be unilateral or bilateral and may occur as an isolated condition or in association with cleft lip and or cleft palate. When the labial pits occur in association with cleft lip and/or palate the condition is referred to as Van der Woude Syndrome. In 1845, Demarquay (1945) first described the lower lip pit and postulated that the condition occurs due to the indentation of the central incisor. Many parents still subscribe to this hypothesis even though the condition is present since birth months before the eruption of maxillary central incisors. Later in 1954, Anne Van der Woude first reported the association of congenital pits of the lower lip with cleft lip and palate. Congenital lower lip pits are present in 88% of all VWS patients, and in 65%–75% of the cases these are associated with cleft lip and palate.[1]

The distinctive feature of VWS is the presence of lower lip pits and/or sinuses, which are present in approximately 85% of cases, but it may be absent in 15% of the cases. In some rare cases, a single barely visible pit might be the only distinguishable feature of VWS, and in others, it may be absent. Other anomalies that are frequently associated with VWS include hypodontia, submucous cleft palate, and bifid uvula, and enamel hypoplasia. Manifestations of the syndrome in other than the oral or facial areas are unusual. More extreme phenotypes in parents tend to produce more extreme expression in their children.[6]

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Case Report: A seven-year-old boy reported with a complaint of difficulty in oral intake of food and liquids due to a cleft in the upper jaw. There was a history of multiple episodes of respiratory tract infections, breathlessness, nasal regurgitation of food, and a nasal twang in the voice. His history revealed that he was delivered through a Cesarean section. The maternal history revealed no history of medication or illness during pregnancy. There was no relevant family history.

An extraoral examination of the child revealed surgical scars of a corrected cleft lip, with two small pits in the center of the lower lip. They just manifested as two simple depressions (Figure 1). When the lower lip was compressed, thick mucus secretion was expressed from both the lip pits. Intraoral examination revealed bilateral clefts of the premaxilla and a median cleft of the hard palate. The uvula was completely absent with a submucous cleft of the soft palate (Figure 2). Radiographic examination revealed a diffuse radiolucent defect in the anterior maxilla, suggestive of a cleft, crowding of the maxillary anterior teeth, and a missing maxillary left incisor. All other teeth were in normal developmental stages and there was no abnormality in the skeletal growth of the jaws.

All the teeth with caries were treated appropriately in the child. The child had already undergone surgical correction of the cleft lip and was referred to higher centers for surgical correction of the cleft palate (Figure 3). Meanwhile, the child was instructed to maintain good oral hygiene. The lip pits were also excised and Z-plasty was done to maintain esthetics (Figure 4).

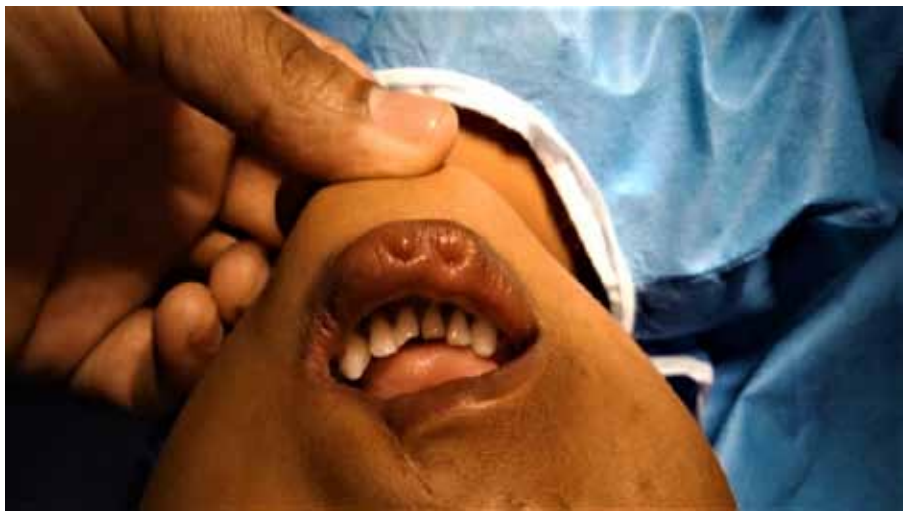

Figure 1. Lip Pits

Figure 2. Cleft Palate

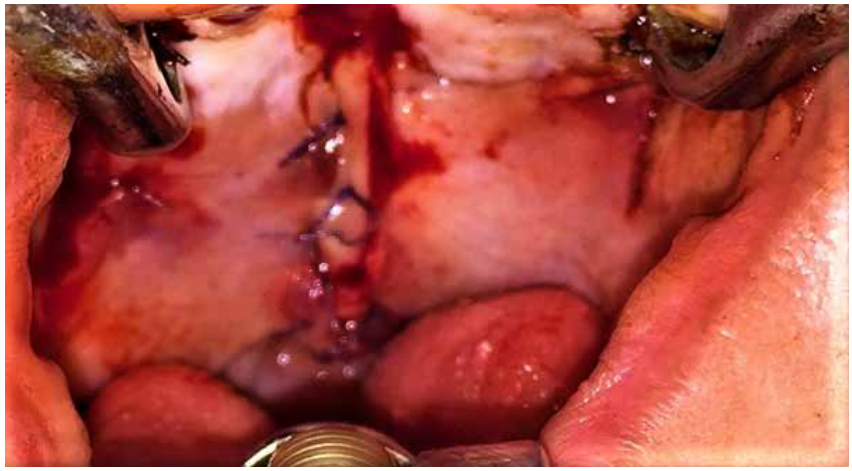

Figure 3. Repaired cleft palate

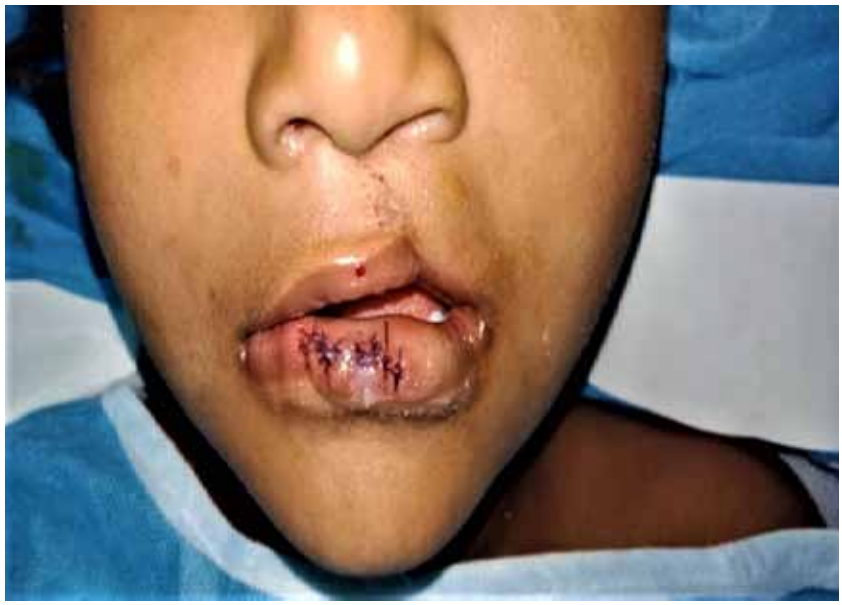

Figure 4. Postoperative picture

Discussion

Lip pits are rare congenital malformations seen on the lower lip, upper lip, or the oral commissure. The condition generally presents as a unilateral or bilateral depression and has a predilection for the lower lip. Lower lip pits are also known as fistula labii inferioris, labial humps, labial cysts, labial fistulae, and paramedian sinuses of the lower lip. These pits are usually circular or oval and generally show bilateral distribution, located on either side of the midline. The unilateral pits are observed mostly on the left side and rarely on the right side. The exact etiology of lip pits is not known. It may result from notching the lip at an early stage of development with fixation of tissues at the base of the notch or it may be due to a failure of complete union of embryonic lateral sulci of the lip. During prenatal development, a fusion of the mandibular arch and sulcus lateralis occurs at 5.5 weeks while the fusion of the maxillary and frontonasal processes takes place at 6 weeks. Probably, a common event may simultaneously disturb fusion in both locations resulting in the strong association between lip pits and cleft lip and/or palate. Although lip pits are generally asymptomatic, in some cases they contain salivary glands. Watery discharge from these pits, especially during mealtime has been reported. Mild cases of lip pits require no treatment. In this report, none of the patients presented salivary discharge during mealtime. However, surgical correction of these anomalies is recommended in more severe cases not only to improve esthetics but

also to remove chronic inflammation and prevention of salivary discharge from lip pits.[1]

The first report on lower lip pits by Demarquay attributed the formation of lower lip pits to the impressions made on the lower lip by the upper central incisors. Most patients tend to adopt this hypothesis even now. According to Kitamura, in a 32-day embryo, the lower lip consists of four growth centers, divided by one median and two lateral grooves. In the 38-day embryo, the lateral grooves disappear, except in the case of impeded mandibular process growth that results in the formation of a lip pit. If a cyst deriving from the epithelial wall communicated with the duct of labial glands, a congenital fistula of the lip is formed. The development of lip pits starts at day 36 of development, CL at day 40, and CP on day 50. The periods of liability of these three tissues probably vary in length and even in sequence, and perhaps they also overlap accounting for the strong association between the lip pits and cleft lip or palate.[2]

The majority of the labial and commissural pits occur without mucus exudation. However, a small percentage may be associated with cleft lip and palate, and therefore not a thorough clinical examination is recommended in individuals with pits to rule out a submucosal cleft palate which may not be obvious to the clinician. Surgical excision of the labial and commissural pits is indicated if the aesthetics of the individual is appreciably affected and exudation of mucous secretions can not be controlled. Surgical excision should include the total removal of the minor salivary glands that exude secretions at the base of the pits to prevent the formation of mucoceles or cysts.[3]

Management of VDWS is mainly focused on the surgical correction of clefts and lip pits. Surgical excision of the labial and commissural pits is indicated if the esthetics of the individual is appreciably affected and exudation of the mucous secretions cannot be controlled. Surgical excision should include total removal of the minor salivary glands that exude secretions at the base of the pits, to prevent the formation of a mucocele or cyst.[4]

Proper evaluation and treatment of VWS along with genetic counseling is important. Care should be taken during the surgical procedure for lip pits, of complete excision of the fistulous or sinus tract, which otherwise may lead to the formation of a mucoid cyst. Patients who do not undergo surgical correction should be instructed about meticulous hygiene care.[5]

More extreme phenotypes in parents tend to produce more extreme expression in their children. However, the lesser expressions of VWS are common and should be actively looked for when counseling families about cleft lip or cleft lip and palate. All the affected parents should be cautioned that they carry a risk of 50% for each child with a cleft lip or palate or both. The potential of embryocopy to detect minor malformations such as cleft lip in early-term pregnancy allows for detection of VWS.[6]

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. As the hallmark features of other syndromes were not manifested, it was confirmed as a classic case of familial VWS.[7]

Many mutations have been described in VWS patients, and also due to the implication of stochastic factors or modifier genes on the IRF6 function, molecular genetic testing does not seem justified at the moment. Because VWS has a high penetrance and a variable expression, genetic counseling is advised for future pregnancy.[8]

Conclusion

The Van der Woude syndrome is a rare condition the phenomenon of a cleft lip and cleft palate combined in the same pedigree makes it unique. The VDWS should be considered in the differential diagnosis of cleft lip and palate. The dentist may be the first person to diagnose this syndrome, and therefore, should be aware of its variable expressions. A proper treatment plan at the correct time will help to satisfy the psychological and esthetic needs of the patients. Further, genetic counseling is highly recommended.

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