Van der Woude Syndrome: Case Reports and a Review


1Professor and Head, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
2Senior Lecturer, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
3Professor and Head, Department of Oral and Maxillofacial Surgery, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
4Professor and Head, Department of Oral Pathology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
5Postgraduate Student, Department of Oral and Maxillofacial Surgery, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India

Correspondence: Vinay Kumar Reddy K, Professor and Head, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Staff Quarters, Penna G-4, Mamata Hospital Campus, Giriprasad Nagar, Khammam-507002 Andhra Pradesh, India, Phone: 9849052434, e-mail: kundoorvinayreddy@yahoo.in

ABSTRACT

Van der Woude syndrome is a rare autosomal dominant disorder characterized by a cleft lip and cleft palate with congenital lip pits. The variable manifestations include lip pits, absent teeth and isolated cleft lip and cleft palate of varying degrees of severity and other associated anomalies though rare have also been reported. It occurs in equal distribution between both genders. We report two cases of Van der Woude syndrome.

Keywords: Lip pits, Autosomal dominant, Cleft lip and palate, Van der Woude syndrome.

INTRODUCTION

Among many disturbances of growth and development that involve the oral and perioral structures, cleft lip and palate are perhaps the commonest. Congenital lip pits are developmental defects that occur on the paramedian portion of the vermilion border of the lower lip. When lip pits occur in association with cleft lip and palate then the situation is termed as Van der Woude syndrome (VWS). Van der Woude syndrome is also called cleft lip syndrome, lip pit syndrome or dimpled papilla of lip. The first case of lip pits was reported by Demarquay in 1845, and the association of cleft lip and palate with lip pits was given by Van der Woude in 1954. It is a rare autosomal dominantly inherited disorder constituting about 2% of all syndromic cases of cleft lip and palate with an incidence of one in 1,00,000 to 2,00,000 and is distributed equally among both genders. It is inherited by an autosomal dominant pattern and shows high expressivity and penetrance. Most cases have been linked to chromosome 1q32-q41 but a second locus has been mapped to 1p34. Affected individuals have a 50% chance of transmitting the syndrome to next generation. The clinical manifestations include cleft lip and palate, paramedian lip pits, hypodontia, missing premolars, bifid uvula, ankyloglossia, hypernasal voice and systemic manifestations, like syndactyly, synthathia, accessory nipples, anomalies of limb and cardiac anomalies, though rare, are reported. We report two cases of Van der Woude syndrome along with their clinical presentation and management.

CASE REPORTS

Case 1

A 20-year-old male patient reported to the Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, with a chief complaint of difficulty in speech and discharge from lower lips since birth. Extraoral examination revealed paramedian lip pits on the lower lip measuring about 1 × 1 cm in diameter present since birth which gradually increased to attain the present size associated with retarded and stuttering speech. Lip pits were associated with salivary discharge, which were nontender on palpation (Fig. 1). Intraoral examination of soft tissue revealed presence of cleft

Fig. 1: Extraoral photograph showing paramedian lip pits of lower lip (Case 1)
in the soft palate with bifid uvula (Fig. 2). Hard tissue examination revealed retained 75 with clinically missing 35, 18, 28, 38 and 48 (Fig. 2). Radiographic examination with panoramic radiograph confirmed missing 35, 18, 38 and 48 with unerupted 28 (Fig. 3).

**Case 2**

A 13-year-old male patient reported to the Department of Oral Medicine and Radiology, Mamata Dental College, with a chief complaint of difficulty in speech. Extraoral examination revealed presence of paramedian lip pits on the lower lip measuring about 1 × 1 cm in diameter, present since birth which gradually increased in size to attain the present size associated with difficulty in speech with hypernasal voice. Lip pits were nontender and no associated discharge (Fig. 4). Intraoral examination of soft tissue revealed bifid uvula (Fig. 5) and ankyloglossia (Fig. 6). Hard tissue examination revealed presence of all the teeth for his age (Fig 5).

**MANAGEMENT**

Complete excision of lip pits, palatoplasty and uvuloplasty were done under general anesthesia in case one and, the second patient is still awaiting for surgery.

**DISCUSSION**

Paramedian lip pits are the most common manifestation of the syndrome occurring in 87.5% of all cases⁴ and they are the only manifestation of the syndrome in case of 66%.³ They are formed as a result of failure of complete fusion of embryonic lateral sulci of the lip which persist and ultimately develop into
pits. The surface of the lip pits is like a transverse or circular slit located at the apex of the nipple-like elevations and measure up to 3 mm in diameter and depth ranges from 1 to 15 mm.\textsuperscript{2,7} Orifices of minor salivary ducts open into these pits causing visible salivary discharge which causes great discomfort and embarrassment to the patient.\textsuperscript{8} In some cases, there can be a submucosal cleft which appears clinically normal but the effected individuals have 50% chance of transmitting the complete syndrome to the next generation.\textsuperscript{2,9} Van der Woude syndrome is usually diagnosed based on its features. A genetic test is available that identifies a mutation in the IRF6 gene, which is found in 70% of people with Van der Woude syndrome.\textsuperscript{4} It should be differentiated from popliteal pterygium syndrome in which anomalies of skin and genital anomalies are found.\textsuperscript{10}

**CONCLUSION**

Van der Woude syndrome is an autosomal dominant syndrome which may present with a variety of clinical features. Management of VWS is primarily to alleviate the discomfort caused by the lip pits due to salivary discharge, to improve speech and the esthetic appearance of the lip by complete surgical excision. Management should be a multidisciplinary approach to enable the patient a better functional efficiency.

**REFERENCES**