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Case Report

VAN DER WOUDE SYNDROME: A UNIQUE CASE REPORT WITH SHORT REVIEW OF LITERATURE

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ABSTRACT

Van der Woude Syndrome is one of the syndromic orofacial clefting accounting for 2 % of all cases. It is a rare autosomal dominant condition with high penetrance and variable expression. Clinical manifestation of this autosomal dominant clefting syndrome includes bilateral midline lower lip pits, cleft lip and/or palate and hypodontia. These congenital lip pits appear as a malformation in the vermilion border of the lip with or without excretion. Hyper nasal voice and cleft or bifid uvula may be present. Although infrequently reported, other symptoms include syngnathia, narrow high arched palate, and ankyloglossia. most cases of van der woude syndrome have been linked with to a deletion in chromosome 1q32-q41: however, a second chromosomal locus at 1q34 has also been identified. Most upper and/or lower second premolars are frequently absent. This case report describes a 17 year old male patient who had reported to the outpatient department of the department of Oral Medicine and Radiology, Maitri College Of Dentistry and Research Center, Anjora, Durg, Chhattishgarh, and eventually diagnosed with this syndrome along with its clinical manifestation, genetic predisposition, differential diagnosis, investigative procedures and management.

Key words: -Cleft Palate, Lip Pits, Van der Woude, Ankyloglossia, Syngnathia.

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INTRODUCTION

Orofacial clefting is a common developmental genetic disorder that occurs with a prevalence which has been estimated at between 1 in 2500 live births depending on geographic origin, racial and ethnic variation and socio – economic status (1). Van der Woude syndrome (VWS) is the most common form of syndromic clefting accountic for 2 % of all cases (2). Lower lip pit(S) and cleft lip and palate are the cardinal features of this syndrome, having a prevalence rates varying from 1:100000 to 1:400000 still born or live births with equal sex predilection (3)

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CASE REPORT

A 17-year-old boy reported to the Department of Oral Medicine and Radiology with a chief complaint of crowding in his upper front teeth region. His past medical history was uneventful. There was no consanguinity in the parents, and antenatal history of the mother was negative for any significant illness and drug intake during pregnancy. History revealed that he was the first child born to his parents, and none of the family members had cleft lip/palate. Extra oral examination revealed mid face retrusion and bilateral cleft of upper lip. His lower lip had bilateral lip pits (paramedian) of 3 mm in diameter and 4 mm deep. When the lower lip was compressed, mucous secretion was expressed from the lip pits. Intra oral examination revealed collapsed V shaped palate with cleft and oronasal communication. There was severe upper anterior teeth crowding . There was obliteration of the upper labial vestibule due to the adhesion of the lip to the gingiva by a thick fibrous band which blanched on retraction of the upper lip. The mandibular teeth were all present and aligned well along with a short lingual frenum causing ankyloglossia. There was hypoplasia of teeth, shrunken uvula in addition to the cardinal features of the syndrome (lip pits, cleft palate & cleft lip). General medical examination was done to rule out any systemic problems. Radiographic examinations like Intra oral periapical radiographs, maxillary occlusal view, orthopantomography, lateral cephalogram, & Cone beam computed tomography of full face was done. Thus, a clinical diagnosis of VWS was made. Multidisciplinary approach for correction of clefts and complete rehabilitation was planned. Patient was referred to the department of oral and maxillofacial surgery for surgical intervention and correction of clefts and the lip pits. Orthodontic treatment for the crowding of teeth was also advised after the surgery. Importance of genetic counselling which is highly recommended as it provides information on likelihood of gene transmission and possible ways of expression was explained and advised to the patient.

FIG 1: EXTRAORAL PICTURE SHOWING UNSYMMETRY

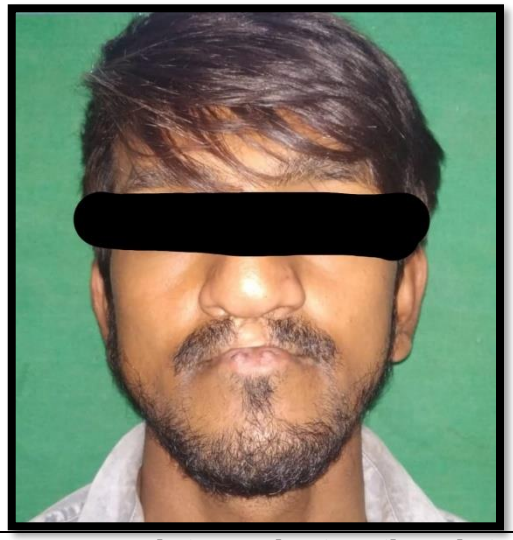


FIG 2: Extraoral Side-View Picture Showing Midface Retrusion And Intruded Upper Lip

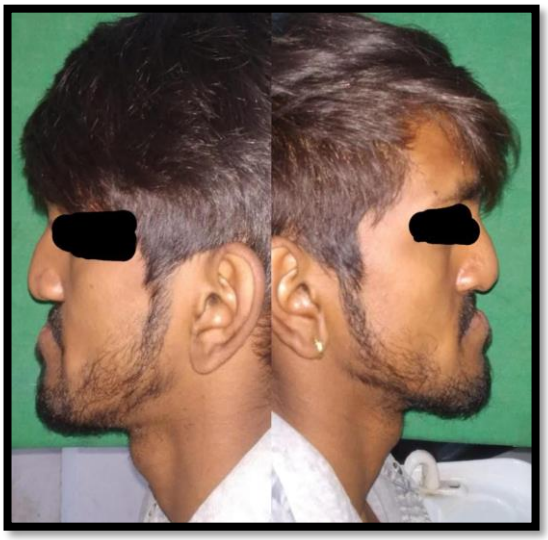


FIG 3: Extraoral Picture Showing Bilateral Lip Pits

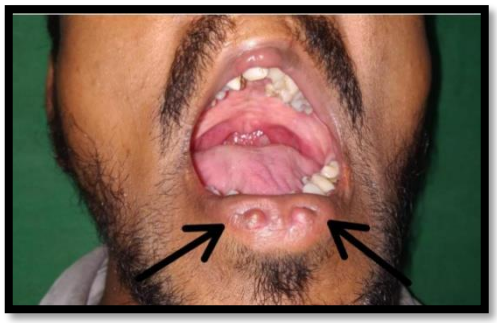


FIG 4: Intraoral Picture Showing Collapsed Maxillary Arch & Severe Malalignment Of

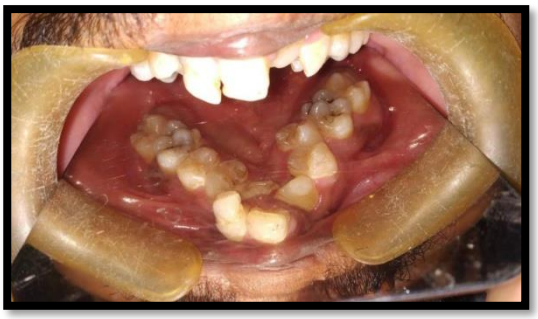
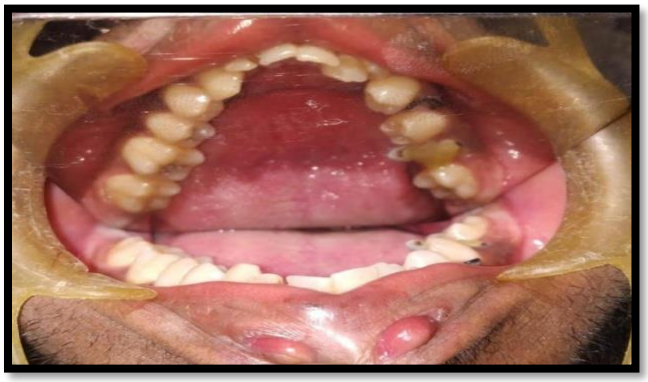


FIG 5: INTRAORAL PICTURE SHOWING IMBRICATION.



DISCUSSION

One of the most common developmental defects seen in India is cleft lip and palate. Amongst them a few cases are associated with congenital lip pits termed as VWS (4). Burdick and Bixler had presented 140 years (1845 to 1985) analysis of medical records of patients (5).

EPIDEMIOLOGY

Prevalence; the incidence of VW syndrome is about 1 in 40,000 to 2,00,000 people. Gender predilection; many authors believe that there is high prevalence of the syndrome in females, though it is noted in male patient in present paper.

CARDINAL FEATURES

Lower their lip pits are the principal trait of VWS and are discussed in relation to their location, morphology, etiology and histopathology.

LOCATION:

The typical presentation of lower lip pits is the bilateral paramedian sinuses of the lower lip placed symmetrically on either side of the midline. The lip pits are usually circular or oval but can also be transverse slit like or sulci. The transverse mucosal ridges, the conical elevations (nipple like) and/or openings with no depth represent microforms of lower lip pits (6).

MORPHOLOGY:

The lip pits form canals and are lined by labial mucosa which extends into the orbicularis oris muscle. They can present as two nipples like protrusions with no sinus openings at their apices as presented in the case reported by the authors here. The canals always end as blind sacs surrounded by mucous glands. A single median or paramedian lip pit is considered as an incomplete expression of trait whereas bilateral lip pits are features of complete expression (7)

EPIGENITICS:

Van der Woude syndrome has an autosomal dominant hereditary pattern with a variable expressivity and its penetrance has been estimated at 80-100%. Eighty percent of gene carriers are not diagnosed because they are nonpenetrant, among the penetrant gene carriers. The VWS loci were initially mapped to human chromosome 1q32-q41 and phenotypes were subsequently demonstrated to result from mutations in the gene encoding interferon regulatory factor 6(IRF6) (2). IRF6 belongs to a family of transcription factors that share a highly conserved N-terminal, penta-tryptophan, helix-turn-helix DNA binding domain and a less well conserved protein-binding domain (2). Affected males and females are equally likely to transmit VWS (6). The potential of embryoscopy to detect minor malformations such as a cleft lip allows for early termination of pregnancy in patients with VWS (8).

DIFFERENTIAL DIAGNOSIS

There are many other syndromes which are considered as allelic variants of the syndrome and present with congenital lip pits like orofacial digital syndrome and popliteal syndrome (4,9).

1. First is the **Popliteal pterygium syndrome (PPS)** that includes popliteal web, cleft lip/palate, lower lip pits in 60 % cases, and cryptorchidism and bifid scrotum in males and hypoplastic labia majora and uterus in females. People with VWS have a risk of giving birth to offspring with PPS(10).
2. Second is the **Hirschsprung's disease** {aganglionic megacolon with cleft lip and lip pits} (11).
3. Third is **Orofacial digital syndrome type 1**, with striking orodental, facial, digital, renal, and central nervous system abnormalities. Orodental signs include cleft palate, bifid tongue, hypodontia and median cleft od upper lip (12).
4. Finally, the following are, moreover, considered: **ankyloblepharon filiforme adnatum** – partial or complete full thickness fusion of the lid margins – cleft lip, palate, hydrocephalus, meningocele, imperforate anus, bilateral syndactyly, anfantile

glaucoma, cardiac problems such as patent ductus arteriosus and VSD's (13).

CONCLUSION

Van der woude syndrome is not frequently reported. Making a distinction between similar diseases presenting cases requires thorough knowledge of various clinical features and investigatory procedures. A multidisciplinary approach of a team compromising of oral medicine & radiology specialist, maxillofacial surgeon, orthodontist, plastic surgeon, ENT surgeon, speech therapist is required. Genetic counseling which would give information on likelyhood of gene transmission, possible ways of expression and a possible embryoscopy to detect minor malformations such as cleft should be encouraged in early diagnosis and management (14).

CONFLICT OF INTERESTS

The authors declare that there is no conflict of interests regarding the publication of this paper.

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