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Oral Medicine

"VAN DER WOUDE SYNDROME: REPORT OF TWO CASES"

Dr. Sivan Sathish	MDS, MFDS, RCPS (Glasgow) (Professor and HOD); Department of Oral Medicine and Radiology.
Dr. S. Manonmani*	(Post Graduate), Department of Oral Medicine and Radiology. *Corresponding Author
Dr. R. Christeffi Mabel	MDS(Professor), Department of Oral Medicine and Radiology.
Dr. A. Priyadharshini	MDS (Senior lecturer), Department of Oral Medicine and Radiology.

ABSTRACT Van Der Woude syndrome is a rare autosomal dominant condition which is associated with developmental malformations involving lips, palate. This syndrome can be a genetic origin due to Microdeletion of chromosome bands 1q32-q41 and also mutation of Interferon regulatory factor 6 (IRF-6) can be a pathogenic cause. We report two interesting cases of Van Der Woude syndrome with lip pits and orofacial features.

KEYWORDS : Cleft lip, Cleft palate, Lip pits, Van Der Woude syndrome

INTRODUCTION:

Van Der Woude syndrome (VWS) is a rare autosomal dominant syndrome causing craniofacial and orofacial disorder with a cleft lip and / or a cleft palate and lip pits(1). The congenital lip pits can be appreciated on bilateral paramedian region on the lower lip or commissure of the lips. This disorder affects both male female gender equally and incidence rate at birth is 1/1,00,00-2,00,00(2).

The first description comes from the year 1845 by Jean Nicolas Demarquay , who first described lower lip pit and postulated that the condition occurs due to the indentation of central incisor. Many parents still subscribe 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 (fistula labii inferioris congenita) with cleft lip and palate.

This syndrome can be a genetic origin due to Microdeletion of chromosome bands 1q32-q41 and second locus has been mapped to 1p34. Also, mutation of Interferon regulatory factor 6 (IRF-6) can be a pathogenic cause. The autosomal dominant gene for VWS has been found to have 80% to 92% penetrance, indicating 3% to 20% of VWS genotypes were missed because they were clinically mostly asymptomatic.

A wide variety of expressions can be observed within the pedigree of the syndrome. Besides the cardinal symptoms of the VWS with cleft lip, cleft palate, paramedian lip pits and lip pits on commissure, they can be accompanied by hypodontia, hypoplasia, congenital hand deformities, congenital heart disease, ankyloglossia etc.

We present the rare cases of Van Der Woude syndrome with variable expressivity in a south Indian patient.

CASE REPORT 1:

A 22 years old male patient reported to the department of Oral Medicine and radiology with chief complaint of decay in right lower back tooth for past 3 years. On extra-oral examination asymmetrical lip pits, two in number which is measuring 1×1 cm on each side of the para-median region of the lower lip were noted. On inspection, the lip pits were shallow in depth with regular margin. The mucosa overlying the pits were normal in colour and surface texture. There were no signs of exudate from the pits. On palpation, lip pits are non-tender and the consistency of the tissue is soft and supple. Vermillion border of lower lip appears to be tissue redundant and cupid bow in shape. A noticeable groove present on the mento-labial sulcus. From personal history patient reports these features were present from birth. On intra-oral examination, revealed highly arched and constricted palate. Facial features show flattened nose bridge with ophthalmic hypertelorism (Figure: 1a &1b). General physical examination revealed no considerable abnormalities. Patient is a full-term delivery baby of non-consanguineous married parents and no family history of oral and facial anomalies.

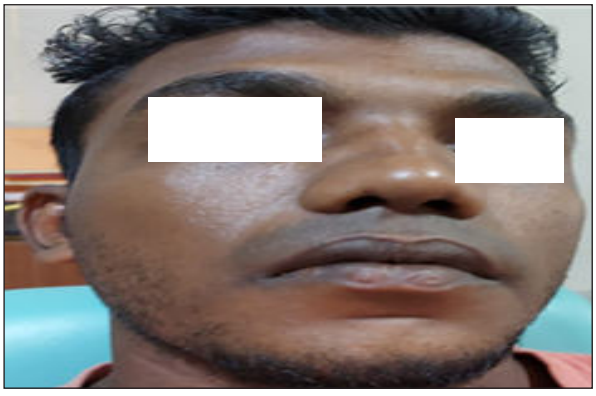

Figure 1A

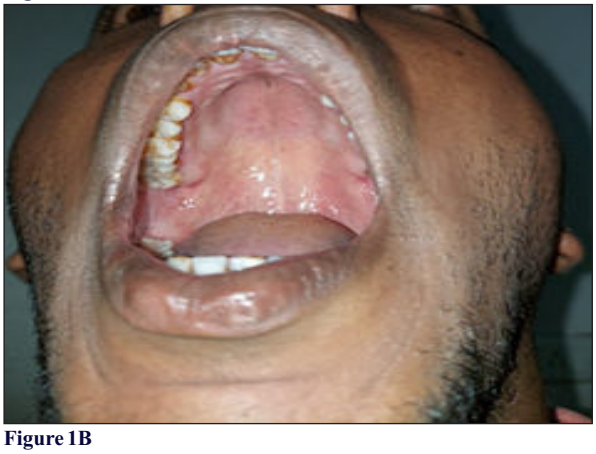

Figure 1B

Figure:1 a. Extra oral examination: para-median lip pits & flattened nose bridge with ophthalmic hypertelorism **& 1b.** Intra oral examination: highly arched & constricted palate

Case Report 2:

A 60 years old male patient reported to the department of Oral Medicine and radiology with chief complaint of mobility in upper and

lower back tooth region for past one month and pain associated with left upper back tooth region. On extra-oral examination, the lip pits, two in number, one on the junction of vermillion border and labial mucosa of left commissure of lip, which is deep, measuring 0.5×0.5 cm in size and one on the right commissure of the lip, which is shallow depressed measuring 0.5×0.5 cm in size. On palpation, lip pits are non-tender and the consistency of the tissue is soft and supple. On intra-oral examination, revealed highly arched and constricted palate. A small furrow on the mid palatine region, measuring 0.5×0.2 cm in size and 0.3 cm in depth was noted and which is bordered anteriorly by hard palate and posteriorly by soft palate (Figure: 2). From personal history patient reports these features were present from birth. General physical examination revealed no considerable abnormalities. Patient is a full-term delivery baby of non-consanguineous married parents and no family history of oral and facial anomalies.

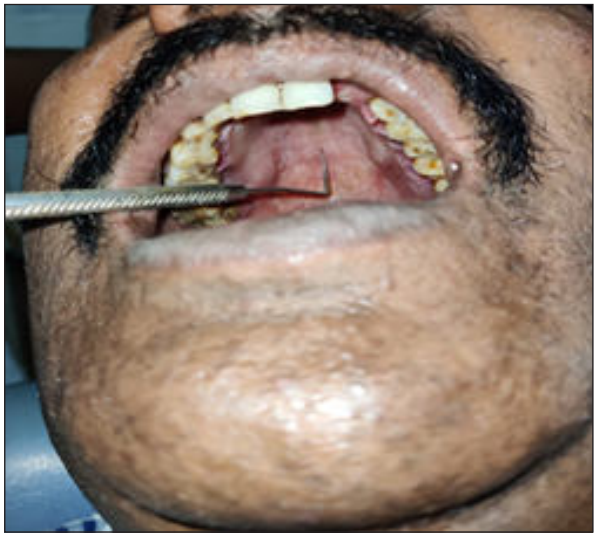

Figure2: Extra oral examination: two lip pits - on junction of vermillion border and labial mucosa of left commissure of lip on the right commissure of the lip & Intra oral examination: reveals highly arched and constricted palate with small furrow on mid palatine region

DISCUSSION:

Van Der Woude Syndrome(VDW) is a genetic disorder categorized by the combination of cleft lip with or without cleft palate , cleft palate ,lower lip pits, commissural lip pits. Globally, prevalence of orofacial clefts ranges from 1:1000 to 1:500 births and in which more than 400 syndromes comprises of cleft lip/cleft palate. VWS is evident from other orofacial cleft syndromes due to the combination of cleft lip and palate , cleft palate only and lip pits within the same family(3).

Lip pits were first reported by Demarquay in 1845 and he thought they were indentations formed by the upper incisors. During embryogenesis, on day 32 four growth centres of the lower lip gets divided by two lateral grooves and one median. By day 38 the grooves get disappeared. When growth of the mandibular process is hindered, a lip pit occurs by the day 36. Cleft lip begins to develop on day 40 and cleft palate develops on day 50 of development.

Lip pits are classified according to their location into three types: midline upper, commissural, and lower lip. The common phenotype is two symmetrical lower lip pits flanking both sides of the midline in the bilateral paramedial sinuses and may also be present bilaterally, unilaterally, or medially asymmetrical.

There are different shapes for lip pits, as circular, oval & slit-like or transverse. The lip pits extend into the orbicularis oris muscle, ending in blind sacs surrounded by mucous glands. In some cases lip pits present as sinuses and mucous is excreted when the muscles contract(4).

Many other syndromes which are assessed as allelic variants of the syndrome and the following are considered in differential diagnosis of Van Der Woude Syndrome as,

Popliteal pterygium syndrome (PPS) which in closes popliteal web, cleft lip and/or cleft palate, lower lip pits, anomalies of genitourinary system, as cryptorchidism and bifid scrotum in male and hypoplastic labia majora and uterus in female. Also, people with Van Der Woude Syndrome have a risk of giving birth to offspring with PPS.

Hirschsprung's disease includes aganglionic megacolon combined with cleft palate and lip pits (5)

Orofacial digital syndrome type 1 has abnormalities in orodental, facial, digital, renal, and central nervous system. Orodental signs in closes as cleft palate, bifid tongue, hypodontia, median cleft of upper lip and/or lip pits. Based on orodigital findings this syndrome should be considered.

Ankyloblepharon filiform adnatum consists of partial or complete full thickness fusion of the lid margins cleft lip and palate, hydrocephalus, meningocele, imperforate anus, bilateral syndactyly, infantile glaucoma and cardiac problems such as patent ductus arteriosus and ventricular septal defects (6)(7)

Most commonly used procedure is fusiform excision with dissection of whole pit, split-lip advancement technique and inverted T-lip reduction provides perfect functional and aesthetic result. Appropriate management of Van Der Woude syndrome requires thorough knowledge of present surgical procedures(8).

CONCLUSION:

Nowadays, Van Der Woude syndrome are common in occurrence due to different mutations. Clinical features related to the VWS were left unobserved. So, we have to create adequate knowledge about the specific identifiable parameters along with the detailed genetic details to Oral Physician for providing prompt diagnosis. Also, Genetic counselling is crucial for diagnosis of VWS. All affected patients should be cautioned that they carry a risk of 50% for each child with a cleft lip or palate or both.

Conflicts Of Interest: Nil

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