

Van der Woude Syndrome (VWS) – A Case Report

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Abstract

Van Der Woude syndrome (VWS) is a rare genetic disorder characterized by paramedian lip pits, cleft lip and/ or cleft palate, hypodontia, syngnathia, narrow high arched palate and hyper nasal voice. Van der Woude syndrome is known to be caused by a mutation in a single gene with equal distribution between the sexes. In the present case report, a case on similar grounds wherein a patient has been diagnosed with this rare syndrome presenting with characteristic orofacial features and an unusual finding of bilateral paramedian commissural lip pits. The present article emphasizes upon etio-pathogenesis, clinical manifestations and multidisciplinary management of Van der Woude syndrome.

Key words : Van der Woude syndrome, lip pits, cleft palate.

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Introduction

Among the many disturbances of development and growth that involve the oral and perioral structures, cleft lip and palate is perhaps the commonest. Dental literature is replete with syndromes associated with cleft lip and palate, one of which is Van der Woude syndrome. The characteristics of Van der Woude syndrome is the association of cleft lip and/or palate with distinctive lower lip pits [1a]. Van der Woude syndrome (VWS), also known as lip pit syndrome or cleft lip syndrome is a rare developmental congenital malformation with an autosomal dominant disorder, with high penetrance and variable expressivity, occurring in about one of every 1,00,000-2,00,000 general population [1b,1c]. It was first observed by Demarquay in 1845, and was first reported by Epstein in 1900 and was extensively reviewed by Anne Van der Woude in 1954, that later received her name [2-4]. VWS is characterized by orofacial manifestations like lower lip pits, cleft lip with or without cleft palate, isolated cleft palate, hypodontia, cleft or bifid uvula, syngnathia, narrow high arched palate, ankyloglossia and hypernasal voice [5].

We hereby, present a case of a twelve year old boy, presented with such rare syndrome. A detailed history and a comprehensive clinical examination in conjunction with histopathological features led to the diagnosis of Van der Woude syndrome.

Case report

A twelve year old male patient had reported for routine dental check up with a past medical history of

surgical correction for congenital upper cleft lip and palate. His parent had history of consanguineous marriage and was only child, born after an uneventful full term pregnancy.



Figure-1: Bilateral Para median lower lip pits with watery discharge



Figure-2: Anterior cross bite, Mixed Dentition and prominent lip pits.

The patient had normal IQ but hyper nasal voice. Extra oral examination revealed straight facial profile, bilateral paramedian lower lip pits and surgical scar for upper cleft lip (Figure-1). Para median lip pits were bilateral, patent and had watery discharge. Intraoral examination revealed mixed dentition, anterior cross bite, bilateral surgically corrected cleft palate extending from premaxilla to palatal vault. (Figure-2, Figure-3). A biopsied specimen from lip pits revealed histologically

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epithelial fragment and connective tissue showed with modest cellular infiltrate. (Figure-4). Based on the clinical presentation, and histopathology the diagnosis of Van der Woude was suggested. The patient is presently being treated by multidisciplinary team comprising of stomatologist, orthodontist and child psychologist, giving prime consideration to growth and development of the facial structures, esthetics along with psychological support.



Figure-3: Surgically corrected palatal cleft with adjoining premaxilla to palatal vault.

Discussion

Van der Woude syndrome can be inherited as autosomal dominant trait or may develop as a spontaneous mutation. Most reported familial VWS cases have been linked to 1q32-q41 and a second locus has been mapped to 1 q 34. Mutations in the interferon regulatory factor 6 (IRF6) gene were demonstrated to cause VWS [6-8].

Although the lip pits are inherited as an autosomal dominant trait, their pathogenesis is not well understood. They are thought to develop from notching of the lip at an early stage of the labial development with fixation of the

tissue at the base of the notch or from failure of a complete union of the embryonic lateral sulci of the lip, which persist and develop into the typical lip pits [9]. Lip pits is the main manifestation of VWS and they occur in 88% of affected individuals.

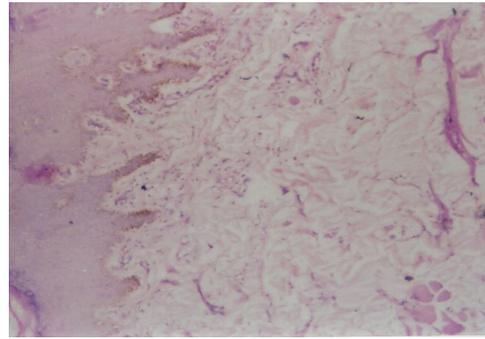


Figure-4: Histologically epithelial fragment exhibiting modest cellular infiltrate

The lip pits form canals, lined by labial mucosa, which extend inside the orbicularis oris muscle, their lengths being 1mm to 25 mm. These depressions are on a blind fundus or have minor salivary glands in their inner part [10]. The majority of the labial and commissural pits occur without exudation. However, an additional feature of mucous discharge was reported in our case. Surgical excision of the lip pits is indicated if the aesthetics of an individual is appreciably affected and exudation of the mucous secretions cannot be controlled [11]. Surgical excision should include the total removal of the minor salivary glands that exude secretions at the base of the lip pits, to prevent the formation of mucoceles or cysts.

The occurrence of weak forms of VWS should be kept in mind. Differentiation from Orofacial digital syndrome type 1 (lip pits, oral frenula, oral clefts, hypoplasia

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of nasal cartilages, malformation of hands, hypertelorism and psychomotor retardation), Popliteal pterygium syndrome (lower lip pits, oral clefts, syngnathia, popliteal web, toenail dysplasia, syndactyly of the toes, congenital heart disease and genital abnormalities) and Kabuki make-up syndrome (dysmorphic face, postnatal growth retardation, skeletal abnormalities, mental retardation and unusual dermato-glyphic patterns [5]. The present case exhibits a peculiar combination of all traits associated with Van der Woude syndrome. In order to carry out the philosophy of "total treatment", multidisciplinary management was carried out and resulted in a remarkable involvement of the patient aesthetics. Proper

evaluation and treatment of VWS along with genetic counseling is important. Care should be taken and meticulous oral hygiene care is imperative in such patients.

Conclusion

Congenital lip pits constitute a rare developmental malformation, transmitted by an autosomal dominant mode, with considerable heterogeneity as regards the expression of the disorder. Nevertheless, with prompt identification and appropriate understanding of clinical features, oral manifestations, the oral physician plays an important role in the diagnosis and management of patient with such rare syndrome.

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