

Congenital Lower Lip Pits (Van der Woude Syndrome): Report of a Case

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Abstract

Van der Woude syndrome (VWS) is a rare autosomal dominant disorder that is characterized by a cleft lip and palate with congenital lip pits. This is a report of a case of VWS with sinuses in the lower lip, a cleft in the upper lip, and a supernumerary tooth in the maxilla. The main characteristics of this disorder are discussed. Dental treatment of the patient was performed, but the surgical removal of the sinus was rejected by the parents. This case report brings this condition to the attention of dentists and surgeons and emphasizes lip pits may not always be identical in appearance.

Keywords: Van der Woude syndrome, congenital lip pits, supernumerary teeth

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Introduction

Van der Woude syndrome (VWS) is a rare autosomal dominant disorder typically involving a cleft lip or palate, distinctive pits on the lower lip, and hypodontia.¹⁻⁴ Pits are usually bilateral but may be unilateral or centrally placed on the vermilion border of the lower lip.^{1,3,5} Hypodontia is only rarely observed.⁶

Congenital lower lip sinuses have been reported in about 0.001% of the population, and 65% to 75% of the cases are associated with cleft lip and palate.^{7,8} Although a higher incidence in females has been reported, no genetic sex linkage has been found.⁹ It was suggested genetic defect of lip pits is due to a micro-deletion on chromosome bands 1q 32-q41.¹⁰ However, more recently Kondo et al.¹¹ reported that mutations in the IRF6 gene underlie a majority of cases with VWS including those with lip pits.

The following report describes a case of VWS with sinuses in the lower lip, a cleft in the upper lip, and a supernumerary tooth in the maxilla.

Case Report

A 9-year-old boy presented with a complaint of pain on the upper right second deciduous molar tooth. He had no significant past medical history. On examination, a sinus was noted next to the midline of the lower lip on the left side. In addition, on the other side of the lower lip, an abnormal appearance could be noted. The sinus was present since infancy. However, no

treatment has been attempted since there were no symptoms of saliva leakage, pain, swelling, discharge, and inflammation. As an associated deformity, a unilateral cleft lip on the left side was observed which was operated on when the boy was 3-months old (Figure 1). The midline of the maxillary teeth was observed to change due to the cleft lip operation. The patient demonstrated normal speech.

On intraoral and radiographic examinations, there was a supernumerary tooth in the maxillary anterior region between the upper left permanent lateral incisors and upper left deciduous canine. An abnormal morphology of the upper left deciduous canine was observed, and all of the primary second molar teeth were decayed (Figure 2).



Figure 1. A sinus is present just to the left of the midline on the lower lip along with an associated unilateral cleft lip which had been treated. The midline of the maxillary teeth was observed to change due to the cleft lip operation.



Figure 2. A preoperative panoramic radiograph.

All permanent teeth had dental fluorosis. No other developmental abnormality was detected on physical examination. Family history revealed the parent's relatives and the patient's grandfather had similar lesions.

We recommended surgical removal of the sinus on the lower lip. However, this treatment option was rejected by the parents of the patient.

Discussion

Congenital pits of the lower lip in association with clefting of the lip or palate or both occur in many syndromes.¹² Lower lip pits are characteristic in VWS, popliteal pterygium syndrome (PPS), alveolar synechia syndrome, type I orofacial digital syndrome, and ankyloblepharon filiforme adnatum.^{3,8-10,13,14}

The differential diagnosis of mild forms of such syndromes with VWS is difficult. Genetic counseling is critical for the diagnosis of VWS.⁶ There is some evidence VWS is associated with

a dominant autosomic gene of a high penetrance and variable expressivity.¹⁵ In this case report the patient had a family history of the syndrome, i.e., his grandfather had similar lesions. Moreover, his parents were children of siblings with VWS. All affected parents should be cautioned that they carry a risk of 50% for each child with a cleft lip or palate or both.¹⁶

Lip pits are generally treated surgically, and the most common indication is esthetic considerations. Our patient rejected this treatment because his lesions were asymptomatic and his parents could not afford the treatment expenses. He had a supernumerary tooth in the maxilla, however, hypodontia has been described in previous reports of VWS.^{2,12,15,17}

Summary

The case described here does not fit any syndromes similar to VWS, and it is suggested this case may be due to a variation of VWS.

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