

The syndrome of pit of the lower lip and its association with cleft palate

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Abstract

The Van der Woude syndrome is a rare autosomal dominant developmental malformation usually associated with lower lip pits, cleft lip and palate. These congenital lip pits appear, clinically, as a malformation in the vermilion border of the lip, with or without excretion. Obligate carriers of this dominant mutation may have lip pits alone, cleft(s) alone, clefts and pits or neither. Here, we present a case report of a 10 year-old male patient with Van der Woude syndrome and a brief literature on its treatment modalities.

Keywords: Ankyloglossia, bifid tongue, cleft palate, pit on lower lip, Van der Woude syndrome

Introduction

The Van der Woude syndrome is a rare autosomal dominant developmental malformation usually associated with lower lip pits, cleft lip and palate. These congenital lip pits appear, clinically, as a malformation in the vermilion border of the lip, with or without excretion.^[1] Obligate carriers of this dominant mutation may have lip pits alone, cleft(s) alone, clefts and pits, or neither.^[2] This syndrome is remarkably variable. Lower lip pits have been described with other anomalies, including syndactyly, ankyloblepharon, ankyloglossia, and polythelia.^[3] Congenital adhesions (synechia) between different parts of the oral cavity rarely occur.^[3-5]

The prevalence of Van der Woude syndrome has been reported to be 1:75,000 to 100,000.^[2,3] Lip pits are depressions of the lower lip that contain the orifice of mucous glands or minor salivary glands. They usually occur on either side of the midline of the lower lip and are generally bilateral.^[2] The mode of inheritance of this syndrome is autosomal dominant inheritance with variable expressivity. The gene responsible for this disorder has

been mapped to the long arm of chromosome 1 at q32 to q41.^[6] Carriers of the dominant mutation of Van der Woude syndrome manifest variably: 44% pits without clefts, 26% pits with clefts, 10% clefts without pits and 20% neither clefts nor pits.^[2,6] Reported here is a case of Van der Woude syndrome associated with lip pit with cleft palate, hypodontia and ankyloglossia.

Case Report

A 10 year-old male came to the Department of Pedodontics with the chief complaint of missing teeth and persisting pit on the lower lip. Past medical history was not significant. Family history did not reveal any similar complaints or findings in the immediate or distant relatives.

Extraoral examination revealed convex profile with retrognathic chin [Figure 1]. History revealed presence of unilateral lower lip pit approximately 2 mm in diameter since birth with no exudation and no sign of regression in the size of the pit [Figure 2]. There was presence of ankyloglossia associated with a significant bifid tongue that resulted in slurred speech [Figures 3 and 4]. There was a submucous cleft palate in the anterior region with a narrow high arch vault [Figure 5]. The lower arch exhibited hypodontia.

Treatment plan included restoration of carious lesions and extraction of grossly decayed root stumps. The patient was scheduled for surgical correction of ankyloglossia and was advised to go in for correction of cleft and subsequent speech therapy. The patient was also advised to go in for prosthetic rehabilitation to compensate for hypodontia.

Discussion

Van der Woude syndrome is an autosomal dominant syndrome characterized by a cleft lip or cleft palate, distinctive pits of the lower lips or both.^[7] It is the most common syndrome associated with cleft lip or cleft palate. The degree to which individuals who carry the gene are affected widely varies,

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Figure 1: Convex profile with retrognathic chin



Figure 2: Unilateral lower lip pit



Figure 3: Ankyloglossia



Figure 4: Bifid tongue



Figure 5: Submucous cleft palate with narrow high arch vault

even within families. These variable manifestations include lower lip pits alone, absent teeth or isolated cleft lip and cleft palate of varying severity. Hypodontia (absent teeth) has been increasingly recognized as a frequently associated anomaly.

Hypodontia has been observed in 10-81% of all Van der Woude syndrome, patients with the number of teeth missing in the upper jaw being more frequent.^[3,8] Our case presented hypodontia with missing teeth in both the arches.

Many other associated anomalies have also been described. Other frequently associated anomalies include syndactyly of the hands, polythelia, ankyloglossia and symblepharon^[9,10] club foot,^[11] thumb hypoplasia,^[12] oral synechiae and tapering finger.^[13] The present case showed presence of ankyloglossia along with other significant findings.

Lip pits in Van der Woude syndrome 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 ultimately develop into the typical pits.^[14]

The surface opening of the lip pit may present as a circular or transverse slit or be located at the apex of nipple-like

elevations and measure up to 3 mm in diameter. The depth of the pits ranges from 1 to 15 mm.^[15] Minor salivary gland orifices open into the pits and hence the salivary exudate. Although the cleft lip and palate are the major esthetic problems for these patients, exudation of mucous from the lip pits onto the lower labial skin is a source of embarrassment to the patient. Our case reported presence of lip pit approximately 2 mm in diameter with no exudation.

The most prominent and consistent features of Van der Woude syndrome are orofacial anomalies caused by an abnormal fusion of the palate and lips at 3050 days postconception. Most cases of Van der Woude syndrome have been linked to a deletion in chromosome 1q32-q41; however, a second chromosomal locus at 1p34 has also been identified. The responsible mutation has been identified in the interferon regulatory factor6 (IRF 6) gene, but the exact mechanism of this mutation on craniofacial development is uncertain.^[3]

Demonstrating the presence or absence of an IRF6 mutation can be helpful when distinguishing between uncomplicated cleft lip and/or cleft palate and Van der Woude syndrome. A wide variety of chromosomal mutations that cause Van der Woude syndrome and are associated with IRF6 gene mutations have been described. A potential modifying gene has been identified at 17p11.2p11.1.^[3]

Because of its variability, obtaining a detailed family history is important in diagnosing Van der Woude syndrome. However, approximately 3050% of all cases of Van der Woude syndrome arise as a *de novo* mutation. The present case showed no relevant family history.

Van der Woude syndrome equally affects both sexes. We reported a male case. A single, small study has suggested that males with the syndrome may have poorer cognitive function than females.^[15]

Conclusion

Van der Woude syndrome is a rare clinical entity. The phenomenon that Congenital lip pit (CLP) and Cleft palate (CP) are regularly combined in the same pedigree makes it unique. A case of Vander Woude syndrome is presented. Additional clinical features noted were enamel multiple congenitally missing teeth, ankyloglossia and cleft palate. Expressivity varies widely and careful clinical examination of parents and relatives may be necessary. Physical examination of relatives, close examination of

family photos or interviews of older relatives may be necessary to identify minimally affected family members. Genetic counselling is highly recommended.

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References

1. Van der Woude A. Fistula labii inferioris congenita and its association with cleft lip and palate. *Am J Hum Genet* 1954;6:244-56.
2. Cervenka J, Gorlin RJ, Anderson VE. The syndrome of pits of the lower lip and cleft lip and/or palate. Genetic considerations. *Am J Hum Genet* 1967;19:416-32.
3. Rizos M, Spyropoulos MN. Van der Woude syndrome: A review. Cardinal signs, epidemiology, associated features, differential diagnosis, expressivity, genetic counselling and treatment. *Eur J Orthod* 2004;26:17-24.
4. Oberoi S, Vargervik K. Hypoplasia and hypodontia in Van der Woude syndrome. *Cleft Palate Craniofac J* 2005;42:459-66.
5. Shaw WC, Simpson JP. Oral adhesions associated with cleft lip and palate and lip fistulae. *Cleft Palate J* 1980;17:127-31.
6. Sander A, Schmelzle R, Murray J. Evidence for a microdeletion in 1q32-41 involving the gene responsible for Van der Woude syndrome. *Hum Mol Genet* 1994;3:575-8.
7. Janku P, Robinow M, Kelly T, Bralley R, Baynes A, Edgerton MT. The van der Woude syndrome in a large kindred: Variability, penetrance, genetic risks. *Am J Med Genet* 1980;5:117-23.
8. Ranta R, Rintala A. Tooth anomalies associated with congenital sinuses of the lower lip and cleft lip/palate. *Angle Orthod* 1982;52:212-21.
9. Neuman Z, Shulman J. Congenital sinuses of the lower lip. *Oral Surg Oral Med Oral Pathol* 1961;14:1415-20.
10. Burdick AB, Ma LA, Dai ZH, Gao NN. van der Woude syndrome in two families in China. *J Craniofac Genet Dev Biol* 1987;7:413-8.
11. Ludy JB, Shirazy E. Concerning congenital fistulae of the lips; their mooted significance; review of the literature; and report of a family with congenital fistulae of the lower lip. *New Internat Clin* 1937;3:75-88.
12. Wong FK, Karsten A, Larson O, Huggare J, Hagberg C, Larsson C, *et al.* Clinical and genetic studies of Van der Woude syndrome in Sweden. *Acta Odontol Scand* 1999;57:72-6.
13. Christian J, Gorlin RJ, Anderson VE. The syndrome of pits of the lower lip and/or palate: Genetic considerations. *Clin Genet* 1971;2:95-103.
14. Everett FG, Wescott WB. Commisural lip pits. *Oral Surg* 1961;14:202-9.
15. Nopoulos P, Richman L, Andreasen N, Murray JC, Schutte B. Cognitive dysfunction in adults with Van der Woude syndrome. *Genet Med* 2007;9:213-8.

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