SHORT COMMUNICATION

Van der Woude syndrome with an unusual intraoral finding

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ABSTRACT

Orofacial manifestations of Van der Woude syndrome (VWS) include cleft lip or palate, lower lip pits, hypodontia, hypernasal voice, cleft or bifid uvula, syngnathia, narrow high arched palate, and ankyloglossia. Extraoral manifestations include limb anomalies, popliteal webs, accessory nipples, congenital heart defects, and Hirschsprung disease. We report an interesting case of VWS with characteristic orofacial features along with an unusual additional finding of fusion of primary mandibular left lateral incisor and canine in a 7-year-old boy.

Key words: Ankyloglossia, cleft lip, cleft palate, fusion, hypodontia, lip pits

Van der Woude syndrome (VWS) is a rare developmental, congenital malformation with autosomal dominant inheritance, high penetrance, and variable expressivity, occurring in about 1 of every 1,00,000–2,00,000 people. It is characterized by pits and/or sinuses of lower lip, cleft lip with or without cleft palate, isolated cleft palate, bifid uvula, and hypodontia.[1] Pits are usually bilateral but may be unilateral or centrally placed on the vermilion border of the lower lip. Hypodontia is only rarely observed. Congenital lower lip sinuses have been reported in about 0.001% of the population, and 65–75% of the cases are associated with cleft lip and palate.[2] Although a higher incidence in females has been reported, no genetic sex linkage has been found. It was suggested that genetic defect of lip pits is due to a microdeletion on chromosome bands 1q 32–q41.10. However, more recently Kondo *et al*.[3] reported that mutations in the IRF6 gene underlie a maturity of cases with VWS, including those with lip pits. The following report describes a case of VWS with pit in the lower lip, a cleft in the upper lip, cleft palate, and a fusion of deciduous lateral incisor and canine.

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CASE REPORT

A 7-year-old boy was referred to the Department of Oral Pathology and Microbiology with the chief complaint of nasal regurgitation and nasal twang. Past surgical history revealed that the patient had undergone surgical correction of upper cleft lip at the age of 18 months. The family history did not reveal consanguineous marriage of his parents. The patient's parents and his 9-year-old sister did not exhibit any related findings after thorough intraoral and radiographic examination but intraoral examination of 6-and 3-year-old brothers revealed ankyloglossia. According to his mother, the patient was born after an uneventful, full-term pregnancy with no exposure to radiation. The extraoral examination revealed surgical scar of the upper operated cleft lip and bilateral, paramedian small pits on the lower lip [Figure 1a]. Intraoral soft tissue examination showed bilateral cleft palate extending from premaxilla to soft palate (Veau's classification: Group 4) and ankyloglossia [Figures 1b and 1c]. Dental examination revealed mixed dentition, missing deciduous maxillary lateral incisors, and the fusion of the deciduous mandibular left lateral incisor and canine [Figure 1d]. An orthopantamograph disclosed bilateral maxillary cleft and fusion of deciduous mandibular left lateral incisor and canine. Deciduous and permanent maxillary lateral incisors were absent [Figure 2d]. Intraoral periapical radiograph confirmed fusion of deciduous mandibular left lateral incisor and canine [Figure 2c]. The depth of lower buccal pits were assessed by placing gutta-percha in it [Figure 2a and 2b].

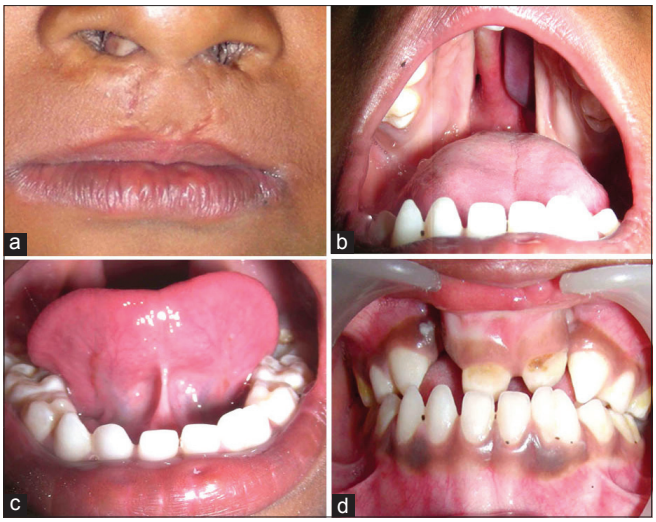

Figure 1: Clinical photographs showing operated upper cleft lip and bilateral paramedian lower lip pits (a), (b) cleft palate, (c) ankyloglossia, and (d) bilateral cleft alveolus and missing deciduous lateral incisors in the maxilla with anterior cross bite

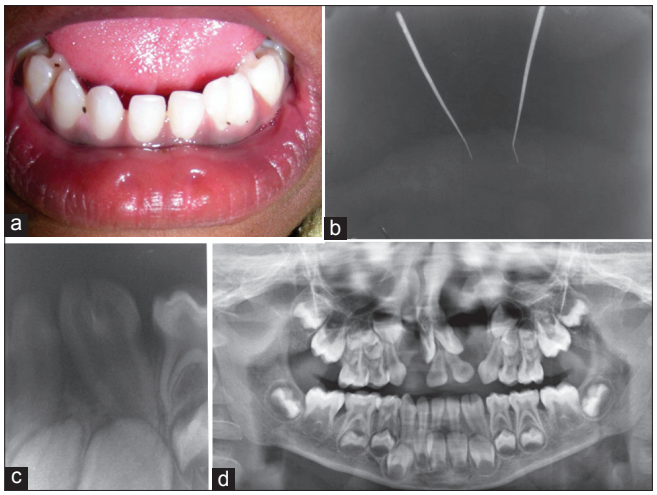

Figure 2: (a) Photograph showing labial pits and fusion, (b) radiograph of lower buccal pits by placing gutta-percha, (c) IOPA showing fusion, OPG showing missing maxillary bilateral deciduous and permanent lateral incisors and mandibular left permanent lateral incisor, (d) cleft alveolus and palate, and fusion

DISCUSSION

Although VWS was mentioned as early as 1845 by Demarquay, the eponymic credit was given to van der Woude for reviewing the syndrome extensively in 1954.[1] It 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 but a second locus has been mapped to 1q34. Mutations in the interferon regulatory factor 6 (IRF6) gene were demonstrated to cause VWS. So far, 46 mutations in IRF6 associated with VWS have been identified.[4-6] Orofacial manifestations of VWS include cleft lip or palate, lower lip pits, hypernasal voice, cleft or bifid uvula, syngnathia, narrow high arched palate, and ankyloglossia. The occurrence of weak forms of VWS should be kept in mind. Differentiation from the Orofacial digital syndrome and from the Popliteal pterygium syndrome, which also occur with lower lip pits and clefts is essential. The present case exhibits a peculiar combination of all traits associated with VWS with an unusual additional finding of fusion of the deciduous left mandibular lateral incisor and canine. However, the fusion of deciduous teeth is not uncommon in nonsyndromic patients, it is a rare finding in syndromic patients. In the present case, the fusion may have occurred incidentally and may not be necessarily related to the VWS syndrome.

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