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Van der woude syndrome. Cardinal signs, epidemiology, associated features, differential diagnosis and treatment: a review

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Corresponding Author:
Dr. Martina Maria D'emidio,
Attender, Oral and Maxillo Facial Sciences Department, Orthognatodontics Unit, La Sapienza - Italy

Submitting Author:
Dr. Martina Maria D'emidio,
Attender, Oral and Maxillo Facial Department, Orthognatodontics Unit, La Sapienza - Italy

Other Authors:
Dr. Emanuele Fantasia ,
Attender, Oral and Maxillo Facial Sciences Department, Orthognatodontics Unit, La Sapienza - Italy
Dr. Giuseppe Rodi,
Attender, Oral and Maxillo Facial Sciences Department, Orthognatodontics Unit, La Sapienza - Italy
Dr. Gabriella Padalino,
Attender, Oral and Maxillo Facial Sciences Department, Orthognatodontics Unit, La Sapienza - Italy
Dr. Elisa Lombardelli,
Attender, Oral and Maxillo Facial Sciences Department, Orthognatodontics Unit, La Sapienza - Italy

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Author(s): D\'emidio M, Fantasia E, Rodi G, Padalino G, Lombardelli E

Abstract

The Van der Woude syndrome (VWS), is a rare autosomal dominant disease due to a genetic mutation. The cardinal sign of this syndrome are labial fistulas. It is often associated with cleft lip, labial and palate. Cleft lip in 66.2% of cases, labial in 16,5% of cases and palatal in 17.3% of cases. The treatment of fistula is exclusively for aesthetic reasons, so the main treatment is that of lip and palatal cleft. It is important so that you can reach good results a multidisciplinary approach.

Introduction

Congenital malformations are known since the dawn of humanity, as shown by engravings and figurines witnesses of past civilizations, found in various parts of the world. Some consider cleft lip fissures date back to 2000 b.c. The birth of a malformed child has always caused consternation, but the explanation of this phenomenon has occurred has varied at different times, according to the magical-religious or philosophical concepts prevalent. Thus, in some cultures a malformed child was considered an impure being, he should not live and then it was destroyed; while in others it was deified and worshiped. In some religions it is considered as the result of sin, and therefore as divine punishment; while others represent harbinger of future events, by disagreements between the divinities or cosmic wars. A mid nineteenth century teratology born as the science that deals monstrosities, and in recent decades has been coined the term dysmorphology to refer to the science of congenital malformations. They are considered congenital malformations, those structural defects present at birth. Due to the complexity of the disease that have these malformed patients, a multidisciplinary approach is needed for rehabilitation.[1]

The Van der Woude syndrome (VWS), is an autosomal dominant disease due to a genetic mutation level IRF6 gene located in the 1q32-q41 chromosome, characterized by the presence of fistulae at the level of the lower lip usually related to salivary glands heterotopic and clefts : cleft lip in 66.2% of cases, labial in 16,5% of cases and palatal in 17.3% of cases.[2-3] In addition known as associated defects: hypodontia or tooth agenesis, in order of frequency: second upper premolars, second lower premolars and lower lateral incisors, tongue tie, submucosal fissures and bifid uvula and adhesions between maxilla and mandible, ankyloblepharon or alterations to the tips like syndactyly. Other commonly associated anomalies are: polythelia, hypoplasia of the fingers, clubfoot, heart disease, side graves in corners of the lip and upper lip but are rare, drum fingers, preauricular pits and double lip.[2]

EPIDEMIOLOGY:

The VWS prevalence ranges from 1: 100000 to 1: 40000. There is no significant difference in sex, but no chord in the literature, being by many authors the same in women and men while others claim to be greater in women for coming more often to the doctor and others say be more frequent in the male.[4]

Methods

In the recent years, several dentistry works have been published on international literature about the syndromes associated with lip and palatal cleft. So a detached research of international literature on the diagnosis and all the possible treatment methods for these anomalies has been performed using the principal medical databases: PubMed (Medline), Lilacs and Scopus. The keywords used were: Van der Woude Syndorme, malformations, cleft lip and palate and labial fistulas; to identify all articles reporting on the topic untill October 2016. No restrictions of time and languages have been fixed. The results have been filtered and valued following our eligibility criteria and then organized following the PRISMA method. The search identified 3460 abstracts, which were reviewed manually and each article of interest was marked for further review. The full text of the marked studies was retrieved and studies that satisfied our eligibility criteria were included in this review. At the end only 13 full articles have been selected.

Discusion

Labial fistulas:

They are the cardinal sign of this syndrome, the most common form of labial fistula is bilateral, being located at the lower lip symmetrically on either side of the midline. They can also occur unilaterally, bilaterally mesial or asymmetrical. Unilateral forms are considered an incomplete expression of the syndrome and occur more often on the left lip. Fistulas often have circular or oval shape, and less frequently shaped slit or groove, due to the fusion of one or more of them together. From its locations canals generate the labial mucosa lining extending into the orbicularis muscle. Through fistulography you can see the path of these channels and their branches. Fistulas occasionally diverge and rarely converge to form a single path. The opening of the pipes can be found flush with the lower lip causing a depression in this area. Labial fistulas are usually asymptomatic, the only symptoms that can cause relates to the continuous or intermittent drainage of secretions salivary can occur spontaneously or be caused by chewing. Mucous secretions are more abundant before and during meals and crying baby.[5]

Differential diagnosis:

The differential diagnosis should include other syndromes associated with lower lip fistulas as popliteal pterygium syndrome and gold-digit-facial dysostosis.[4]

Cleft lip and palate:

The problem of cleft lip and palate, occurs between the 6th and 10th weeks of embryo-fetal life. A combination of failure in the normal binding and inadequate development, can affect soft tissue and bone components of the upper lip, alveolar ridge, and the hard and soft palates. The causes of congenital malformations are very different and varied, however we can gather them into 2 groups: genetic and environmental.[1] In the case of VWS we have a genetic cause with monogenic autosomal dominant inheritance pattern.

Classification of cleft lip and palate:

The varied morphology that can result in clefts alveolus lip palatal to involve the deformity of 4 different structures: the lip, alveolar process, hard palate and soft palate, coupled with the possibility that the alteration is unilateral or bilateral, it has always been a challenge to universally adopt a single classification; and if this is added the modern idea that the classification should be based not on anatomical facts of the fetus to term, but in the embryological data that led to deformity, is that virtually every student of this problem has made its own classification.[1] Among the traditional classifications used to establish cleft lip and palate are the Davis and Ritchie (1922), Veau (1931), Pfeiffer (1964), Kernahan (1971), Millard (1976) and Tessier (1979). All these makes a description of the anatomical segments involved in the cleft but not calculate the magnitude of the defect. There classifications like Mortier, who considers tissue deficiency, however does not consider the diameter of cleft lip and palate. Another more recent classification is Posadas Ortiz, describing the severity of the cleft (magnitude of tissue deficiency) considering three components: nose-lip, palate primary and secondary. This classification is based on the vertical and horizontal deficiency of tissues in the cleft considering even quite specific aspects such as muscle integrity, lip thickness, depth of sulcus, and other more detailed but still complex and difficult to remember.[6] The classification of cleft is made according to the structures involved: lip, gums, hard palate, soft palate. It also defines whether it is complete or incomplete unilateral; or bilateral, which can be symmetric or asymmetric.[7] Kernahan classification is the most widespread in the world for its simplicity of use filling and reading, but does not provide information regarding the extent of tissue deficiency in the cleft.[7-8]

Another diffuse classification is accepted by the international confederation in Rome in 1967 which divides clefts into four groups depending on the location relative to incisive foramen: [10-11-12]

I. Clefts previous palate incisor foramen (primary palate)

II. Clefts anterior and posterior palate (transforaminal)

III. Clefts palate posterior to anterior incisor foramen (secondary palate)

IV. Rare facial clefts

Treatment:

As mentioned earlier lip dimples are generally asymptomatic but can perform surgical excision for aesthetic reasons or to reduce the secretion of mucus or salivary flow. So the treatment of VWS consists mainly of surgery cleft lip / palate.[4]

Patients with VWS who have cleft lip and palate need a multidisciplinary team care setting priorities according to the general condition of the patient, the severity of the defect and the age of it. On the site of the malformation aesthetic impact is important and

vital functions are involved: breathing, swallowing, phonation and mastication being the most affected swallowing and phonation.[11]

Treatment protocol:

This treatment protocol, fits the diagnosis of each case (cleft lip and palate, cleft palate, cleft lip isolated) and is based on six pillars:[7]

1. Early assessment and guidance to the patient and their parents by a multidisciplinary team. The initial diagnosis of Cleft Lip Palatine can be in utero (increasing), starting at 16 weeks of gestation by an ultrasound, or at birth. In this first evaluation we need to diagnose the existence of other malformations or pathology associated as may be the VWS.
2. Pre-surgical, orthopedic involves the use of devices that allow stimulation and bone remodeling nasal segments, alveolar and palatine cleft, decreasing the size of clefts, during the first 3 months of life, shaping it as close to the anatomy normal before surgery lip and cleft palate.[13]
3. Primary cleft lip surgery with nasal surgery and in cases where there alveolar cleft with gingivo make contact (closure of the alveolar ridge).
4. Postoperative nasal modeling with nasal hook, or other nasal splint to avoid scar retraction and decrease residual deformity.
5. Plasty of the soft palate and hard palate in a time between 12 and 16 months.
6. Assess need for secondary surgeries in each individual case.

Conclusions

The Van der Woude syndrome is a rare condition. Its cardinal sign the presence of labial fistulas whose treatment by surgical excision is performed only if aesthetic need. It is often associated with cleft lip and palate which is difficult to find a unanimous classification between different professionals. It requires a multidisciplinary approach both dental treatment: prosthetic, orthodontic and surgical as child care and your family: speech therapy and psychological.

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