CASE REPORT

Indonesia Journal of Biomedical Science (*IJBS*) 2020, Volume 14, Number 2: 118-121
P-ISSN.2085-4773, E-ISSN.2302-2906

Published by DiscoverSys

Van der woude syndrome: A case report and review

Nyoman Ayu Anggayanti[1*], Endang Sjamsudin[2], Farah Asnely Putri[2]

ABSTRACT

Introduction: The prevalent autosomal disorder, with or without cleft palate and lip, is Van der Woude Syndrome (VWS). The condition has been known for its lower lip pits. Interferon Regulatory Factor 6 (IRF6) gene mutations are causing a range of cases of VWS. The furrows caused an accumulation of mucous and felt esthetic pain during meals and crying.

Purpose: To determine VWS causes, prevalence, treatment, symptoms, and diagnosis.

Case report: A six-month-old baby was with lip pits, a bilateral cleft lip and palate since birth. The extraoral exam showed bilateral pit swellings with dome form on the lower vermilion boundary laterally to the midline. Simultaneous operations with the Nordhoff technique and simple excision were conducted combined with a breakthrough procedure under general anesthesia to correct bilateral cleft lips. The patient showed better clinical condition after the seventh-day post-surgery without infection sign, and bilateral cleft in lower lip pit closed in a month after surgery.

Conclusion: VWS is generally not known and not always diagnosed. It is peculiar that the cleft palate and lip phenomenon is routinely integrated into the same pedigree. The patient with a lip pit can be thoroughly examined, for example, by revealing the secret shape of the cleft. In this case, the surgical repair was handled simultaneously to both functional and esthetic aspects to achieve satisfying results.

Keywords: van der woude syndrome, cleft lip and palate, lip pits

Cite this Article: Anggayanti, N.A., Sjamsudin, E., Putri, F.A. 2020.Van der woude syndrome: A case report and review. *IJBS* 14(2): 118-121. DOI: 10.15562/ijbs.v14i2.269

[1]Departement of Oral and Maxillofacial Surgery, School of Dentistry, Faculty of Medicine, Universitas Udayana, Denpasar, Indonesia
[2]Departement of Oral and Maxillofacial Surgery, Faculty of Dentistry, Universitas Padjadjaran, Bandung, Indonesia

*Corresponding author:
Nyoman Ayu Anggayanti;
Departement of Oral and Maxillofacial Surgery, School of Dentistry, Faculty of Medicine, Universitas Udayana, Denpasar, Indonesia;
ayu.anggayanti@unud.ac.id

Received: 2020-09-21
Accepted: 2020-11-23
Published: 2020-12-01

INTRODUCTION

The Van der Woude Syndrome (VWS) is a sinus disorder that included lower lip, a flaccid lip and palate.[1] In 1954, Van der Woude described this function, investigated it extensively, developed a connection between the lower lip or sinus, and introduced a new clinical perspective to the heritage mode.[1] The lip-pit typically has salivary glands, which drain into the lip-pit and result in salivary flow. The patient lip pits can associate with patient dizziness symptoms, velopharyngeal abnormalities or genital or cardiovascular problems.[2] They are typically positioned on the lower vermilion portion on either side of the midline with symmetrical and bilateral. They may also be asymmetrical on a unilateral, medial, or bilateral. One lesion is typically used as an incomplete syndrome.[2] Lip pits are oval shape, transverse, and even sulcious. Cross-cut mucosa and conical (nipple-like) elevations are considered lower lip microforms. The sinuses penetrate to a depth of 1-25 mm in the orbicular muscle and interact with the small salival glands beneath them. They are mostly asymptomatic, but watery or salivary secretions are intermittently or continuously drained.[3]

VWS has high penetration and variable expressiveness from 89% to 99%. Live births from 1:35 thousand to 1:100 thousand and 1 to 2 percent. Seventy percent of lip wells are associated with the palate and even lip.[4] Thirty percent have minimum results like isolated lip wells and hypodontia. Most cases of VWS are due to IRF6 mutations. In the 1q32-41 field, VWS was related to 500 to 800 kilobases deletion.[5] These cases are unusual. The VWS case and its literature, we are interested in covering below.

CASE REPORT

A six-month male infant with lip pits, cleft palate, and bilateral cleft lip came to the Cleft Center University of Padjadjaran Bandung. Parents complained about cleft lip, palate, and drooling issues since childbirth. Uncomplicated deliveries and normal births have been confirmed, and there has never been a history of drug and radiation exposure during pregnancy.

A general physical condition test was found with 7 kg body weight within the normal range. We observed bilateral dome-shaped swellings with pitches on the midline on the lower vermilion boundary with a complete cleft palate and lip had been diagnosed. (Figure 1).

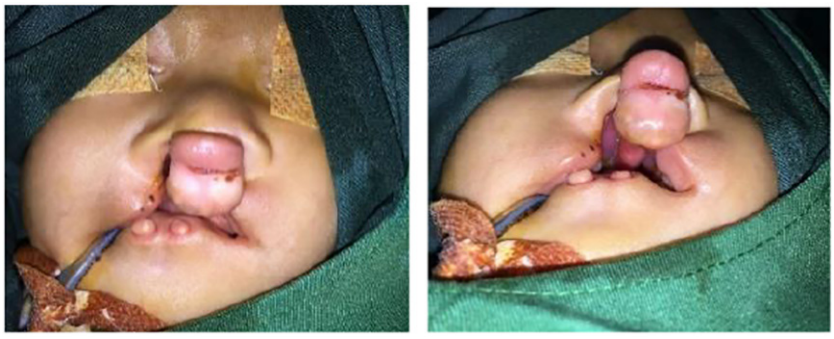
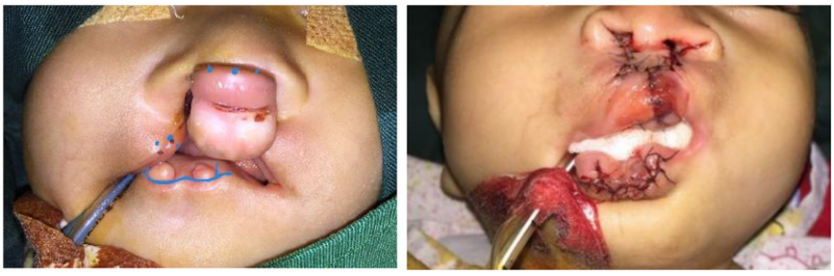
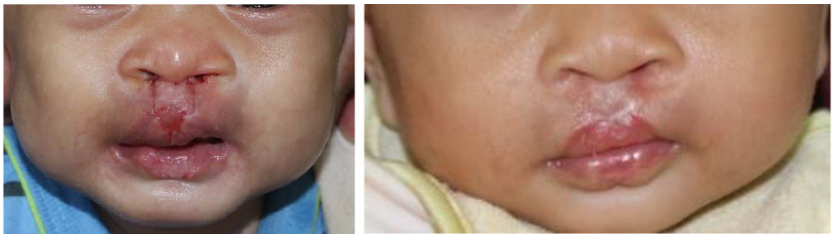


Figure 1. Bilateral cleft lip and lower pits lip preoperative picture



(A) (B)

Figure 2. During operation (A). The lips with marking side, (B). Patient's condition after surgery



(A) (B)

Figure 3. (A). The suture was extracted seven days after surgery (B). A month after the operation

Treatment for bilateral cleft lip correction and simple excision with general anesthesia is envisaged. The infant made eligible for preoperative and perioperative care with a laboratory test, a thorax X-ray, and cardiopulmonary in the Department of Pediatrics and the Department of Anesthesia. Her general anesthesia treatments did not include a contraindication, and the patient's parents signed informed consent.

The Nordhoff technique and an exact cut combination with a split-lip advancing approach were used to correct the bilateral cleft lip under general anesthesia surgically. The patient was aseptic and antiseptic and had a supine role under general anesthesia. Marking for this cleft lip repair is the same as the methylene blue mark (Figure 2A) and follows the same principles. Following the diagram's completion, lidocaine 2 percent plus epinephrine 1:200,000 is infiltrated into the lip clamp has been inserted during the procedure to decrease bleeding. The incisions and the dissections are rendered as usual by other methods according to the mark, and the repair is closed by three layers: first, the mucosa, muscles, and skin (Section 2B). The procedure must be performed simultaneously to correct the bilateral cleft lip with lower lip pits of the infant to achieve appropriate findings both in functional and esthetic terms.

Following surgery, the patient has treated with amoxicillin 100mg. After the treatment, his lip looked like mild swelling oedematous, no bleeding, no fever. Patients use a spoon for drinking milk. Patients are permitted to return, and the treatment is continued. Polyclinic monitoring is promoted in patients. After seven days of surgery, a suture has been removed, no signs of infection exist (Figure 3A), and the bilateral cleft, lower lip pit were closed, and a follow-up has been closed for a month after surgery. (Figure 3B).

DISCUSSION

Van der Woude was the firstperson companion to combine lower lip troughs with cleft palate and lip which introduce a newly clinical perspective, thus defining the heredity mode as a whole. Bocian et al. documented a lip pit patient in 1887 and 1q32-q41 deletion. Murray et al. followed by a link between VWS and the markers in the same area. Bocian et al. found in families with VWS in 1q32-q41 microdeletions.[6] The second locus of VWS was located in 1p34 chromosome. The IRF6 mutation in patients with VWS has recently been identified. VWS cases are usually related to chromosome 1q32-q41 (VWS 1), and chromosome 1p34 for the second locus (VWS 2). IRF6 are coded by the gene that located in chromosome 1q32-q41. IRF are a family of transcription factors. Nine IRFs in humans are registered. IRF6 is still uncertain for its role, meanwhile the other IRFs has the known role.[6]

The mandibular arch and the lower lip sulcus lateral fusion occurs at 5.5 weeks during normal development, while the maxillary and nasofrontal fusions take place at a period of six weeks. A joint event can, at the same time, interrupt fusion at either location. It is potentially that lip pit and cleft lip and palate are closely related.[5] Parents frequently guested these pits due to eruption of maxillary incisors, although the pits have been present since the beginning, months before the maxilla incisors' eruption. As standard, our patients' depressions were thought to have formed due to tooth pressure, and there was no any other evaluation in our patient. Therefore, any other lip-pit anomalies need

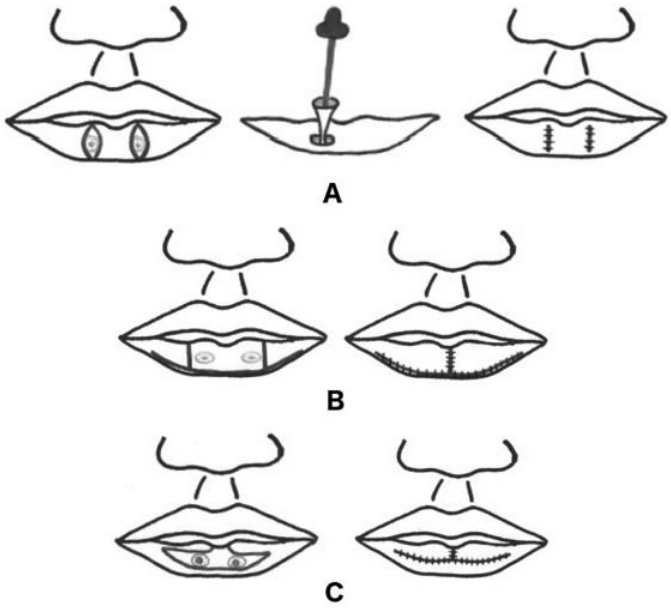


Figure 4. Drawings of operational strategies available in VWS to treat lower lip pits. A. The lower lip pit has fusiform excision, helped by the use of a tear-sond. B. Mutaf et al. identified the split-lip progression flap. C. Chen and Chen outlined the technique for inverted T-lip reduction.

to be investigated, since the pits are generally not noticed by the physician.[4]

Operational excision is the treatment of the pits. It is important to ensure the entire tract is appropriately removed when cutting the pits because salivary tissue can result in cyst formation in any residual tract. The tract will vary in diameter between a small orifice and a 6 mm long orifice. Since tracts can bifurcate, they can leave tract branches behind with a lacrimal probe to trace the track. We use bacitracin ointment regularly combined with methylene blue teal to trace and use during excision and, in this case, preauricular pits, sinuses, and cysts. Cautious precautions should also be taken when exploring neonates with the lip or the palate to search for the lesions of lower lips, because will imply to diagnosis of VWS and future genetic counseling will be required.[7]

Due to the variable expressivity of VWS, one should be aware of microforms that are often difficult to determine, such as lower lip pits and cleft lip, including cleft alveolar or bifid herb, and cleft palate submucous. Family history is significant and three generations of pedigree are recommended. VWS is a divided lip or palate, so need a palate or lip surgical reconstruction and multidisciplinary follow-up.[8] The lower lip wells may be asymptomatic, while watery drainage has been seen regularly or intermittently in most patients. There are also cosmetic effects. Our experience has shown that the diameter of the lower lip fistulae is more than the clinical trials initially predicted (average 15.7 mm). This depth means that the orbicularis oris muscle is similar to its aesthetic effects if not well remedied. This must be taken into account before surgery.[7] Mutaf et al. explains the classic elliptical cutting and split-leap technique, includes two methods used for removal of fistula. We used elliptical simple for fistula excision and had good results (Figure 4).

CONCLUSION

Doctors should be aware for congenital condition of the variable lip pit, which has clinical variables of VWS, and this disorder is hard to identify.[6] While lip pits show that the VWS is present as the exclusive clinical finding, the negative correlation with previously recorded chromosome 1q32-Q41 deletion and the lack of a mutation of IRF 6 in our case indicates that the phenotypic expression of the 2nd modifier gene may be affected. Genetic therapy is recommended for a potential pregnancy because VWS has high penetration and a variable voice.[9] Surgery correction, combined with the technique of Nordhoff and simple excision combined with dividing layer advancement technology, must be rendered at the same time to achieve acceptable results on the practical as well as on the esthetic aspects. Therefore, the analysis was made to be aware of VWS, such as signs, symptoms, and treatments.

AUTHOR CONTRIBUTION

All authors have contributed to all processes in this case report, including preparation, data gathering, and data presenting, drafting, and approval for publication of this manuscript.

CONFLICT OF INTEREST

The authors declare no conflict of interest regarding the publication of this case report.

FUNDING

This case report was not funded by any grant or any external funding.

ETHICAL STATEMENT

This case report has approved by local ethic commission.

REFERENCES

1. Tehranchi A., Behnia H., Nadjmi N., et al. Multidsiciplinary management of a patient with van der Woude syndrome: A case report. International Journal of Surgery Case Reports. 2017;30:142–147.

2. Bertin H., Diallo-Hornez, Isidor B., et al. Surgical management of lower lip pits in Van der Woude syndrome. J Stomatol Oral Maxillofac Surg. 2018;119:67–70.
3. Retno Widayanti, et al. Congenital double lip pits: A case report. Padjadjaran Journal of Dentistry. 2016;28(1):62-67.
4. Baghestani S., Sadeghi N., Yavarian M., et al. Lower lip pits in a patient with van der Woude syndrome. J Craniofac Surg. 2010;21:1380-1381.
5. Lam AK., David DJ., Townsend GC, et al., Van der Woude syndrome: dentofacial features and implications for clinical practice. Australian Dental Journal. 2010;55: 51–58.
6. Trishala.A et al., Van Der Woude Syndrome – A Review. J. Pharm. Sci. & Res. 2016;8(6):495-497.
7. Krauel L., Jose P., Munoz E., Gean E., et al. Van der Woude syndrome and lower lip pits treatment. J. Oral Maxillofac Surg. 2008;66:589-592.
8. Hasan S., Ishrat Khan N., Van der Woude syndrome-a case report. Recent Research in Science and Technology. 2011;3(12):53-57 50.
9. Ziai MN., Bensin AG., et al., Congenital lip pits and van der Woude syndrome. The journal of craniofacial surgery. 2008;16(5).