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RARE EXPRESSION OF VAN DER WOUDE SYNDROME: A CASE REPORT REVISITED

Kristaninta Bangun[1]*, Nurina Widayanti[2], Gentur Sudjatmiko[3]

1. *Universitas Indonesia, Department of Surgery, Division of Plastic Reconstructive and Aesthetic Surgery, RSUPN Cipto Mangunkusumo, Jakarta, Indonesia*
2. *Research Assistant of Lingkar Studi Bedah Plastik, Jakarta, Indonesia*
3. *Universitas Indonesia, Department of Surgery, Division of Plastic Reconstructive and Aesthetic Surgery, RSUPN Cipto Mangunkusumo, Jakarta, Indonesia*

ABSTRACT

Background: Van der Woude syndrome (VWS) is a rare developmental malformation, characterized by pits in the lower lip. Van der Woude syndrome is an autosomal dominant craniofacial syndrome with various expression: lower lip pits, cleft lip with or without cleft palate, syngnathia, hypoodontia, and ankyloglossia. Extra-oral abnormalities findings can be found: syndactily, corpus callosum dysgenesis, megacolon, ventricular septal defect and genital abnormality.
Methods: We reported a case of 5-month-old male with rare expression of VWS: bilateral cleft lip and palate, syngnathia, lower lip pits, ptosis of upper left eyelid and macropenis.
Results: We perform surgery to release the fibrous band to achieve satisfactory maximum mouth opening. Next we perform cheiloplasty and lower lip pit removal.
Conclusion: Proper surgical intervention in VWS patients can improve feeding and prevent further temporomandibular complications. Careful examination of patients with cleft lip and lower lip pit should be done to avoid misdiagnosis.
Keyword: *lower lip pits, bilateral cleft lip and palate, syngnathia, ptosis, Van der Woude Syndrome, macropenis*

Latar Belakang : Sindrom Van der Woude (VWS) adalah malformasi kongenital yang jarang, ditandai dengan adanya lekukan pada bibir bawah. Sindrom Van der Woude merupakan sindrom kraniofasial yg diturunkan secara autosom dominan dengan presentasi klinis: "sumur" bibir bawah, bibir sumbing dengan atau tanpa langit-langit sumbing, singnatia, hipodonsia, dan ankyloglossia. Kelainan ekstra-oral yang dapat ditemukan adalah: sindaktili, disgenesis korpus kallosum, megakolon, defek septum ventrikel, dan kelainan genitalia.
Metodologi : Kami laporkan pasien VWS laki-laki usia 5 bulan dengan presentasi klinis yang jarang: bibir sumbing dan langit-langit sumbing bilateral, singnatia, lekuk bibir bawah, ptosis palpebra superior kiri, dan makropenis.
Hasil : Kami lakukan tindakan untuk melepas jeratan fibrosa (*fibrous band*) sehingga tercapai pembukaan mulut yang baik. Selanjutnya kami lakukan labioplasti dan eksisi lekuk bibir bawah.
Kesimpulan : Intervensi bedah yang tepat pada pasien VWS dapat memperbaiki pemberian nutrisi dan mencegah komplikasi temporomandibula di masa depan. Pemeriksaan fisik yang cermat harus dilakukan pada pasien bibir sumbing dengan lekuk bibir bawah untuk menghindari misdiagnosa.
Kata Kunci : *lower lip pits, bilateral cleft lip and palate, syngnathia, ptosis, Van der Woude Syndrome, macropenis*

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INTRODUCTION

Van der Woude Syndrome (VWS) is an autosomal dominant syndrome characterized by unilateral or bilateral pits of the lower lip. The pits is an accessory salivary glands.[1-5] Another clinical appearance of VWS is very variably : cleft lip and or palate,[1,2,4,5] syngnathia,[6] syndactily,[5,8] dysgenesis of corpus callosum,[5] megacolon congenital, [5,8] and ventricular septal defect.[7] The incidence of VWS has been reported to be 1: 60.000-75.000,[9] to 1:200.000.[10] We report a case of 5-month old baby with very rare expression of Van der Woude Syndrome: bilateral cleft lip and palate, syngnathia, lower lip pits, ptosis of upper left eye lid and macropenis.

CASE REPORT

Our VWS patient is a five-month-old male, with body weight 6 kg, who came to our clinic with bilateral cleft lip and palate. The baby was born as the third child of 28-years-old mother, delivered spontaneously, aterm. Measured birth weight was 2600g with length 49 cm. The mother had never experienced serious illness during her pregnancy and had only reported one visit to the district hospital due to gastritis. There was family history reported, which was a cleft lip proband on his cousin from the father's side. The laboratory finding showed: hemoglobin 11.4mg/dl, white blood count 11,600/mmc, platelet 238,000/mmc and hematocrite 35%.

According to the diagnosis from the outpatient clinic we planned to perform labioplasty in RSUPN Dr.Cipto Mangunkusumo. At the operating room, we administered injection of 10 mg pethidine and give halothane induction by mask. When we started to perform intubation we found the fusion of maxilla and mandible due to bilateral fibrous band. The intubation failed because of narrow mouth opening (only 4 mm wide) so we decided to postpone the operation. One minute later the oxygen saturation decreased to 60% followed by bradicardy and apneu. Cardiopulmonary resuscitation was performed with an injection of 0.1 mg atropine. Few minutes later the oxygen saturation increased to 99-100%. Oxygen saturation depletion can occur after inhalation of halothane due to bronchial spasm—which can lead to hypoxia followed by bradycardia and apneu.

On examination, in addition to (1) bilateral cleft lip and palate, and (2) syngnathia, we found: (3) Bilateral lower lip pits and (4) ptosis of left upper eyelid and (5) macropenis based on the length (4.5 cm) of non-stretch penis. According to the clinical findings we diagnosed the patients with VWS and planned to perform surgical procedure to release the band, labioplasty and pits excision. The pre-operative problem was the estimation of how much and how long the fibrous band that restricted the maxilla and mandible. To anticipate that problem, we performed axial, coronal and sagittal CT-Imaging, and found multiple fibrous bands at left and right side of the mouth and oropharynx.Intra-operative, the baby was put on halothane by mask. We released the fibrous band by cutting with clamp and scissors, bleeding was minimal. We released three bands on the right side connecting alveolar ridge of maxilla to the mandible, and two on the left side connecting alveolar ridge of maxilla to the mandible and maxilla to the tongue. After all of the bands have been released, we are able to achieve 15 mm of mouth opening and the intubation with endotracheal tube could be performed. We performed labioplasty with Millard modification procedure and excision of bilateral lower lip pits. Before excising the pits, first we identified the pits using a gentian violet solution. The tracts run proximally to the gingivobuccalis sulcus, so we made a conical shape design with the external orifice of the pits as a base. We excised the epithelium of the pits and closed the defect by primary suture. Two weeks post-operatively the wound was good and the baby could open the mouth wider.

Figure 1 and 2. Clinical picture of the patient, shows bilateral cleft lip and palate, syngnathia caused by fibrous bands (arrows), bilateral lower lip pits and ptosis of left upper eyelid

Disclosure: *The authors have no financial interest to disclose.*

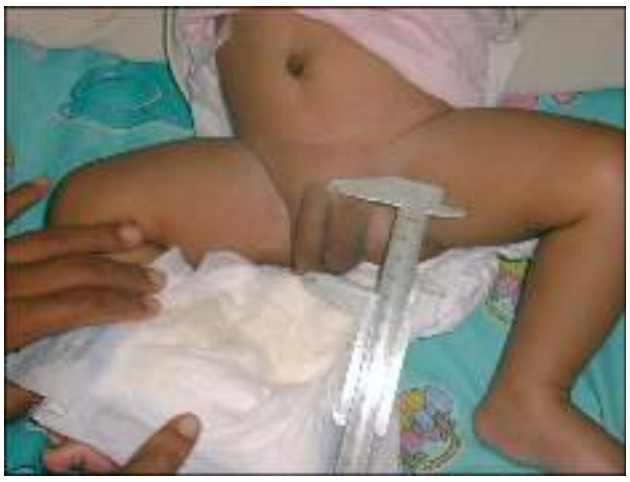

Figure 3. Length of the non- stretched penile is 4,5cm

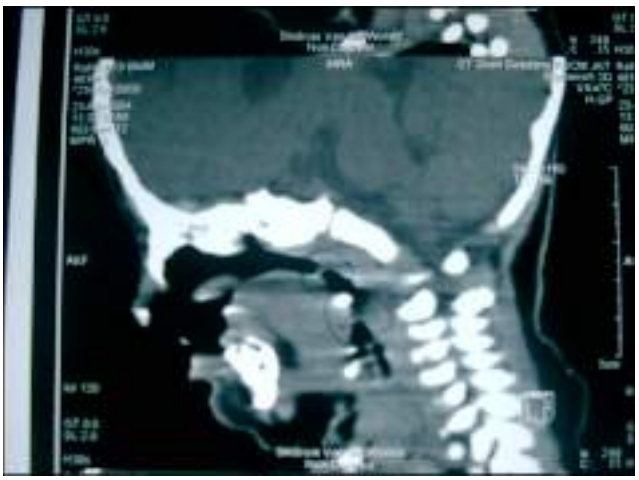

Figure 4. From CT imaging 2 fibrous band (arrows) seen at the left backside of oral cavity

DISCUSSION

Van der Woude syndrome is an entity characterized by lower lip pits and transmitted in autosomal dominant fashion.[1,4,6] The gene responsible for this disorder has been mapped to the long arm of chromosome 1 at q32-q41.[1,2,4,6] Demarquay in 1845 was probably the first to report a child with van der Woude syndrome.[3] This syndrome affects both sexes equally. This malformation is caused by a genetic defect that occurs during the embryonic stage and does not allow complete fusion of the mandibular arcade and subsequent union of the lips and palate.[4] The pathogenetic basis of the pits and depression is the incomplete reduction of the lateral wrinkles of the lower lip, which occurs between the 40th and 50th day of embryonic life. It is normally during this period that fusion of the lip and the palate occurs simultaneously.[11]

Persistence of buccopharingeal membrane is the most commonly cited cause of syngnathia.[15] The buccopharingeal membrane is derived from the cranial aspect of two linear areas in the axially differentiated embryonic disc in which the ectoderm and endoderm remain in contact and are never invaded by mesoderm. It has been suggested that oral adhesions due to fibrous band represent remnants of the buccopharingeal membrane, which normally disintegrates during the fourth week of intrauterine life.[6]

Extra facial malformation that was found in our patient is macropenis. The length of the stretched penile is 5.5 cm, normal length is 3.9 0.8 cm.[16] Other malformations like syndactily, dysgenesis of corpus callosum, megacolon congenital, and ventricular septal defect were not found. The differential diagnoses of this case were (1) lateral synechia syndrome, and (2) popliteal pterygium syndrome. Lateral synechia syndrome, reported first by Fuhrman in 1972, consist of cleft palate and alveolar synechia,[13,14] until now only 5 cases has been reported worldwide. [14] Popliteal pterygium syndrome, is a very rare case with incidence 1:300.000, consist of syngnathia, pterygium of popliteal fossa and ankyloblefaron filiforme.[12] There is speculation that some reported cases of VWS may in fact be mild variants of popliteal pterygium syndrome and that some genetic etiologic relationship exist between the two syndromes.[6]

The main intra-operative problem on this case was the difficulties to secure airway, Puvabanditsin[6], Patel [12] and Cronin [14] proposed some alternative methods: (1) orotracheal intubation under partial visibility, (2) using laryngeal mask, (3) the bands could have been released before intubation, (4) fiber optic bronchoscope- guided nasopharingeal intubation (5) tracheostomy, especially in case of synostosis or bony fusion. We prefer the third method, by cutting all of the bands immediately before intubation. We choose this method because we already know the situation of the bands according to the CT imaging.We released the band using scissors. Before cutting, we clamp the fibrous band to avoid bleeding. Verdi[13] released the band in a nursery room without anesthetic agent. Patel[12] excised the bands with cautery device and the patient was in alert condition. Cronin[14] excised the bands with sagittal incision along the length of the band, and using the remnant fibrous tissue as a flap to cover the raw edges. The result of pathologic study of the fibrous band showed fibrous tissue covered by parakeratotic complex squamous epithelium

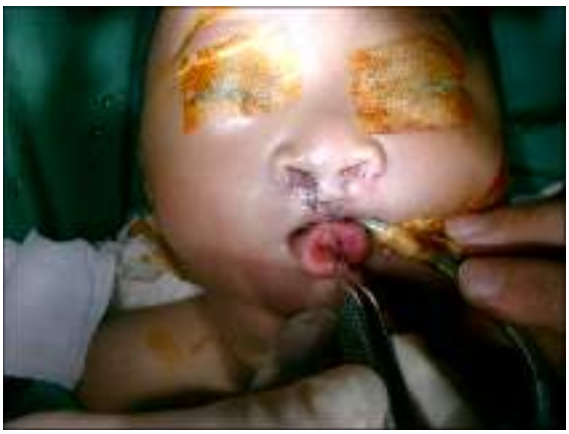

Figure 5. Bilateral pits at paramedian of the lower lip

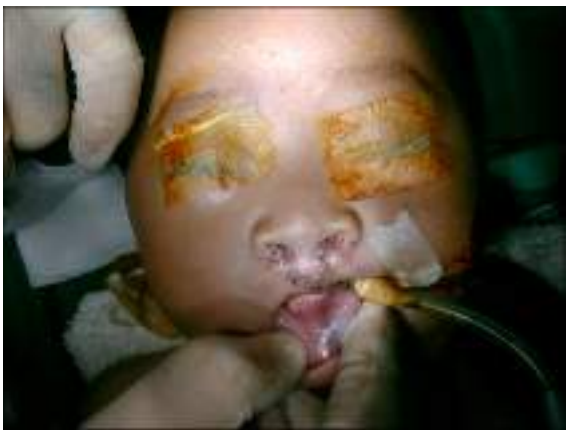

Figure 6. The tracts (arrows) run proximally to the gingivobuccalis sulcus

Figure 7. Post-operative view, note: left eyelid ptosis

Figure 8. Two weeks post operative

As the adhesion of maxilla and mandible released, satisfactory maximum mouth opening can be achieved. The intubation could be performed, followed by labioplasty, and excision of bilateral lower lip pits. In our patient, pre-surgical maximum mouth opening is only 4 mm with post-surgical mouth opening improvement (15 mm). This narrow mouth opening causes feeding difficulties. Prolonged delay in releasing of the fibrous band may result in temporomandibular complications, such as temporomandibular joint ankylosis.[14]

Ptosis correction was postponed until the patient reaches 4-years-old. This will provide time for the levator muscle to be fully developed. Further consultation to the ophthalmologist is needed to assess pupillary axis and discuss strategies to prevent amblyopia.[17] At last, careful examination of patient with lower lip pits with cleft lip or palate may prevent delayed diagnosis, so delayed treatment in this case can be avoided.

DISCUSSION

Van der Woude Syndrome is a rare autosomal dominant congenital malformation. Meticulous physical examination should be carried in order to identify patient with VWS. Careful examination of patients with cleft lip and lower lip pit should be done to avoid misdiagnosis that can delay treatment. Proper surgical intervention on VWS patient can improve feeding and prevent further temporomandibular complication.

Corresponding author :
Kristaninta Bangun
kristaninta@yahoo.com

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