Congenital double lip pits : A Case Report

Retno Widayanti*, Melita Sylvyana**, Endang Syamsudin*, Rizki Diposarosa***

*Departement of Oral and Maxillofacial Surgery, Fakulty Dentistry Universitas Padjadjaran, Bandung, Indonesia
**Departement of Oral and Maxillofacial Surgery, RSUP Dr. Hasan Sadikin, Bandung, Indonesia
***Departement Pediatry Surgery, SMF Bedah, RSUP Dr. Hasan Sadikin, Bandung, Indonesia

ABSTRACT

Introduction : Lip pits are unusual congenital anomalies affecting the lip, first described by DeMurquay in 1845. Lip pits can encounter aesthetic problem thus the patient ask for surgical overcome. Case Report : A 4-year-old girl patient came with double lower lip pits that present since birth with bilateral cleft lip and palate. The pits made a mucous accumulation occurs during mealtimes and crying, and felt aesthethic discomfort. The surgery was performed by simple excision combined with split-lip advancement technique under general anaesthesia and the excised pits was then analized for histopatological structures. The patient has no aesthetic defect after surgery. Discussion : Congenital lip pits are developmental anomalies that occur as an isolated defect or either in association with other developmental disturbances. It happens due to notching of lip at an early stage of development with fixation of tissues of the base of the notch or from a failure of complete union of embryonic lateral sulci of the lip. Lip pits can be shallow or deep, and may be associated with accessory salivary glands. The treatment is usually surgical excision with removal of entire fistulous tract. Conclusion : Surgical removal of lip pits is commonly for cosmetic purpose. It must be treated wisely because lips are essential part of someones face.

Keywords : *congenital anomalies, lip pits, excision*

Correspondence author: Retno W, Department of Oral and Maxil Surgery Faculty Dentistry Universitas Padjadjaran, Jl. Sekeloa Selatan No. 1 Bandung, Tel./Fax: +6222-2504985/2532805 Email: retnowidayanti99@yahoo.co.id

ABSTRAK

Pendahuluan: Lip pit merupakan anomali kongenital pada bibir yang jarang terjadi, pertama kali didefinisikan oleh DeMurquay pada tahun 1845. Lip pit dapat menyebabkan permasalahan estetik dan menyebabkan pasien datang untuk diatasi dengan pembedahan. Obyektif : Memahami patogenesis dan managemen lip pit kongenital. Laporan Kasus : Pasien anak usia 4tahun datang dengan keluhan terdapat dua lekukan pada bibir bawah, yang sejak lahir telah ada bersamaan dengan celah bibir dan langit-langit bilateral. Dilakukan pembedahan eksisi sederhana dikombinasi dengan tehnik split-lip advancement dalam anestesi umum. Lip pit yang dieksisi kemudian dianalisis struktur histopatologisnya. Pasien kontrol dengan tanpa defek estetik pasca pembedahan. Diskusi: Lip pit kongenital merupakan anomali perkembangan yang terjadi sebagai defek tunggal atau dapat merupakan defek yang berhubungan dengan kelainan perkembangan lain. Lip pit terjadi karena deformitas bibir pada awal tahap perkembangan dengan fiksasi dasar jaringan bibir atau dari kegagalan penyatuan sulkus lateral bibir masa embrio. Lip pit dapat dangkal atau dalam, dan dapat berhubungan dengan kelenjar saliva. Penatalaksanaan bedahnya adalah eksisi dengan mengambil seluruh traktus fistula. Kesimpulan : Pengangkatan lip pit secara bedah umumnya untuk tujuan kosmetik dan harus ditatalaksana dengan baik karena bibir adalah bagian penting wajah seseorang.

***Kata kunci** : anomali kongenital, lip pit, eksisi*

INTRODUCTION

Lip pit is an indentation and/or sinus tract in the lower lip that is usually located to one or both sides of the lip midline.[1] Lip pits are unusual congenital anomalies affecting the lips, can occur in the region of upper lip, lower lip, or the oral commissure.[2,3] Congenital lip pits or congenital fistulas or paramedian sinus or humps or labial cysts are rare congenital invaginations of the lower lip.[3-6]

Lip pits are first described by DeMurquay in 1845.[2,3,6] Lip pits are developmental anomalies that occur either in association with other developmental disturbance or as an isolated defect.[2,4-6] This minimally deforming anomaly is remarkable chiefly for its association with facial clefts.[4] The association of lip pits with cleft lip and palate was given by Van der Woude in 1954, and called Van der Woude syndrome.[5,6] Two-third of congenital lip pits are associated with cleft lip or palate and the other one-third have minimal findings such as hypodontia, or isolated lower lip pits.[2]

Lip pit is more common among females.[5,6] The clinical manifestation of lower lip pits covers a wide spectrum.[6] These include slight depressions on the vermilion border of the lip, and fistulas that penetrate into subjacent salivary glands and drain small amounts of saliva.[6]

Lip pits has not been carried out for its survey among the general population; hence the frequency of this rare anomaly has been only estimated roughly from its incidence in hospital records.[7] Assuming that 70% to 80% of patients with pits of the lower lip have associated cleft lip and/or palate, and that the frequency of clefts is 1:650 (one in every 650 births), it can be estimated that the frequency of lip pits among the general population is about 1:75,000-1:100,000 (one in every 75,000 to 100,000 births).[7]

In this case report we will discuss a 4 year old girl with double lower lip pits, that present since birth with bilateral cleft lip and palate. The patient came to Oral and Maxillofacial Surgery Department because of cosmetic reason.

CASE REPORT

A 4-year-old girl patient came to Oral and Maxillofacial Surgery Departement with her parents in January 2015, with two pits on her lower lip. The lip pits were present since birth with bilateral cleft lip and palate. Her double lip pits made the patient uncomfortable appearance because of unusual shaped of her lower lip. The pits was also made a liquid accumulation occurs during mealtimes, or in relation to crying. History

of past surgery was repaired of her lips due to bilateral cleft lip in August 2010 when she was 3 months old, followed by repaired of her bilateral palatochizis with palatoplasty when she was 1,5 year old. History of family with cleft lip, cleft palate, and/or lip pits was denied. History of routine followed up by medical practitioner when her mother was pregnant was admitted, and there was no history of trauma in the first 3 months of her mother pregnancy, no history of taken medicines or medicinal herbs during pregnancy. The patient was spontaneously born in medical practitioners with fully term of pregnancy. The patient has two siblings without any abnormality, and the patient was the youngest.

Examination of general physical status found within normal limit. From local status extra oral examination, there was symmetrical face, found with scar mark post bilateral labioplasty in her upper lip. In her lower lip, there was double pits revealed bilateral paramedian pits, appeared wet but without pain and swelling (Figure 1).

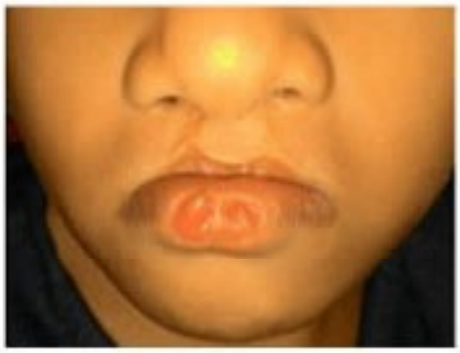

Figure 1. Double lower lip pits.

From intra oral examination, there was scar post palatoplasty in her palate, ten deciduous teeth in her lower jaw, and eight deciduous teeth without lateral incicors with cleft on her lateral incicors of upper gingival, in her upper jaw. The tongue, floor of the mouth, vestibulae, buccal mucosa and tonsils was found no abnormality. As the defects of the patient lower lips were present at the time of her birth, it was diagnosed as a congenital lip pits. The surgery was planned excision under general anaesthesia, and the patient was prepared with laboratory examination, thorax x-ray and was consulted to Paediatric Department for cardiopulmonary examination and to Anaeshtesia Department for preoperative and perioperative treatment. There was no contraindication found for her treatment under general anaesthesia.

The surgery was performed in the end of February 2015. The size of the double lip pits was measured and was found 0,5x0,8x1,2cm and 0,5x0,3x1,2cm (Figure 2). On applying pressure, watery secretions was expressed from the opening pits. The pits was then marked for its excision pattern (Figure 2). The lip was injected with adrenalin, then excision was done carefully by saparating the pit from the muscle. During the dissection, special attention was paid to the lip muscle so as not to cause a whistling deformity later. In the deepest part of the one sinus we found the base is very thin line of labial mucous so its easily break the intraoral site. The track of the pits or sinus should all excised to prevent retention cyst. The defect than reconstructed by suturing the lip muscle carefully to get as ideal as possible for its good esthetics outcome with vicryl 5-0 after careful control of bleeding (Figure 4). The excised pits was then analized for its histopatological structures to the Anatomy Pathology Departement.

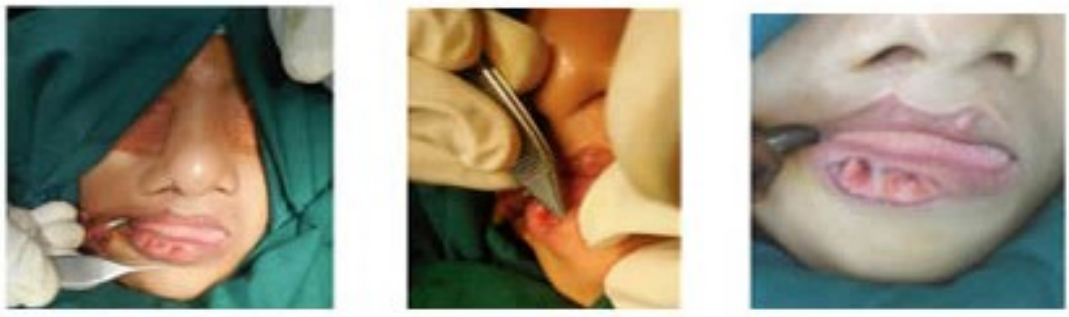

Figure 2. The depth of the double lip pits was measured and incision pattern was marked.

Patient was managed by given amoxycillin 250mg and analgetic suppositoria after surgery. One day after surgery her lower lip looked minimum oedematous swelling, no bleeding, and patient did not feel pain on her lips intra oral and extra orally. Seventh day after surgery the suture was removed and two weeks after surgery the patient came with histopatological result. The lips did not have aesthetic defect after and seems symmetrical without loss of the lip muscle or a whistling deformity (Figure 6).

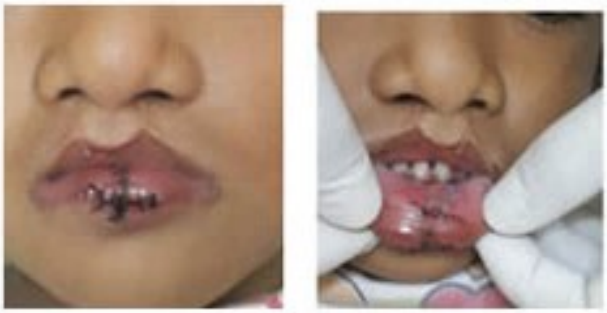

Figure 5. First day after surgery.

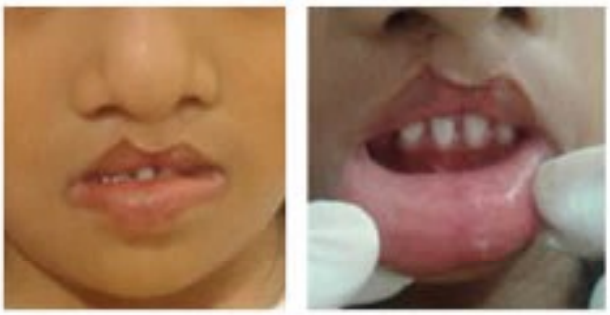

Figure 6. Two weeks after surgery, the lower lip had no defect.

The excised pit histopatology structures was macroscopicly a skinned tissues size 1,2x1x0,5cm and 1,2x0,7x0,5cm, brownish white and elastic. Microscopicly appeared hyperplastic stratified squamous epithelium with normal nucleus. There was a salivary gland duct, fibrocolagenous connective tissue stroma in subephitelial region that hyalinated with lymphosit inflammatory cell. Inside it appeared salivary gland with stratified thoracic epithelial with normal nucleus. There was no malignancies cells appeared, and the histopatologically was concluded as hyperplastic squamous cell with fistulous salivary gland duct in inferior lip.

DISCUSSION

Congenital lip pits are developmental defects that occur on the paramedian portion of the vermilion border of the lower lip.[2] Pits of the lower lip (fistulas of lower lip, paramedian sinuses of lower lip, humps of lower lip, labial cysts) is a very rare congenital malformation.[4] The first case of lip pits was reported and discribed by DeMurquay in 1845.[2-10]

The clinical picture of lip pits may vary ranging from a single pit in the centre of the lip to two pits (one on the right and one on the left) or one pit on either the right side or left side. Their occurrence can be on the inner lip surface, outer lip surface or on the margin between the inner and outer lip.[2-5,7]

Most of the double lip pits are located in the lip vermilion and on the muco-cutaneous line at a distance of about 5-25 mm from each other, and often enlarged and swollen.[5,10] Their appearance can range from subtle depressions to prominent humps, may be shallow or deep, varying from asymptomatic slight depression on vermilion border to pits that form canals ranging in length from 1- 25 mm, which generally extend into the orbicularis oris muscle.[2,3,5-7, 10]

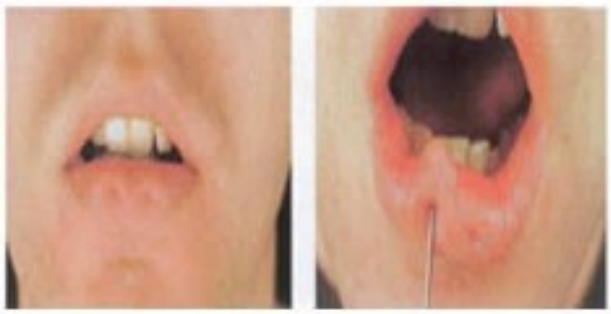

Figure 7 : Congenital lower lip pits appears enlarged and swollen and measured on probing 11mm deep fistula. Quoted from : Dissemond, Haberer, Franckson & Hillen.[10]

Lip pits are usually circular or oval shaped, but some have also been described as transverse, slit-like, or sulci shaped.[6] On occasion, lip pits may be located at the apex of nipple-like elevation. Rarely, the elevations may fuse in the midline, producing a snout like structure.[5] Lower lip pits are usually asymptomatic.[2,7] The only symptom might be the continuous or intermittent drainage of water

or salivary secretions, because the lip pit maybe associated with minor salivary gland.[3,5,7,9,10] The mucous accumulation occurs more rapidly before and during mealtimes, or in relation to crying.[5]

The association of cleft lip and palate with lip pits was given by Van der Woude in 1954. When lip pits occur in association with cleft lip/palate the condition is referred as Van der Woude Syndrome (VWS). [2-6,8-11] The patient in our case report seems to have the condition of VWS since her parents admitted that the cleft lip, cleft palate and the double lip pits appeared since the girl was born. Other malformations associated with lip pits are syndactyly of hands and cleft lip and palate; mental retardation and cleft, type not specified; ankyloglossia and cleft lip and palate; polythelia; symblepharon and cleft lip and palate; and ankyloblepharon, adhesion between maxilla and mandible, and cleft uvula. [4,7,11]

Various syndromes associated with lip pits beside VWS are popliteal pterygium syndrome, oral facial digital syndrome and Marres and Cremer's syndrome.[2,11] Hypodontia and lower lip pits are also seen in the Kabuki make-up syndrome.[11] Patients often perceive these pits as depressions made by the maxillary central incisors, even the pits are present from birth, much before maxillary incisors erupt.[2]

Congenital pits of the lower lip seem to run stronger in family. Gurney (1940) reports four cases of lip pits in one family. Fogh-Andersen (1943) reports eleven cases of lip pits in three family groups; and Test and Falls (1947) reports lip pits in five generations of the same family. Vander Woude (1954) found, after careful study of five pedigrees, that the combination of pits of the lower lip with cleft lip and palate is based on a single dominant gene. In 1987 Bocian et al. reported a patient with lip pits and a deletion in 1q32-q41, and subsequently Murray et al. found linkage between VWS and markers from the same region. Microdeletions in 1q32-q41 have also been reported in families with Vander Woude.[5,11] However, chromosome 1p34 was mapped as the second locus for the VWS.[3,5,9,10]

Recently, mutations have been found in the interferon regulating factor 6 gene in patients with VWS and popliteal pterygium syndrome. However, the lack of 100% concordance in monozygotic twins suggests that genetic events alone are not responsible for the clefting phenotype. The process of monozygotic twinning is in a sense a teratogenic event, and monozygotic twins have an increased incidence of (often discordant) structured malformations.[5]

The fact that clefts occur with lip pits seem happening stronger in families than clefts without lip pits, has attracted the attention of professionals dealing with cleft patients.[7,8] Some cases of single lip pit have occurred in families with members with double lower-lip pits. It can be assumed that a single pit is not a distinct entity but rather an incomplete expression of the trait. On the other hand, the rarely described fistulas of the upper lip have not shown any inheritance pattern.[7,11]

The origin of lip pits is related to failure of complete union on the embryonic mandibular arch and can result due to notching of lip at an early stage of development with fixation of tissues of the base of the notch.[3-5,9] These sulci normally disappear by 6 weeks of embryonic age.[3] Warbrick et al. explained that the sulci gradually become obliterated except at the cephalic end. These furrows become deeper as growth proceeds, thus establishing canals that are gradually incorporated in the substance of the lower lip and, persisting, become congenital labial fistulas.[4]

At 5.5 weeks during the developmental stage of the head and neck, the fusion of the mandibular arch and sulcus lateralis of the lower lip occurs, while the fusion of the maxillary and frontonasal processes come about at 6 weeks. It is hypothesized that a common event may simultaneously disturb fusion in both locations. This event results in the strong association between the lip pits and cleft lip or palate.[2]

Lip pits are among the rarest congenital deformities recorded. Congenital lower lip sinuses have been reported in about 0.001% of the population, and 65% to 75% of the cases are associated with cleft lip and palate.[5] The prevalence of VWS varies from 1:100,000 to 1:40,000 still born or live births. No significant difference between sex is reported as regards to the prevalence of the syndrome.[5]

The first histological examination of lip pits was performed by Madelung in 1888. Since then his findings have been confirmed by many other authors.[4] Microscopic examination of a paramedian lip pit shows a tract that is lined by stratified squamous epithelium. Minor salivary

glands may communicate with the sinus. A chronic inflammatory cell infiltrate often is noted in the surrounding connective tissue.[3] It seems that the histological finding in this case report was equal to the literature. The lip pits may require no treatment if they are mild.[1] If necessary, the labial pits may be excised for cosmetic reasons.[2,3,6,11] The relatively high incidence in females has been attributed to the established fact that they are more likely to seek surgical intervention for cosmetic considerations, like occurred in this patient case report. The goals of treatment are removal of sinuses and providing cosmetic relieve for the disfigurement.[6]

Surgical excision is especially indicated in patients with resulting recurrent inflammation.[10] Electrocoagulation techniques and marsupialization of the sinuses into the oral cavity have been discontinued because of high complication rates and poor results.[5,11] The simple excision of the sinuses and adjacent glands that empty into the sinuses is the most commonly accepted procedure. Transverse elliptical excision is a well-known and accepted method for treatment of congenital sinuses of the lower lip. Several authors emphasized that simple excision and transverse elliptical excision had inferior results, such as lower lip muscle loosening and whistling deformity.[5,11]

Another surgical approach to achieve radical excision is the split-lip advancement technique. This technique was described by Mutaf et al and has attempted of the dysplastic tissue and repair of lip muscles to restore good lip functionality and bilabial symmetry.[11] In this technique, two opposite labial artery-based flaps, including the whole thickness of the vermillion and the mucosal surface of the lip, are used to repair the median defect that results from excision of the sinuses.[5]

CONCLUSION

Double lip pits is a rare case. The excision surgery that performed in this case combined with split-lip advancement technique had a satisfying outcome to the patient and her parents. To manage lip pits, evaluate the outcome of surgical intervention, and to review some literatures for its etiologies and pathogenesis are important because lips is one of aesthetic issue. The most significant problems that was not performed in this case are related to associated congenital anomalies, such as cleft lip and/or cleft palate, and the potential for transmission of the trait to subsequent generations. The identification of familial lip pits is crucial for genetic counseling.

REFERENCES

1. Dixon MJ, Marazita ML, Beat TH, Murra JC. Cleft lip and palate: Understanding Genetic and environmental Influences. Macmillan Publisher Limited. 2011;3(12):167-78.
2. Shakil M, Mohtesham I, Jose M, Prabhu V. Non Syndromic Bilateral Congenital Lip Pits: A Case Report. Asian Pac. J Health Sci 2014;1(3):197-9.
3. Sapp JP, Eversole LR, Wysocki GP. Contemporary oral and maxillofacial pathology. 2nd ed. St. Louis Missouri: Mosby. 2002. p. 27-8.
4. Cervenka J, Gorlin RJ, Anderson VE. The syndrome of pits of the lower lip and cleft lip and/or palate: Genetic consideration. U.S. Public health service program grant in oral pathology. Am J Hum Gen 1966. p. 416-32.
5. Prakash R, Verma S, Singh U. Agarwal N. Congenital lip pits: Report of cases with variable expressivity. ejournal of dentistry. 2013;3(3):453-6.
6. Ibrahim A and Ajike S. Congenital symmetrical lower lip pits: Van der Woude Syndrome. Oman Med J 2015;10:5001.
7. Berkowitz S. Cleft lip and palate. 2nd ed. Germany: Springer. 2006. p. 201-13.
8. Kummer AW. Cleft palate and craniofacial anomalies. 3rd ed. United States of America: Cengage Learning. 2014. p. 84,122,360,730.
9. Arangannal, Muthu, Nirmal. Vander Woude Syndrome: A case report. J Indian Soc Pedo Prev Dent. 2002;20(3):102-3.
10. Dissemond J, Haberer D, Franckson T, Hillen U. The van der woude syndrome: a case report and review of the literature. European Academy of Dermatolog and Venereology JEADV. 2004;18;611-613.
11. Guner U, Celik N, Ozek C, Cagdas A. Van der Woude Syndrome. Scand J Plast Reconstr Surg Hand Surg 2002;36:103-5.
12. Gupta R, Kaul B, Kotwal B, Mahajan N. The syndrome of pit of the lower lip and its association with cleft palate in contemporary clinical dentistry. 2014;5(3):383-5.