Case Report

Non-syndromic bilateral lower lip pits: A case report and literature review

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ABSTRACT

This case report describes a rare occurrence of non-syndromic lower lip pits. The 16-year-old female patient had bilateral pits on the lower lip vermilion, without any other associated abnormalities, which sets this case apart from the syndromic lip pits seen in Van der Woude syndrome. The report outlines the clinical features, diagnostic evaluation, and chosen treatment approach, emphasizing the importance of recognizing and distinguishing isolated lip pits from syndromic presentations.

Key words: Congenital, Lip anomalies, Lower lip pits, Non-syndromic

Congenital lower lip pits are very uncommon, occurring in only about 0.001% of the general population [1]. Non-syndromic lower lip pits are even rarer, and the exact prevalence is not known, as limited literature is available on this specific condition [2]. Non-syndromic lower lip pits are a rare congenital anomaly characterized by depressions on the vermilion border of the lower lip, typically without any other associated abnormalities [1-3]. This condition, though often benign, can present management challenges due to potential cosmetic concerns and occasional complications, such as infection or salivary drainage.

This case report contributes to the limited literature on this rare condition and will explore the clinical presentation, diagnosis, and management of non-syndromic lower lip pits, highlighting key differences from syndromic lip pits and emphasizing the importance of a thorough evaluation and appropriate intervention.

CASE REPORT

A 16-year-old female student was presented by her parents due to the unsightly appearance of her lower lip (Fig. 1). The patient reported feeling depressed about the appearance of her lips and had been subjected to bullying by others as a result. The patient had one younger brother. The patient was born to non-consanguineous parents and no other family members were reported to have similar conditions or any other identifiable congenital abnormalities.

The patient was well nourished and proportionate for her age. Her intelligence was within normal limits, and she exhibited less sociable behavior. General examination findings included a pulse rate of 82 beats/min and a blood pressure of 110/70 mmHg. On examination, there were two horizontally ovoid pits with nipple-like projections in the center measuring 1 × 0.5 cm and 0.7 × 0.5 cm in size on the right and left sides of the lower lip, respectively. These pits were located on either side of the midline, along the vermilion border of the lower lip. The inferior rims of the pits were continuous with the white line of the lower lip. Saliva was observed secreting from the pits. The overall size of the lower lip was normally developed and proportional to the upper lip. The upper lip and commissure were also normally developed. The patient's teeth were normally erupted in number for her age, though the maxillary canine teeth were smaller. There was no cleft lip or cleft palate, and the uvula was normal. The tongue was normally developed, and there was no ankyloglossia. The eyes were normal, and there was no hypertelorism or pterygium. The nose, chin, and digits were all normally developed, with no popliteal webbing, polydactyly, or syndactyly. The spine was also normal. The patient had attained menarche, and her genitalia were normally developed.

Diagnostic imaging studies, including a computed tomography scan of the face, ultrasonography of the abdomen, and echocardiography, were conducted and found to be within normal limits (Fig. 2). In addition, routine blood investigations yielded normal results. Given the absence of any other associated congenital anomalies, the patient's parents were reluctant to pursue genetic studies.

The patient underwent surgical excision of the lower lip pits under general anesthesia (Fig. 3a). An elliptical excision was planned as there were no other structural abnormalities or

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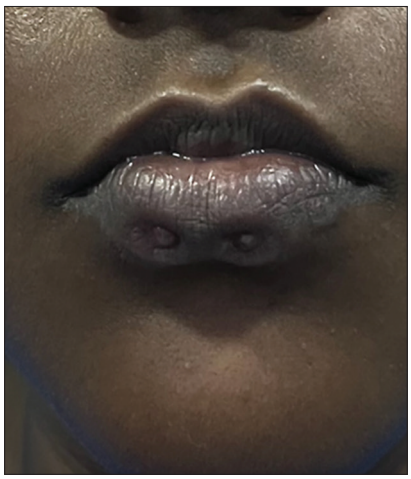

Figure 1: Clinical picture of the patient

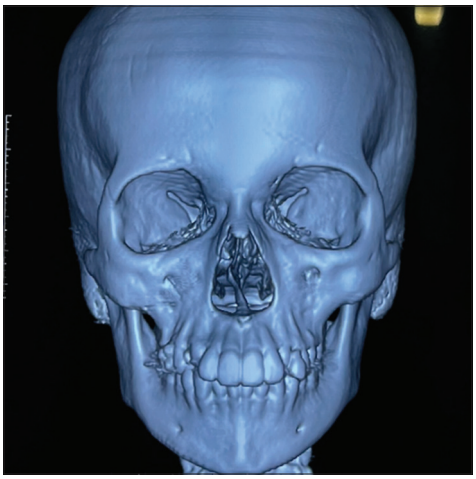

Figure 2: Computed tomography scan of face

additional pits present (Fig. 3b). The pits were excised in their entirety and sent for histopathological examination (Fig. 3c). The resulting defects were closed in layers (Fig. 3d). The final appearance of the lower lip showed no significant alteration in shape and structure. The post-operative period was uneventful, and the patient was followed up for 6 months. Both the patient and her parents expressed satisfaction with the final outcome. The patient reported an improvement in her self-esteem following the procedure.

Histopathological analysis revealed that the margins of the pits were lined with non-pigmented squamous epithelium with irregularly arranged bundles of muscle fibers observed surrounding the pits, and multiple minor salivary glands were found to be draining into the pits (Fig. 4).

DISCUSSION

Non-syndromic lower lip pits typically present as bilateral depressions or pits situated paramedian to the midline in the vermilion border of the lower lip [1-6]. Unilateral or midline presentations are also possible. These pits can vary in size and depth. In one case, the pits are "doughnut-shaped," while other presentations can include a circular or transverse slit and

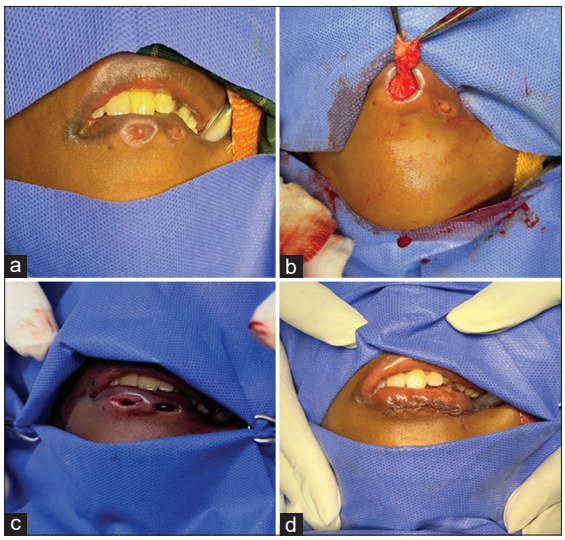

Figure 3: (a) Surgery under general anesthesia; (b) excision of pits; (c) excision defects; (d) closure of defects

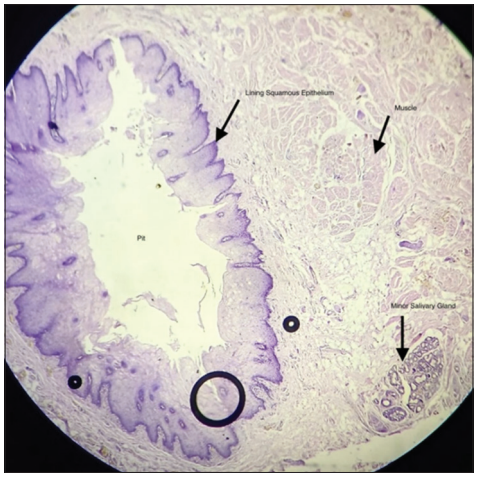

Figure 4: Histopathological examination

may be associated with nipple-like elevations [1-3]. While some cases are asymptomatic, others involve sinus tracts that may secrete saliva, potentially leading to embarrassment or infection. However, in some instances, there are no sinus tracts or associated discharge[1,2]. Non-syndromic lower lip pits exist as an isolated anomaly, without any other associated features. The exact prevalence of non-syndromic lower lip pits is difficult to determine due to its rarity and underreporting [2,6].

Lip pits can be associated with several syndromes [7-13]. Van der Woude syndrome is the most common form of syndromic orofacial clefting [10,14]. It is characterized by lower lip pits, which are depressions on the lower lip, sometimes moist due to salivary glands in the pits. Cleft lip and/or cleft palate, hypodontia, and, rarely, limb deformities, can also be associated with Van der Woude syndrome [10,14]. This has an autosomal dominant pattern transmission [10].

Popliteal Pterygium syndrome is a rare, inherited autosomal dominant condition characterized by anomalies affecting the face, limbs, skin, and genitalia. The clinical manifestations are highly variable. The most striking feature is the popliteal pterygium,

a web of skin behind the knee [8,12]. Other common features include cleft lip and/or palate, lower lip pits, syndactyly, and genital anomalies.

Oral facial digital syndromes are a group of genetic disorders affecting the development of the oral cavity, facial features, and digits [7,11]. The features of the various types overlap significantly, and the classification system continues to evolve. Some common components include cleft lip and/or palate, lip pits, tongue abnormalities (e.g., lobulated or bifid tongue), dental abnormalities (e.g., missing teeth), and excessive oral frenula [7]. Facial asymmetry, hypertelorism, and hypoplasia of the nasal cartilages, polydactyly, syndactyly, and brachydactyly [11]. Neurological problems, brain structure changes, bone abnormalities, vision loss, and heart defects can also be present [7].

Marres Cremer syndrome is a rare condition associated with lower lip fistulas [9]. The most notable feature is the presence of congenital conductive or mixed hearing loss, pre-auricular sinuses, external ear anomalies, and commissural lip pits [9]. The syndrome follows an autosomal dominant inheritance pattern. Marres-Cremer syndrome is distinct from branchiogenic syndromes due to the absence of cervical cysts or fistulae [9].

The exact etiology of non-syndromic isolated lower lip pits remains unclear. Van der Woude Syndrome is known to be caused by mutations in the IRF6 gene located on chromosome 1q32-q41 [6]. This gene plays a crucial role in the development of the face and palate. Most cases of lip pits are linked to chromosome 1q32-q41 and a second locus at 1p34, with about 30–50% arising from *de novo* mutations. Several theories focus on disturbances during embryonic development [5]. One prominent theory suggests that lower lip pits result from incomplete fusion of the mandibular processes during early facial development [5]. This disrupted fusion may involve the lateral sulci of the lip, leading to the formation of pits, suggesting a common event disrupting fusion in both mandibular and maxillary processes, which explains the frequent association of lip pits with cleft lip/palate in Van der Woude syndrome [1,5]. Another theory proposes that early notching of the lip during development, is followed by fixation of tissues at the base of the notch [5]. Less supported theories include abnormal invagination of the lip mucosa, intrauterine disease of labial glands, amniotic adhesions, and the presence of epithelial pearls [5]. None of these adequately explain the occurrence of lower lip pits and are likely to arise from a complex interplay of genetic and developmental factors.

Diagnosis is primarily based on clinical examination by the presence of characteristic lip pits without any other associated systemic anomalies. A thorough family history is essential to assess for a potential genetic predisposition, although many cases arise as *de novo* mutations. It is important to differentiate these pits from commissural lip pits, which occur at the corners of the mouth [15].

Treatment for non-syndromic isolated lower lip pits is primarily for cosmetic reasons or to address complications, such as recurrent infections or persistent salivary drainage. If the pits are asymptomatic and the patient is not bothered by their appearance, no active treatment is necessary [1,2]. Surgical excision remains the mainstay of treatment [16]. The choice of surgical technique depends on individual factors such as pit size, depth, and associated tract involvement. The difficulty index helps to assess the complexity of cases, which guides surgical approaches and predicts outcomes (Table 1). The difficulty index is scored as follows: 2 indicates an easy case, 3 indicates a moderately difficult case, and 4 indicates a difficult case [14].

Table 1: Difficulty index

Score	Involvement of white skin roll	Presurgical depth of lip pit
1	Uninvolved	Shallow (#6 mm)
2	Involved	Deep (>6 mm)

Simple excision involves direct removal of the pit and underlying sinus tract. It carries a higher risk of recurrence and mucocele formation due to the potential for incomplete removal of the sinus tract [16]. The vertical wedge excision or inverted T lip reduction techniques offer better results and lower recurrence rates compared to simple excision [16]. A wedge of tissue encompassing the pit and tract is removed, reshaping the lip. The inverted-T lip reduction allows for tissue rearrangement to minimize scarring and achieve more esthetically pleasing results. These techniques offer the best results because the scar remains hidden at the red line of the vermilion [14]. The Mutaf-Goldstein technique involves injecting methylene blue dye along with bacitracin ointment can be used to clearly delineate the tract during surgery, minimizing the chance of incomplete removal. The entire stained tract is then excised. Modified simple excision techniques of excising both pits in a single elliptical pattern have been developed to decrease recurrence risk and may be considered if other techniques are deemed unsuitable. Whatever the technique used, it is of utmost importance to bring about complete excision of the tracts to prevent recurrence or development of mucous cysts [14].

Post-operatively, patients are advised on oral hygiene practices. Genetic counseling is recommended, especially in familial cases or if there is a history of cleft lip or palate in the family [1]. Psychological counseling is beneficial, particularly if the patient experiences emotional distress.

CONCLUSION

While primarily a cosmetic concern, non-syndromic lower lip pits are a rare unique clinical condition, careful evaluation is needed to differentiate it from Van der Woude syndrome and other syndromes. Surgical intervention remains the primary treatment for symptomatic or cosmetically concerning cases. Further research on the genetic and environmental factors influencing non-syndromic lower lip pit formation will undoubtedly refine our understanding and management of this rare anomaly.

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