A rare nonsyndromic presentation of bilateral doughnut shaped lip pits in an Indian child

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ABSTRACT

Lip pits are a rare congenital anomaly that presents on the upper or lower lip or the commissure of the lips. Lip pits are an autosomal dominant trait occurring almost always in association with cleft lip or palate. They most commonly occur in association with developmental disturbances such as Van der Woude’s syndrome, popliteal pterygium syndrome, oro-facial-digital syndrome, Marres-Cremers syndrome, and Hirschsprung disease. Its occurrence in nonsyndromic individuals is extremely rare with only a handful of cases reported. The identification of lip pits with other associated anomalies is crucial for genetic counseling; we report a case of nonsyndromic presentation of bilateral lip pits.

Key words: Cleft lip, cleft palate, congenital, lip anomalies, pit on lower lip

CASE REPORT

A 9-year-old male patient reported to us with a complaint of swellings on his lower lip. History revealed that the swellings were present since birth, was initially small and increased proportionally to the size of the lip. His medical history was insignificant, and family history revealed that his parents had consanguineous marriage. There was no history of any pernicious oral habits. On general examination, he was mentally sound; there was no skeletal abnormalities, cardiovascular anomalies, genital and nail anomalies. On extraoral examination, there were two symmetrical, doughnuts shaped swelling noted on either side of midline of the lower lip. The periphery of the swellings merged with surrounding mucosa and rose to dip in the center to form of depression, hence resembling a doughnut. The swellings roughly measured about 0.5 cm in size and the color was similar to that of adjacent mucosa. There was no evidence of sinus, fistula, or any discharge from it [Figure 1]. Palpation revealed the swellings to be nontender, soft in consistency with no evidence of discharge on pressure [Figure 2]. Further examination revealed the absence of cleft lip/palate or any other anomalies. Correlating the history and clinical findings a diagnosis of an isolated case of nonsyndromic congenital lower lip pits was made. The patient and his parents were educated about the condition, and surgical management was suggested for cosmetic reasons. Patient’s father was not

Lip pits were first described by DeMurquay in 1845. Among the cases reported since then, two-third occur in association with cleft lip or palate and the remaining one-third present with minimal findings such as hypodontia or as isolated lower lip pits, the same as in this case.[1] The etiology of lip pits in either sporadic or syndrome associated presentations is not clearly known, however, it is generally believed that lip pits can result due to notching of lip with fixation of tissues at the base of the notch or due to failure of complete union of embryonic lateral sulci of the lip.[2] The presentation of sporadic lip pits is extremely rare so existing literatures does not state a specific prevalence rate, but in syndromes like Van der Woude syndrome (VWS) syndrome, the prevalence rates varies from 1:40,000 to 1:100,000 stillborn or live births.[3] Although it is an indicator of certain syndromes, it can also pose a cosmetic defect with associated psychological consequences.

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willing for surgical intervention. The patient was cautioned about the consequences of possible infection or salivation from lower lip due to communication with a minor salivary gland. Follow-up is being performed every 6 months.

DISCUSSION

In the intrauterine developmental stage of the head and neck, the fusion of mandibular arch and sulcus lateralis occurs at 5.5 weeks, whereas the fusion of the maxillary and frontonasal processes occurs during the 6th week. A common event may simultaneously disturb fusion in both locations. This results in the strong association between the lip pits and cleft lip or palate in 21% of patients. Most commonly lip pits occur due to microdeletion on chromosome bands 1q 32-q41.10. Lip pit is a congenital anomaly, usually bilateral, often symmetrical, on the vermilion portion, or in mucocutaneous line of the lip. Congenital lip pits may be classified according to their location as Commissural, midline upper lip, and lower lip. Lower lip pits are also called as fistula labii inferioris, labial humps, labial cysts, labial fistulae, and paramedian sinuses of the lower lip. It can occur as an isolated defect or in association with a syndromic condition.

The surface of the lip pit may present as a circular or transverse slit or be located at the apex of nipple-like elevations. The lip pits measure up to 3 mm in diameter and are usually 1–25 mm in depth. Lip pits commonly have sinus tracts which extend into the orbicularis oris muscle. The congenital lip pit sinus tracts are reported in 0.001% of the population and 65–75% of the cases are associated with cleft lip and palate. In 10–14th month of intrauterine life, during the development of face, failure in closure of evanescent sulci leads to fistulae formation in lip pit patients. With the help of fistulography it has been estimated that these sinus tracts can be long and bifurcated and has a total length of 5–6 cm. Accessory salivary glands usually drain into these tracts and due to this saliva is present as a mucous secretion from these lip pits, which causes embarrassment to the patient. Syndromes that are usually associated with lip pits are VWS, popliteal pterygium syndrome, oral-facial-digital syndrome and Marres-Cremer's syndrome, Kabuki make-up syndrome. In the year 1954, Van der Woude described an autosomal dominant syndrome based on his observation on five pedigrees, manifesting with a combination of cleft lip and/or palate, lower lip pits. Mutations in interferon regulatory factor 6 gene have been determined to cause VWS. The incidence of lip pits among cleft patients is approximately 2%, and the combination of lip pits with cleft lip and/or palate is 24–88%. Hypodontia is present in 10–81% of cases. Other associated anomalies in VWS are syndactyly of the hands, clubfoot, genitourinary abnormalities, and cardiovascular anomalies.

Popliteal pterygium syndrome is autosomal dominant syndrome involving skin and genital anomalies. Clinical manifestation of popliteal pterygium syndrome is popliteal webbing, cleft palate with or without cleft lip, congenital heart disease, genital and nail anomalies, lower lip pits, syngnathia, and syndactyly. The manifestations of oro-facial-digital syndrome Type I are lip pits, oral frenula, oral clefts, hypoplasia of nasal cartilages, malformations of the hands, hypertelorism, and psychomotor retardation. Kabuki make-up syndrome manifestations include dysmorphic face, postnatal growth retardation, skeletal abnormalities, mental retardation, and unusual dermatoglyphic patterns.

In our case, complete examination was done, and there was absence of cleft lip and palate, skeletal abnormalities, cardiovascular anomalies, genital and nail anomalies.
The identification of familial lip pits is crucial for genetic counseling. Genetic counseling is considered important as there is a 10-fold likelihood for the offspring of a cleft patient with lip pits than in those without lip pits, to have cleft lip, with or without cleft palate.[3] Due to autosomal dominant nature, VWS affected patient has 50% risk of transmitting the condition to their offsprings.[2]

Usually, if the lip pits are mild, no treatment is needed. For cosmetic purposes or psychological depression due to mucus secretion from lip pits surgical excision can be considered.[1] Because, the sinus tracts can bifurcate, using a lacrimal probe to trace the tract may lead to leaving tract branches behind.[4] The use of bacitracin ointment with the methylene blue dye provides a dyed viscous coating of sinus tract course which does not readily extrude nor discolor the surgical field.[4] If residual sinus tract is present, they usually manifest as mucosal cyst which could be easily removed using a buccal incision.[4] Bacitracin ointment mixed with methylene blue dye are most commonly used to identify preauricular pits, sinuses, and cysts during excision and can be used in case of lip pits as well.[2]

CONCLUSION

Lip pits are rare congenital anomalies and are usually associated with other co-morbidities. Isolated lip pits with nonfamilial involvement as seen in the present case are sporadic in the literature. Hence, it is imperative that the clinicians should have a thorough knowledge of the condition and play a key role in combating the psychological trauma of the affected individuals.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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