Congenital Sinuses of the Lower Lip in Connection with Cleft Lip and Palate

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Congenital sinuses of the lower lip constitute one of the most uncommon types of congenital anomaly. Watanade and associates (*27*) found about 100 cases reported in the literature, and according to Wang and Macomber (*25*) the total published was around 140. Twenty-two new cases are described in the following, with comments on the etiology and treatment of the anomaly.

Review of the Literature

Disease Picture. The anomaly is usually a sac-like depression in the lower lip, somewhat laterally to the midline, roughly on the boundary between the skin and the mucosa. Both the depth and width of the gap vary in the approximate range of 1–15 mm. The sinus is usually in the middle of a conical protrusion. It ends in a blind pouch between the fibers of the orbicularis oris muscle and is lined with squamous epithelium reminiscent of normal labial mucosa and encircled by mucous glands which open into the sinus (*11*).

Although a bilateral sinus localized symmetrically in the lower lip is by far the commonest type, other types have also been reported: unilateral (*4, 16, 24, 27*), median (*16, 17, 24*), and bilateral asymmetrical (*24*). Wang and Macomber (*24*) also presented a case in which no actual sinuses were seen but there were typical elevations at the corresponding sites. Ludy and Shirazy (*10*) encountered in the literature five cases of congenital sinuses of the upper lip, but not a single case seems to have been described subsequently.

Apart from the cosmetic handicap, the anomaly causes the patient no actual subjective distress. There may be, at most, annoying secretion of mucous from the sinus extraorally.

Etiology. Many different hypotheses have been advanced about the etiology of congenital sinuses of the lip. Only the most commonly known are summarized here. According to Hamilton (*6*), it appears to be a

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compensatory tendency of the lower lip to grow excessively in connection with upper lip anomalies (defects). Demarquay (*2*), Touchard (*21*), and Unterberger (*23*), among other authors, have suggested that the sinus may be an anomaly caused by a developmental disorder during the fetal period such as glandular hypertrophy, mucosal invagination, inflammation, or an inclusion cyst. Keith (*8*) considered a phylogenetic anomaly possible, one corresponding to the ducts of mucous glands in some lower animals (shark) which open into corresponding place. Trendelenburg (*22*) and Hilgenreiner (*7*) held the sinus to be a remnant of the horizontal sulcus of the lip in the fetus from which, on the other hand, so-called double lip could also develop (*12*). Stieda (*19*) advanced a hypothesis which was developed later by Huber (from Nancrede, *13*), among others. According to this the sinuses are remnants of the lateral sulci of the lip during the fetal period. Basing themselves on embryonal studies, Warbrick and associates (*26*) crystallized this hypothesis, and today it is regarded as the most probable.

It is quite obvious that hereditary factors have a role in the genesis of this anomaly. Several studies have established its familial occurrence in many successive generations, for example, Koechelin (*9*), Warbrick and associates (*26*), Wang and Macomber (*24*). Likewise, it occurs fairly frequently in connection with cleft lip and/or cleft palate, but also without them. Wang and Macomber (*25*) attributed the anomaly to a genetic defect that causes retardation or inhibition of the normal development of the lip in a certain phase. On the other hand, cleft lip and palate may also be caused by genetic defect in these cases.

TREATMENT. Transposition of the sinus to open intraorally (*15*) has been proposed as a therapeutic method. Its destruction by electrocoagulation (*1*) has also been suggested, and especially in recent times, by more radical surgical excision. The excision can be made in the longitudinal or transversal direction of the lip or even as a wedge resection which cuts across the entire lip (*9, 24, 18*). As important in connection with the excision of the sinus itself is regarded the removal of the mucous glands which surround it in order to avoid the formation of secondary retention cysts.

Clinical Material

Twenty patients with sinus of the lower lip were examined at the Finnish Red Cross Hospital for Plastic Surgery in 1956–1967, that is, during the last twelve years (Table 1). In addition to these subjects, two other definite cases were detected among the patients' next of kin. All except two of the cases (№15 and №18) were bilateral and symmetrical in their location. In №15, the sinus was on the left side only, and №18 had a median sinus.

Ten of the sinus patients had cleft palate, of which three were of the submucosal type. Six patients had bilateral cleft lip and palate. One of the patients with symmetrical sinuses had bilateral cleft lip and alveolus

TABLE 1. Summary information of 22 patients with lower lip sinuses.

no.	*sex*	*heredity*	*type of cleft*	*type of sinus*	
1	f	+	palate	bilateral	nos. 1–5, members of one family
2	m	?	palate	bilateral	
3	m	+	palate	bilateral	
4	f	+	palate	bilateral	
5	m	+	palate—submucous	bilateral	
6	m	−	palate—submucous	bilateral	nos. 6–9, members of one family
7	m	+	lip, alveolus and palate, bilateral	bilateral	
8	f	+	lip, alveolus and palate, bilateral	bilateral	
9	f	+	lip, alveolus and palate, bilateral	bilateral	
10	f	+	no cleft	bilateral	nos. 10 and 11, members of one family
11	f	+	lip, alveolus and palate, bilateral	bilateral	
12	m	?	palate, submucous	bilateral	nos. 12 and 13, members of one family
13	m	+	palate, submucous	bilateral	
14	f	−	lip, alveolus and palate, right side	bilateral	
15	f	−	lip and alveolus, right side	unilateral left side	
16	m	+	lip, alveolus and palate, bilateral	bilateral	
17	f	+	lip and alveolus, bilateral	bilateral	
18	m	−	lip, alveolus and palate, bilateral	bilateral	
19	f	−	palate	bilateral	
20	f	?	no cleft	bilateral	
21	f	+	lip, alveolus and palate, left side	bilateral	
22	f	−	palate	bilateral	

and two had complete unilateral clefts. The patient with sinus of the lower lip only on the left had a right-sided cleft lip and alveolus, but intact palate. Only two sinus patients presented no cleft at all.

In 13 cases, including one of the two patients without cleft, there were clefts of lip and/or palate in the close relations of the patients, suggestive of positive heredity. In five cases no clefts could be found in the previous generations, and in four cases, including one of the two sinus patients without any cleft, the information available was not adequate.

Four patients, a father, two daughters, and a son (№6–9), belonged to the same family: the father had submucous cleft palate and all three children had bilateral cleft lip and palate. Four other patients (№2–5) belonged to a family in which a total of six members in four generations had cleft palate and three had cleft lip and palate (Figure 1). Subjects 10 and 11 were mother and daughter. The daughter had a bilateral com-

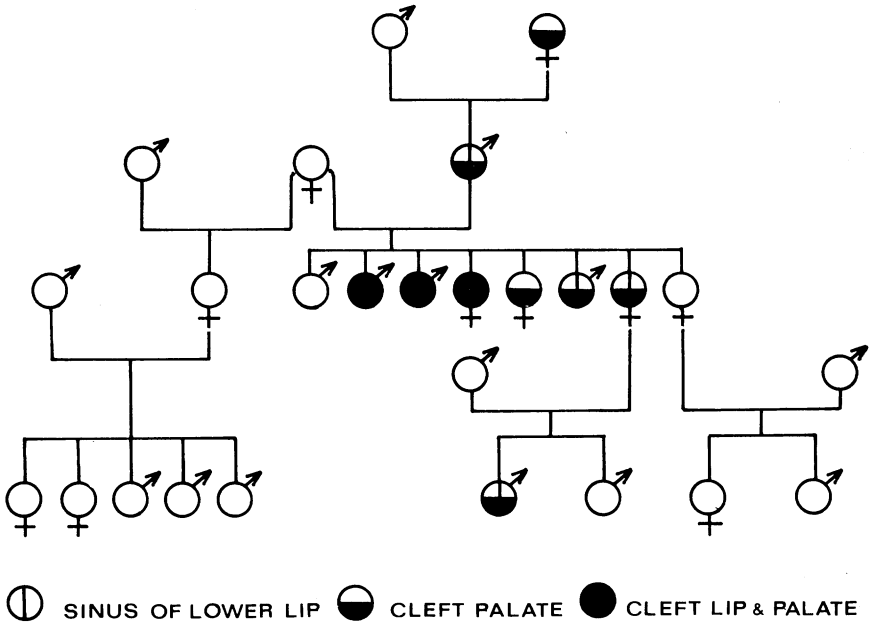

FIGURE 1. A family with cleft lip and palate. In three generations, four cases had lower lip sinus.

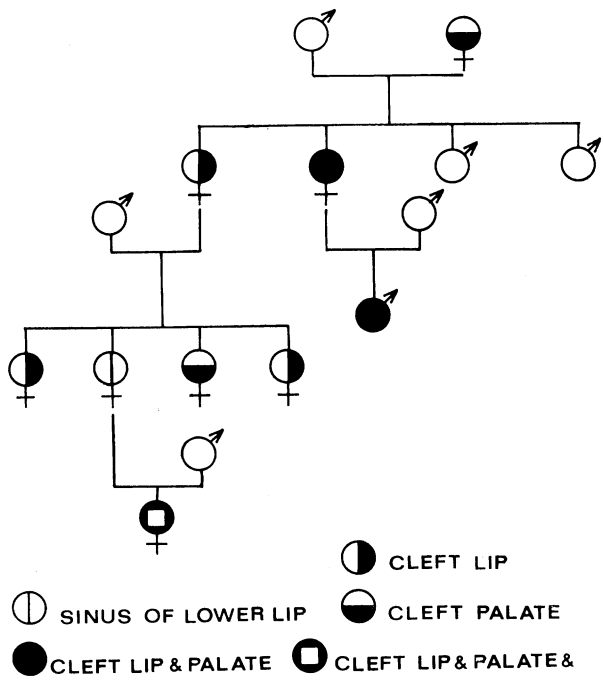

FIGURE 2. A family with cleft lip and palate. In two generations, two cases had lower lip sinus.

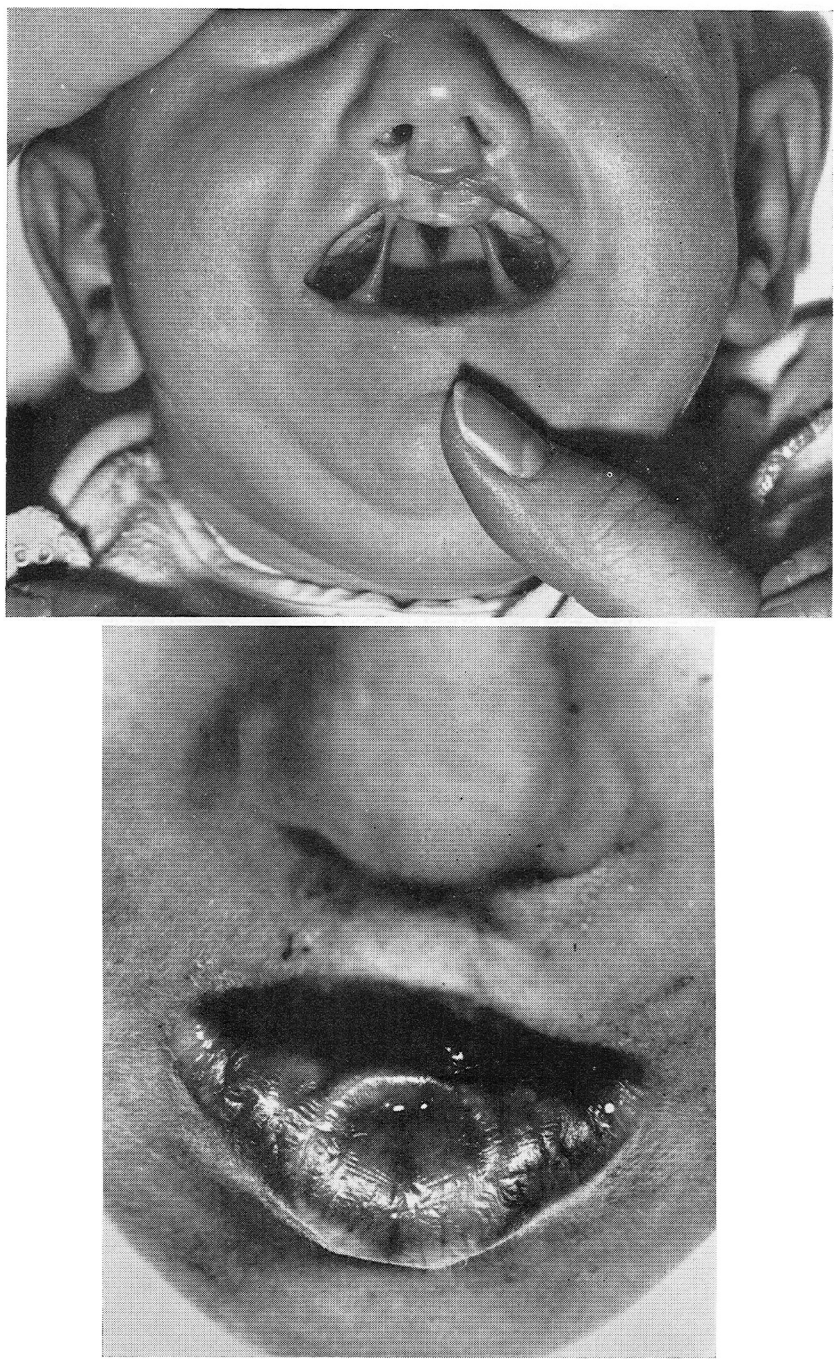

FIGURE 3. Subject #16, *top,* age three months, bilateral cleft lip and palate, and symmetrical epithelial strands running from the lower lip to the alveolar ridge of the maxilla. *Bottom,* age six years, fairly extensive and deep median sinus in the lower lip, cleft lip and palate have been corrected primarily.

plete cleft, but the mother had only sinuses without any cleft. The grandmother on the maternal side had no sinuses, but had unilateral cleft lip, and her mother, again, had cleft palate. There were several other clefts of varying types in near relations of the family (Figure 2).

Subject 18 had a median lower lip sinus (Figure 3) as well as symmetrical, thin epithelial strands which ran roughly from the usual site of the lateral sinuses in the lower lip to the maxillary alveolar ridge on both sides. He also showed bipartite scrotum; prepuce resembling hypospadias; and in both popliteal spaces alar folds which caused flexion contracture of the knee joints. There was no family history of anomalies.

Histologic examination showed that the sinuses were lined with fairly thick and irregular squamous epithelium closely reminiscent structurally of labial mucosa. Salivary gland ducts opened here and there into the sinus (Figure 4).

In subject 11, the chromosomes were analyzed from peripheral leuco-

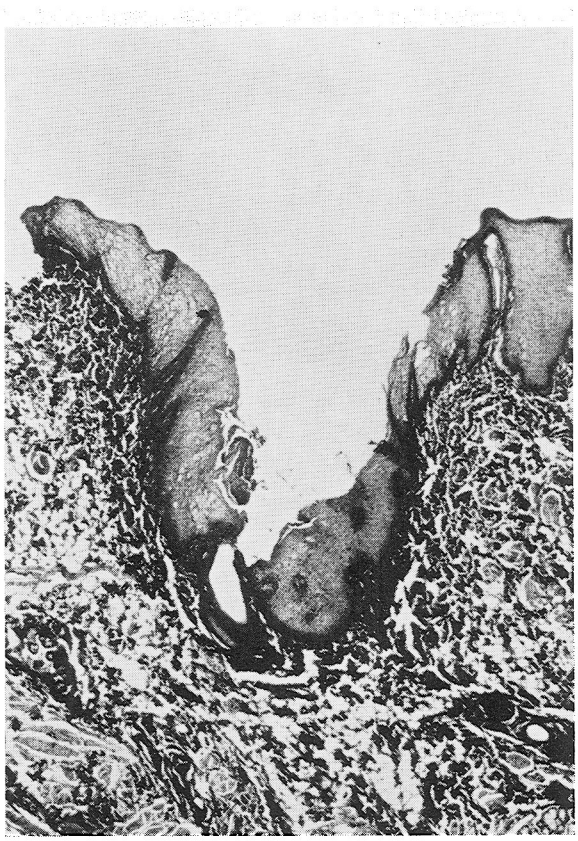

FIGURE 4. Cross section of the lower lip in the longitudinal direction of the sinus. The epithelial structure of the sinus corresponds to normal labial mucosa (haematoxylin van Gieson × 60).

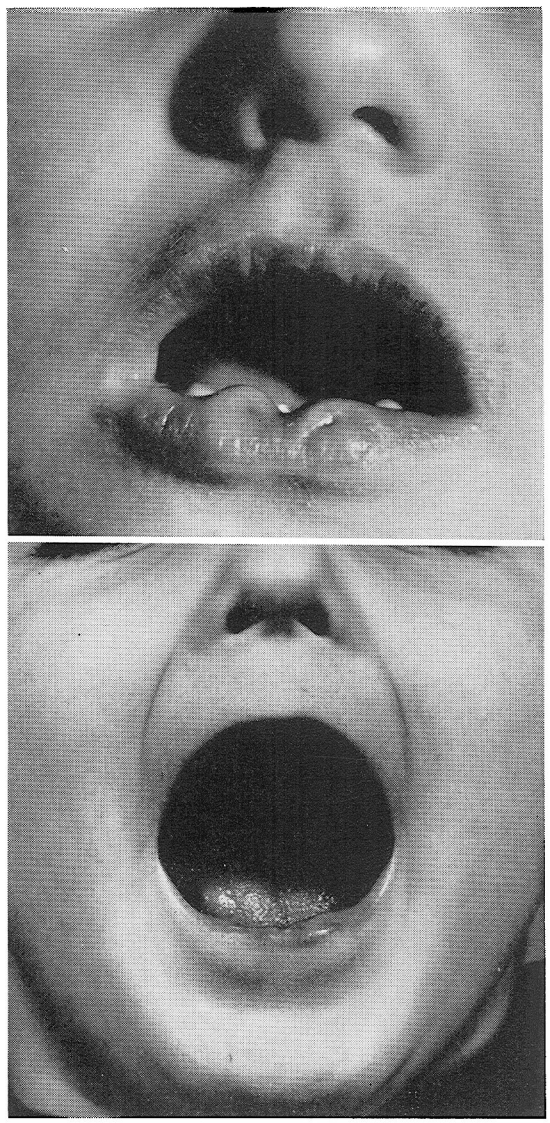

FIGURE 5. Subject #5. *top,* bilateral symmetrical sinuses of the lower lip. *Bottom,* after excision of the sinuses longitudinally to the lip.

cytes. The patient had a normal karyotype with 46 chromosomes and normal XX sex-chromosomes.

Twelve of our patients were born in the years 1955–1964; three were born in 1948–1954; and all the others were older. Since a total of 1347 cleft lip and/or palate patients born in 1955–1964 and 2206 patients born in 1948–1964 have been treated in our hospital, the incidence of lip sinuses in patients with cleft lip and palate was 0.7% in the whole series and 0.9% after 1954.

In five subjects (№s 4, 5, 13, 17, 20) mere excision of the sinuses in the longitudinal direction of the lip was performed (Figure 5) and in three subjects (№s 7, 8, 16) transversal excision or wedge resection were performed. In one of the former and in all the latter cases secondary trimming proved necessary for a satisfactory cosmetic result. In one case (№9), the part of the labial mucosa that corresponded to the posterior wall of the sinus was excised and the wall of the sinus was turned 180 degrees, to form the mucosa of the lip. The resultant mucocele was excised secondarily. There was one instance (№11) in which the posterior walls of the sinuses were excised together with the corresponding parts of the labial mucosa, and the lateral walls of the sinuses were turned to the sides and sutured to the labial mucosa so that the anterior wall of the sinus became the mucosa of the lip. Secondary correction for cosmetic reasons was required also in this case. Operative correction has been postponed in 12 cases, either because the child has been considered too young or because the anomaly has not inconvenienced the patient.

Discussion

The best explanation about the etiology of lower lip sinuses is that a lower lip sinus is a remnant of the lateral sulci originating from a genetic defect. If sinuses really do occur also in the upper lip, as has been asserted, they would be difficult to explain on the basis of this theory. The same applies to median sinuses. The latter might possibly be a remnant of the median sulcus. On the other hand, if upper lip sinuses were some form of cleft lip, it is surprising that they have not been reported more often.

The vertical, symmetrical, groove-like deep folds of skin from the jaw to the lower lip, encountered in Freeman-Sheldon's syndrome and only recently described by Rintala (*14*), must perhaps also be regarded primarily as remnants of the distal and medial parts of the lateral sulci. If we compare the lower lip sinuses with cleft lip and palate anomaly in which the full range of degrees and variations are represented fairly evenly, it appears strange that the remnants are always at the cranial end of the sulci, except in the three cases of Freeman-Sheldon's syndrome mentioned above. To the best of our knowledge, no transitional forms between typical lip sinuses and the skin folds in Freeman-Sheldon's syndrome have been described. The pathogenesis of lower lip sinuses probably cannot, therefore, be regarded as explained.

Our own cases support the view of Wang and Macomber (*25*) that the sinuses are more common in females than in males. They argue strongly for the theory of familial occurrence suggestive of heredity and the connection with cleft lip and palate. All cases of cleft lip and palate in Finland are concentrated in our hospital where other simultaneous anomalies are also taken into consideration. It is thus possible that some cases in which there was no connection between sinuses and

clefts did not come to our attention, as evidenced by the fact that several of our patients were not interested in treatment of the sinuses.

As the pedigrees of several of our cases show, different types and degrees of clefts may appear in successive generations, with or without simultaneous sinuses (*3*). On the other hand, the member transmitting the cleft anomaly may only present the sinuses. This suggests that the anomaly may be caused by a combination of genetic and environmental factors. The last-mentioned would be decisive for the severity, and, depending upon the time of its action, the exact type of the defect. The genetic factor may only act as a sensitizing agent for certain types of anomalies.

The incidence of lip sinuses in cleft lip and palate patients in our series is considerably higher than the 0.5% reported by van der Woude (*28*). This may be due to a real increase in the incidence, as our series suggests. On the other hand, it is possible that in the early years not all sinuses have been reported in the case reports. That discrepancy may be responsible at least in part for the increase in the number of cases in our series too.

Taylor and Lane (*20*) draw attention to the occurrence of other anomalies in connection with sinuses and clefts. The incidence of other anomalies in our cleft material was about 15% (*5*). A noteworthy observation is that unilateral sinuses do not appear to occur solely on the same side as the cleft.

Regarding the therapeutic approach, we have found excision in the sagittal direction and wedge resection to be unsatisfactory methods which always require additional corrective measures. Excision of the sinus in the longitudinal direction of the lip, which takes in also the surrounding mucous glands, seems to be the most beneficial and simplest method. If the sinus is extensive and the lip correspondingly thick, removal of the tissue between the labial mucosa and the posterior wall of the sinus may give a satisfactory result. This leaves intact the glandular ducts which may open into the anterior wall of the sinus and helps to reduce the risk of retention cysts.

Summary

Twenty-two patients with sinus of the lower lip are described. All except two appeared in connection with cleft lip and/or palate. Only one sinus, encountered on the contralateral side to the cleft lip and alveolus, was unilateral. Another was median, appearing in connection with bilateral cleft lip and palate. All the other sinuses were bilateral and symmetrical. A distinct familial occurrence was established in the majority of the cases. None of the numerous hypotheses proffered regarding the etiology of these sinuses appears to account completely satisfactorily for their pathogenesis. The incidence of lower lip sinuses in a cleft lip and palate material was approximately 0.9%. Elliptical excision of the sinus

longitudinally to the lip would seem to be the simplest therapeutic method.

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