Palatal Anomalies in the Syndromes of Apert and Crouzon

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Patients manifesting the syndromes of Apert and Crouzon present anomalies of the palate which may or may not include actual clefting. Previous descriptions of these anomalies have varied both in content and in reported frequency of occurrence. Confusion may have stemmed from the fact that (1) certain of the anomalies are age dependent, becoming fully manifest and identifiable only with increasing age; and (2) the configuration resulting from the lateral palatal swellings is such that a pseudocleft is produced which is often misdiagnosed as a true cleft. Indeed, it is the appearance of this pseudocleft which has resulted in the referral of such cases to cleft palate clinics.

Apert Syndrome

Apert syndrome (acrocephalosyndactyly) is characterized by oxycephaly (acrocephaly) and syndactyly of the hands and feet (*1*). The cranial synostosis in this syndrome most often involves the coronal complex of sutures, alone or together with fusion of the sagittal suture. Hypoplasia of the middle third of the face, including the maxilla, contributes in part to the exophthalmus and relative prognathism (*1, 2, 5, 15*) (see Figure 1). Associated findings include skeletal abnormalities as well as deformities of the temporal bone, eyes, cranial base and cervical spine (*5*).

THE PALATE IN APERT SYNDROME. According to Gorlin and Pindborg (*5*), most frequently observed in Apert's is a high-arched palate, occasionally with a marked median groove. This description coincides with that given by Smith (*12*). Gorlin and Pindborg describe a cause-effect sequence leading to this configuration: maxillary hypoplasia → compression of the upper dental arch which becomes V-shaped → irregular positioning of teeth → marked thickening of the alveolar process.

Solomon et al. (*13*) have described these lateral swellings on the palatine processes as present in infancy and increasing in mass as the child grows

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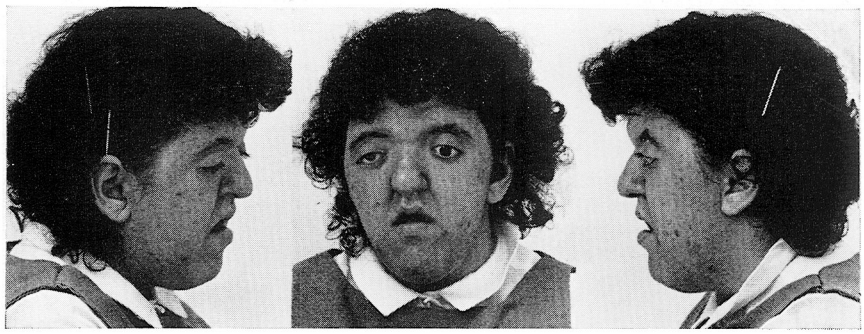

FIGURE 1. Patient (CCFA #1013) with typical facial features of Apert syndrome. Note hypoplasia of middle third of the face and obligatory mouth-open posture at rest.

older (see Figure 2). The resulting bilateral swellings produce a deep median groove which can lead to a mistaken diagnosis of cleft palate (*10*). Coronal tomograms have shown the lateral palatal swellings to consist mainly of soft tissue, and histologic studies have revealed acid mucopolysaccharide deposits (*13*).

The frequency with which this palatal configuration is found in Apert patients is subject to question. Rubin (*11*) states that "malformation of the hard or soft palate" is found in one-half the cases of Apert, but does not specify the type of malformation. Park and Powers (*9*) found a similar incidence (12/21) of "abnormalities of the hard or soft palate." Cohen (*3*) found the bilateral swellings and deep median groove in 34 of 37 patients. Solomon et al. (*13*) found this configuration in all 13 Apert patients they examined. As pointed out by the latter authors, the lateral palatal swellings are less marked in infants and young children than at later ages and may be overlooked.

The swollen configuration of the hard palate may, of course, be combined with actual clefting (*3, 12, 13*). Gorlin and Pindborg (*5*) state that posterior cleft palate or bifid uvula is found in at least 25% of Apert patients. Cohen (*3*) found cleft soft palate in 13 of 40 patients, and bifid or notched uvula in 17 of 40. Solomon et al. (*13*) found cleft soft palate in 3 of their 13 cases, and bifid uvula in 6. However, the latter authors obtained most of their cases from the Center for Craniofacial Anomalies, a unit to which patients with clefts are commonly referred.

Survey of CCFA Apert Cases, Clinical Findings. The Center for Craniofacial Anomalies, Abraham Lincoln School of Medicine, has longitudinal records on 19 Apert cases (10 ♂, 9 ♀). On clinical examination, all 19 cases showed the narrow, high-arched palate with lateral accumulations of soft tissue on the palatine processes. Bifid uvula was present in 6 of the 19 (incidence = 32%), and overt cleft palate in 2 (incidence = 11%, combined incidence = 43%).

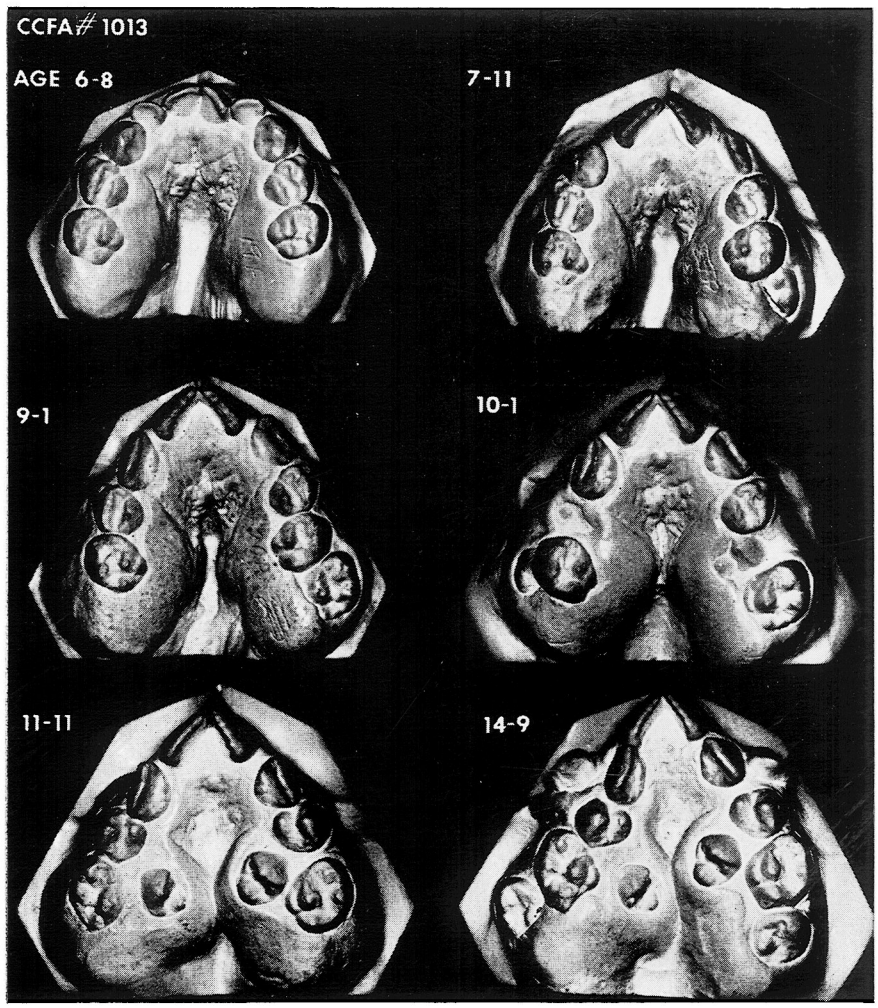

FIGURE 2. Serial casts of Apert patient (CCFA #1013) from the age of 6 years, 8 months to 14 years, 9 months. Note progressive increase in size of swellings on lateral margins of palate. In this case, the deformity was accentuated by the ectopic eruption of the second premolars.

of our Apert patients appeared unusually long. Accordingly, lateral cephalometric radiographs on each of the Apert (and Crouzon) patients were traced and measured to determine length and thickness of the soft palate at rest. (See Figure 3.) In each case, the measurements were taken from the most recent cephalometric films available. These measurements were then compared to norms for length and thickness of the soft palate obtained by Subtelny (*14*) from serial cephalometric roentgenograms on 30 normal subjects. Figure 4 shows the means for length of the soft palate (sexes combined) obtained on Subtelny's subjects, with the shaded area

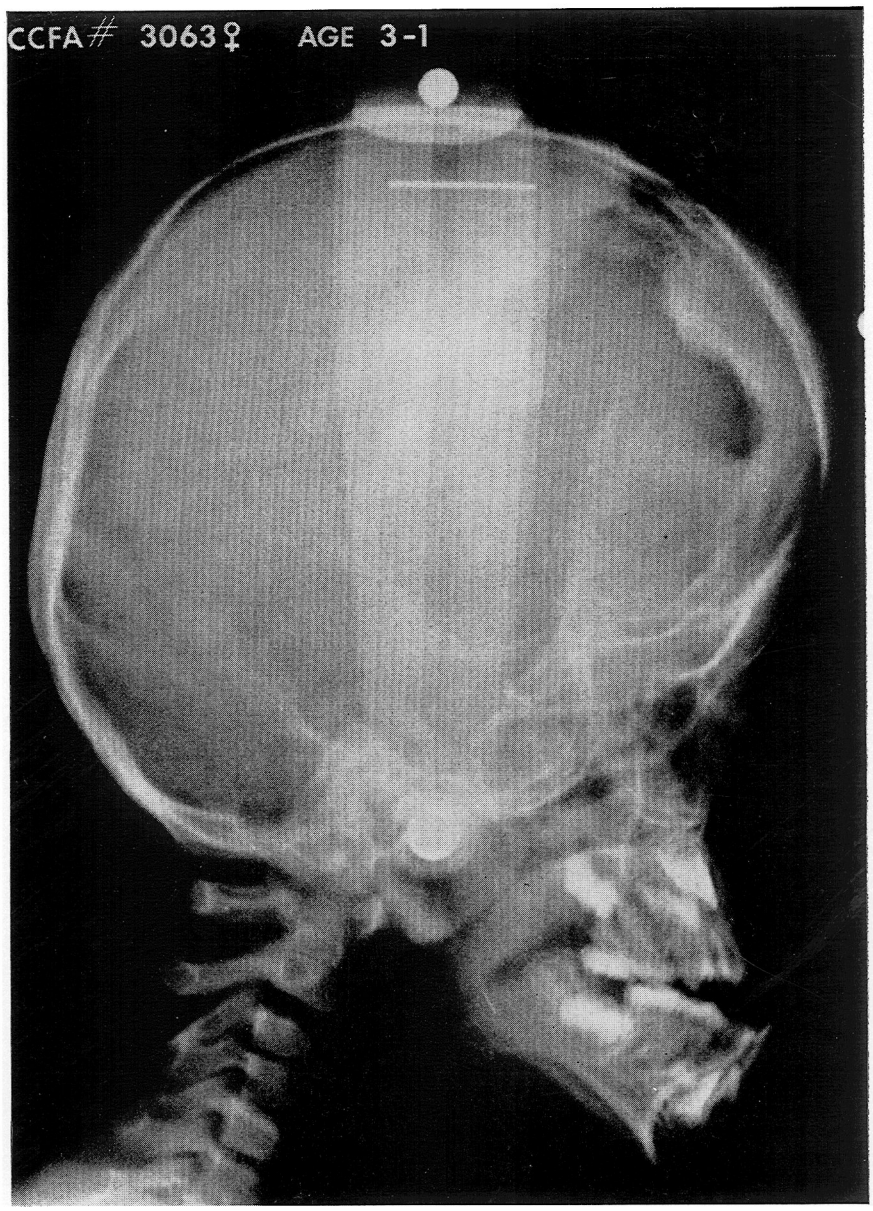

FIGURE 3. Lateral cephalometric roentgenogram of child with Apert syndrome (CCFA #3063). Note midface hypoplasia, cervical spine anomalies, and constricted nasopharyngeal airway.

representing the 95% confidence interval for these means. Each of the CCFA Apert and Crouzon patients is plotted on this graph according to the nearest age level at the time the cephalometric films were taken. (CCFA patients above the age of 18 are plotted to the right of the graph, since 18

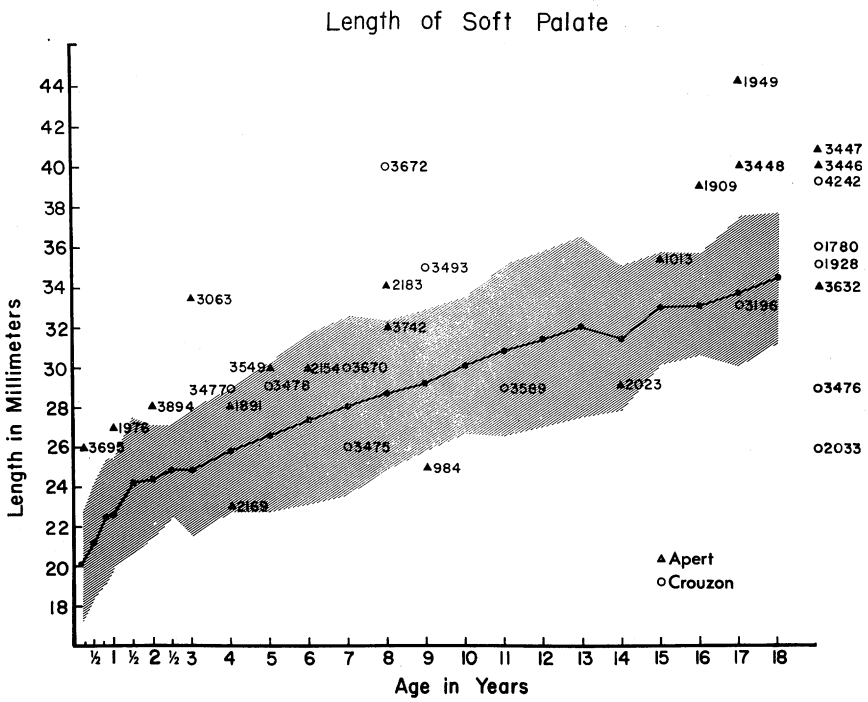

FIGURE 4. Soft palate length at rest (in millimeters) of CCFA Apert and Crouzon patients compared to Subtelny's norms (represented by ●——●, sexes combined). The shaded area represents the 95% confidence interval for the mean length of the soft palate in Subtelny's sample at each age level. CCFA patients who were over 18 years of age at the time the measurements were taken are plotted to the right of the graph. (See Subtelny, 1957.)

Radiographic Findings. On peroral examination, the soft palate of many was the highest age for which Subtelny obtained norms.) Figure 5 plots the CCFA patients in comparison to Subtelny's norms for thickness of the soft palate.

Ten of the 19 Apert patients fell above the upper limit of the 95% confidence interval for length of the soft palate. Two of these (※3063 and ※2183) had bifid uvulas, and one (※3448) had a repaired cleft of the palate.

Eight of the Apert cases fell above the upper limit of the 95% confidence interval for thickness of the soft palate. Two of these patients (※3448 and ※2023) had repaired clefts of the palate, and four (※3063, ※2183, ※984 and ※1013) had bifid uvulas. Four cases (※3063, ※2183, ※3448 and ※1949) fell above the upper limit of the 95% confidence interval for both length and thickness of the soft palate.

No statistical tests were carried out on the Apert cases in comparison to Subtelny's normal subjects due to the fact that we lacked a sufficient number of cases at each age level.

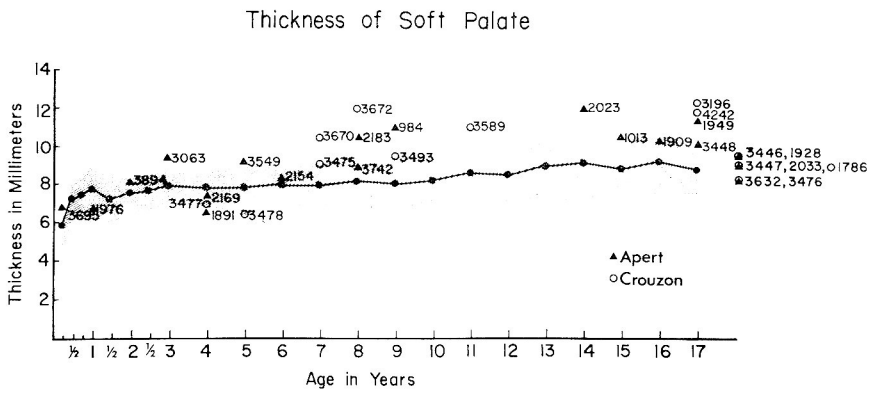

FIGURE 5. Soft palate thickness at rest (in millimeters) of CCFA Apert and Crouzon patients compared to Subtelny's norms (represented by ❶——●, sexes combined). The shaded area represents the 95% confidence interval for the mean thickness of the soft palate in Subtelny's sample at each age level. CCFA patients who were over 17 years of age at the time the measurements were taken are plotted to the right of the graph. (See Subtelny, 1957.)

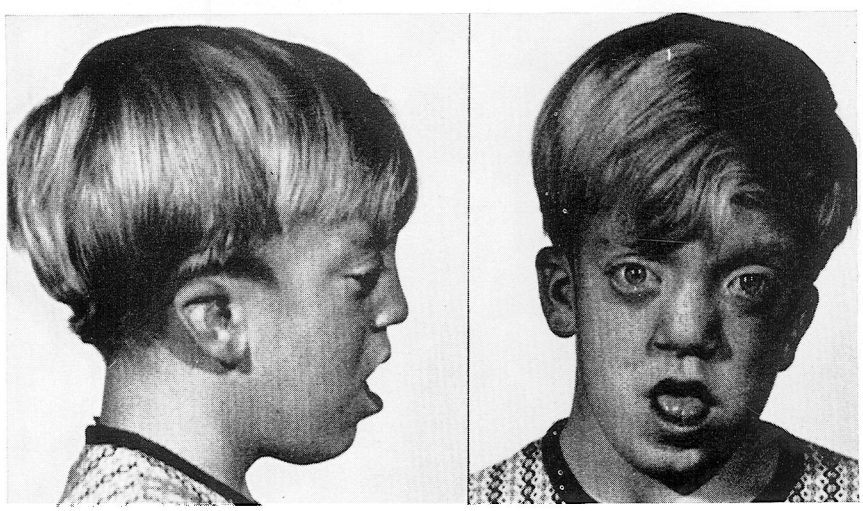

FIGURE 6. Patient with typical facial features of Crouzon disease (CCFA #3493).

Crouzon Disease

The cranial and facial characteristics of Crouzon disease (craniofacial dysostosis) are very similar to those of Apert syndrome, involving brachycephaly, maxillary hypoplasia, relative prognathism, exophthalmus, and hypertelorism (*5*) (see Figure 6). In Crouzon disease, the coronal, sagittal and lambdoidal sutures are prematurely synostosed in the majority of cases (*1*). In a recent roentgenographic study (*8*), it was found that Apert and Crouzon patients could not be distinguished on the basis of skull shape alone.

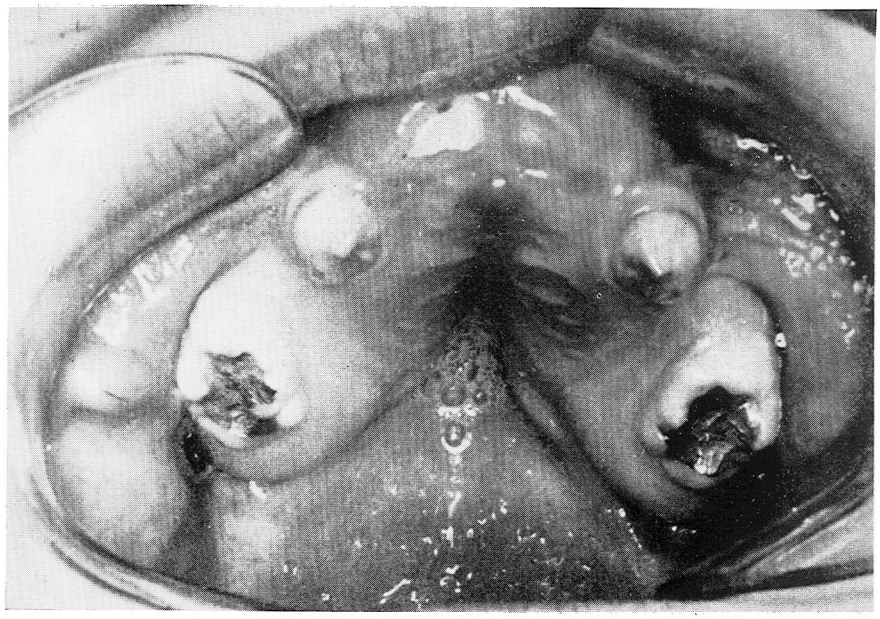

FIGURE 7. Palate of Crouzon patient (CCFA ※3493) pictured in Figure 6. Note similarity to palatal configuration of Apert patient in Figure 2.

THE PALATE IN CROUZON DISEASE. As in Apert syndrome, hypoplasia of the maxilla leads to crowding of the maxillary teeth (*1, 6, 7*) and V-shape of the maxillary arch (*6*). Gorlin and Pindborg (*5*) state that, in most cases, the palate is described as high-arched and short. Cleft palate and bifid uvula have been reported in this syndrome (*4, 6, 7, 11*) but incidence figures have not been given.

Survey of CCFA Crouzon Cases, Clinical Findings. The CCFA currently has records on 13 cases of Crouzon (7 ♂, 6 ♀). On clinical examination, 3 patients (※3672, ※3493 and ※1780) showed narrow, high-arched palates with lateral accumulations of soft tissue barely separated by a deep median groove (see Figure 7). These palates were indistinguishable from that seen in Apert syndrome. One of these 3 cases (※3493) had a bifid uvula. An additional 8 of the 13 cases showed high-arched palates not so extreme as that seen in Apert syndrome (see Figure 8).

Radiographic Findings. Figure 4 indicates that 3 of the Crouzon patients (※3493, ※3672 and ※4242) fell above the upper limit of the 95% confidence interval for palatal length. It is interesting to note that 2 of these 3 cases were among those showing palatal vaults similar to that seen in Apert syndrome.

Five of the Crouzon cases (※3670, ※3672, ※3589, ※3196 and ※4242) fell above the upper limit of the 95% confidence interval for thickness of the soft palate (see Figure 5). Only one of these (※3672) was also above the upper limit of the 95% confidence interval for length of the palate.

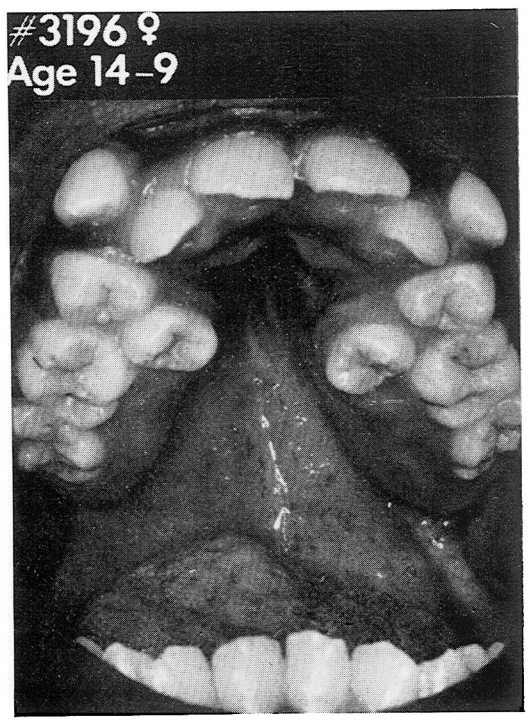

FIGURE 8. Palate of Crouzon patient (CCFA #3196). Note that palatal arch is high and narrow but not so extreme as that seen in Apert patient (see Figure 2).

Again, no statistical tests were carried out on the Crouzon cases in comparison to Subtelny's normal subjects due to the lack of a sufficient number of cases at each age level.

Discussion

The above findings should alert the clinician to the probability of structural abnormalities of the hard and soft palate in patients with the syndromes of Apert and Crouzon. These abnormalities, in combination with other cranial and oral features of the syndromes, can affect respiration and speech, among other functions. Indirectly, they can also contribute to the abnormal appearance of the face.

The extremely shallow oropharynx (see Figure 3) has been previously recognized as an etiologic factor in the obligatory mouth breathing and forward carriage of the tongue so often seen in these patients (*13*). Obviously, the presence of an abnormally long and/or thick soft palate in the shallow oropharynx adds to the severity of the problem. Under these circumstances, it is not surprising that speech is often markedly denasal. It is also not surprising that the amount of palatal elevation seen on phonation is often minimal: The soft palate simply has no room in the oropharynx for superior-posterior movement.

The incidence of cleft palate and bifid uvula in CCFA Apert and Crouzon patients may be spuriously high due to the reputation of the Center as a cleft palate treatment unit. Our findings do not immediately suggest a relationship between the presence of clefting and abnormal length and/or thickness of the soft palate.

The potential effects on speech of the several oral anomalies in these syndromes have been described previously (*10*). The specific effects of the palatal anomalies may be postulated as follows:

1. If there is an actual cleft of the palate, control of the airstream in speech may be a problem (depending upon the configuration of the oropharynx) until the cleft is closed.
2. The hyperplastic soft tissue filling the anterior palatal vault may interfere with tongue tip placement on lingua-alveolar consonants.
3. A soft palate that is abnormally long and/or thick may cause or contribute to denasality.

The radiographic data suggest a trend of greater deviation in palatal length with increasing age in our Apert and Crouzon patients (see Figure 4). However, this apparent trend must remain merely a "suggestion" until sufficient longitudinal data on individual patients are available. The major problem in collecting such data is that of obtaining cephalometric films on very young patients: The use of even mild sedation is often contraindicated in the infant whose respiration is threatened by a constricted nasopharyngeal airway.

To date, we have no information regarding the nature of the velar tissue in those patients showing unusual length or thickness of the soft palate. Thus we cannot postulate a cause for this phenomenon. It is possible that the acid mucopolysaccharide deposits found in the hyperplastic soft tissue filling the palatal vaults in some of the patients may be present in the velum as well.

Studies are currently under way (1) to track the growth of the soft palate over time in these syndromes, (2) to determine the nature of the velar tissue, and (3) to document the effects of the several oral anomalies on articulation.

Summary

Patients with the syndromes of Apert and Crouzon present anomalies of the palate which may or may not include actual clefting. In a sample of 19 Apert patients ranging in age from 4 months to 30 years, all 19 were found to have narrow, high-arched palates with later accumulations of soft tissue barely separated by a deep median groove. This configuration frequently leads to a misdiagnosis of cleft palate. Previous studies have revealed the presence of acid mucopolysaccharide deposits in the hyperplastic soft tissue filling the palatal vault. Bifid uvula was present in 6 of the 19 patients, and overt cleft palate in 2. On radiographic examination, 10 of the Apert patients were shown to have soft palates of abnormal length in com-

parison to Subtelny's norms, and 8 showed abnormal velar thickness (4 overlapping cases). In 13 cases of Crouzon disease ranging in age from 4 years to 30 years, 3 showed palatal vaults indistinguishable from that seen in Apert patients, and 8 showed high-arched palates not so severe as that seen in Apert. One Crouzon patient had a bifid uvula. There was no apparent relationship between clefting and abnormal size of the soft palate. There appeared to be a trend of greater deviation in velar length with increasing age, but additional longitudinal data are required. The cause for the unusual length and thickness of the soft palate in these patients is not known.

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