Cleft Palate and Congenital Palatopharyngeal Incompetency in Mandibulofacial Dysostosis: Frequency and Problems in Treatment

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In a series of 25 cases of *mandibulofacial dysostosis*, 7 patients had isolated *clefts* of the palate, 1 had a complete unilateral cleft of the lip and palate, and 8 had *congenital palatopharyngeal incompetency* (CPI). The CPI appeared in four forms: (a) complete agenesis of the soft palate, (b) foreshortening of the soft palate associated with a submucous defect of the hard palate, (c) submucous defect of the hard palate with adequate palatal length but inadequate elevation in speech, and (d) lack of adequate palatal elevation in the absence of a submucous defect or reduced length of the soft palate. Inadequate velopharyngeal function, whether congenital or subsequent to palatal repair, may be masked by the presence of other speech problems in this syndrome, particularly by the "muffled" voice quality which appears to be associated with an elevated and retracted *tongue* posture. Both prosthetic and surgical treatment of inadequate velopharyngeal function are complicated in patients with MFD by other structural anomalies, particularly inadequate oral opening and constriction of the airway.

The morphologic features of mandibulofacial dysostosis as described by Berry (1889), Treacher Collins (1900), and Franceschetti and associates (1944, 1949), among others, include hypoplasia of the malar bones, antimongoloid slant of the palpebral fissures, lower lid defects, bilateral deformities of the auricle and middle ear, and hypoplasia of the mandible and maxilla (Figure 1). When present, the mandibular hypoplasia results in a peculiar curvature of the lower border of the mandible with consequent malocclusion usually involving an open bite (Roberts *et al.*, 1975).

Cleft palate has been reported to occur in approximately 40% of the patients manifesting mandibulofacial dysostosis (MFD) (Gorlin and Pindborg, 1964). Cleft lip and palate have been reported in two cases (Smith and Jones, 1975; Wildervanck, 1975) where MFD facial stigmata were associated with limb malformations. Cleft lip was also present in one of Berry's patients (1889). Bowen and Harley (1974) described a child with Nager's acrofacial dysostosis (mandibulofacial dysostosis with limb malformations) who had a "short palate." Two investigations of submucous cleft palate (Fara *et al.*, 1971; Weatherly-White *et al.*, 1972) each reported one case in which the palate defect occurred in

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conjunction with MFD. Walker (1974) described a family in which one of two female siblings with MFD (in a total sibship of 5) showed submucous cleft and bifid uvula.

At the University of Illinois Center for Craniofacial Anomalies, we have become increasingly aware of the frequency of occurrence of congenital palatopharyngeal incompetency (CPI) among patients with mandibulofacial dysostosis. In our series of cases, congenitally short and/or immobile palates have been observed both (a) in conjunction with a submucous defect of the hard palate and (b) in the absence of any such defect. The possibility of an ascertainment bias is recognized in view of the Center's reputation as a treatment facility. Thus, the presence of cleft palate might have influenced the number of MFD patients who were referred because of the palatal problems. However, two facts should be noted. First, in those cases of congenital palatopharyngeal incompetency reported below, the condition was not discovered until *after* the patient had begun treatment through the Center. Second, in several instances of overt cleft palate occurring in conjunction with MFD, the cleft was not reported in the original referral.

As a result of our clinical experience, we have become concerned that both overt cleft palate and congenital palatopharyngeal incompetency may be underreported in the MFD population. The following report is thus concerned

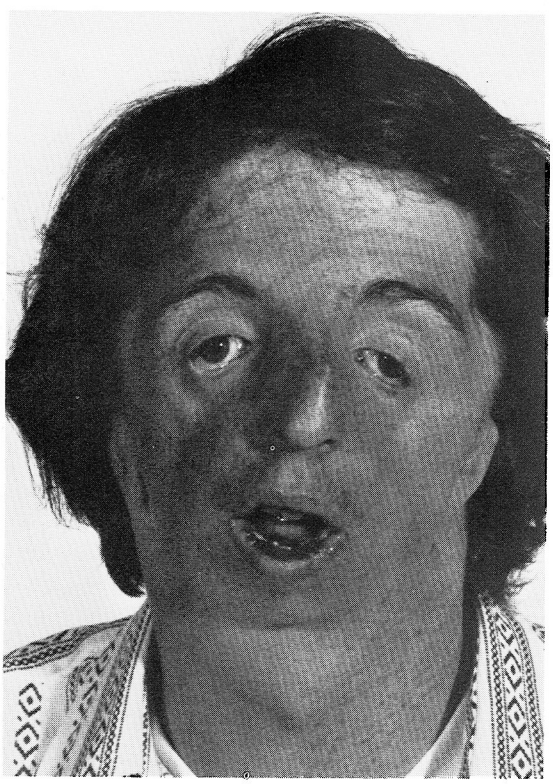

FIGURE 1. Typical facies of mandibulofacial dysostosis. Note hypoplasia of malar bones, antimongoloid slant of palpebral fissures, lower lid defects, bilateral deformities of auricles, mandibular and maxillary hypoplasia.

with (1) the frequency of overt cleft palate and CPI in our MFD population, and (2) problems in treatment of palatopharyngeal incompetency in these patients.

Survey of CCFA Cases

Reliable records were available on a series of 25 patients with mandibulofacial dysostosis (19 active cases, six inactive; 9♂, 16♀). At the time this survey was made, the active patients ranged in age from five months to 31 years, seven months.

Of the 25 patients (Table 1), seven had isolated clefts of the palate (not involving the lip or alveolar ridge) and one had a complete cleft of the lip and palate. The latter patient has a twin of the same sex (zygosity has not been fully established at this time) who also has MFD but no cleft. Eight additional patients in our series of 25 had congenital palatopharyngeal incompetency which appeared in four forms:

a. complete agenesis of the soft palate, with a uvula attached directly to the posterior border of the hard palate (one case);
b. submucous cleft of the posterior ⅓ of the hard palate and foreshortening of the soft palate (one case, see Figure 2);
c. submucous defect of the hard palate with adequate length of the soft palate but lack of adequate palatal elevation during speech (two cases). One of these patients has a bifid uvula, while the other cannot open her mouth sufficiently to allow visualization of the uvula.
d. lack of adequate or consistent palatal elevation during speech production in the absence of any submucous defect or reduced length of the soft palate (four cases).

TABLE 1. Frequency of cleft palate and four forms of congenital palatopharyngeal incompetency in a series of 25 cases of mandibulofacial dysostosis.

	♂	♀	TOTALS
Normal	4	5	9
Cleft lip and palate		1	8
Cleft palate only	3	4	
Agenesis of soft palate		1	8
Submucous defect, short soft palate		1	
Submucous defect, inadequate palatal elevation		2	
Inadequate or inconsistent elevation (adequate length, no submucous defect)	2	2	
TOTALS	9	16	25

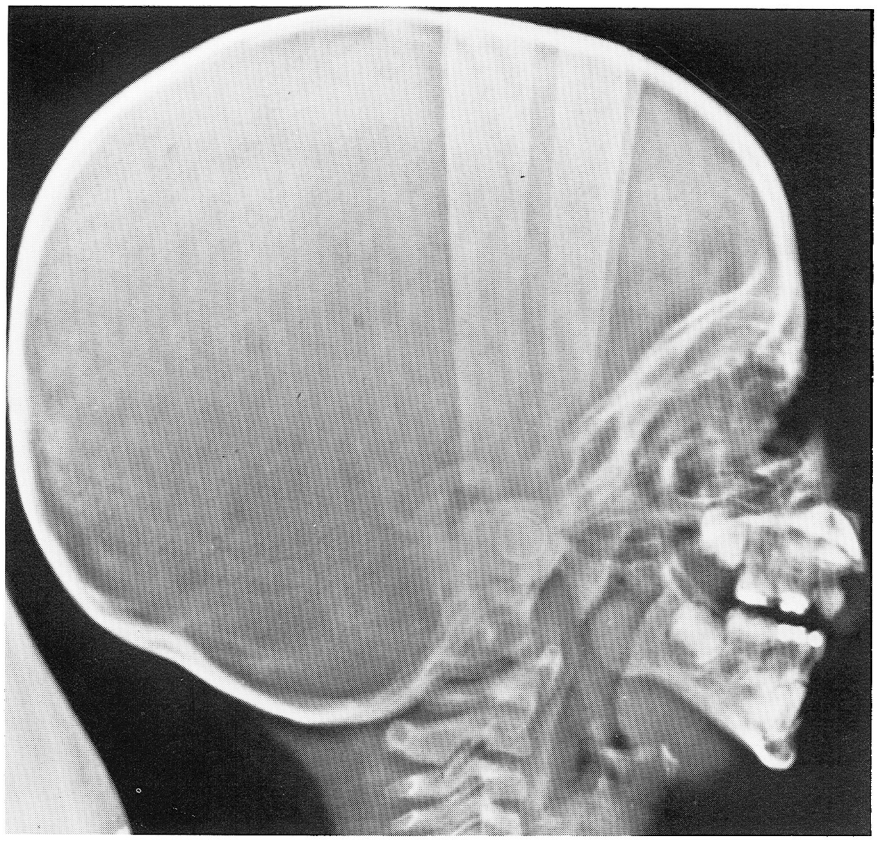

FIGURE 2. Lateral cephalometric roentgenogram of MFD patient with foreshortening of the soft palate and submucous defect of the hard palate.

The variability in palatal findings in this series of cases is not surprising in view of the fact that mandibulofacial dysostosis is known to vary widely in both penetrance and expressivity (Axellson, *et al.*, 1963; Cotter, 1968; Rogers, 1964).

Treatment Histories of CCFA Cases

A. CLEFT PALATE. Of the eight cases of mandibulofacial dysostosis with cleft palate, two became inactive to the clinic before they could be treated, and two are new to the clinic. The remaining four all showed hypernasal speech following closure of the palate. In two of these cases, prosthetic treatment failed to improve speech, and in two speech continued to be hypernasal following superior-based pharyngeal flaps.

B. CONGENITAL PALATOPHARYNGEAL INCOMPETENCY (CPI). Four cases of CPI have not been treated to date for their hypernasality because of the priority of other treatment. A fifth case became inactive before treatment was initiated. Two patients (ages five and seven) have recently been fitted with prostheses designed to totally obstruct the velopharyngeal port. It is hoped that this "over-correction" will help to break down established patterns of nasal emission;

initial results are encouraging. Finally, the patient with complete agenesis of the soft palate is a young adult who had been treated prosthetically over a period of years, unfortunately without significant alteration in nasal emission or resonance balance.

Comments on Treatment

Physical management of the velopharyngeal port in MFD can be perilous (and unsuccessful) for a number of reasons:

1. The nasopharynx may be inaccessible because of restricted oral opening resulting from hypoplasia of the mandible. Lack of growth of the ramus and abnormalities of the temporomandibular joint may result in such severe limitation of ability to open the mouth that surgery cannot be performed nor can a prosthesis be introduced.

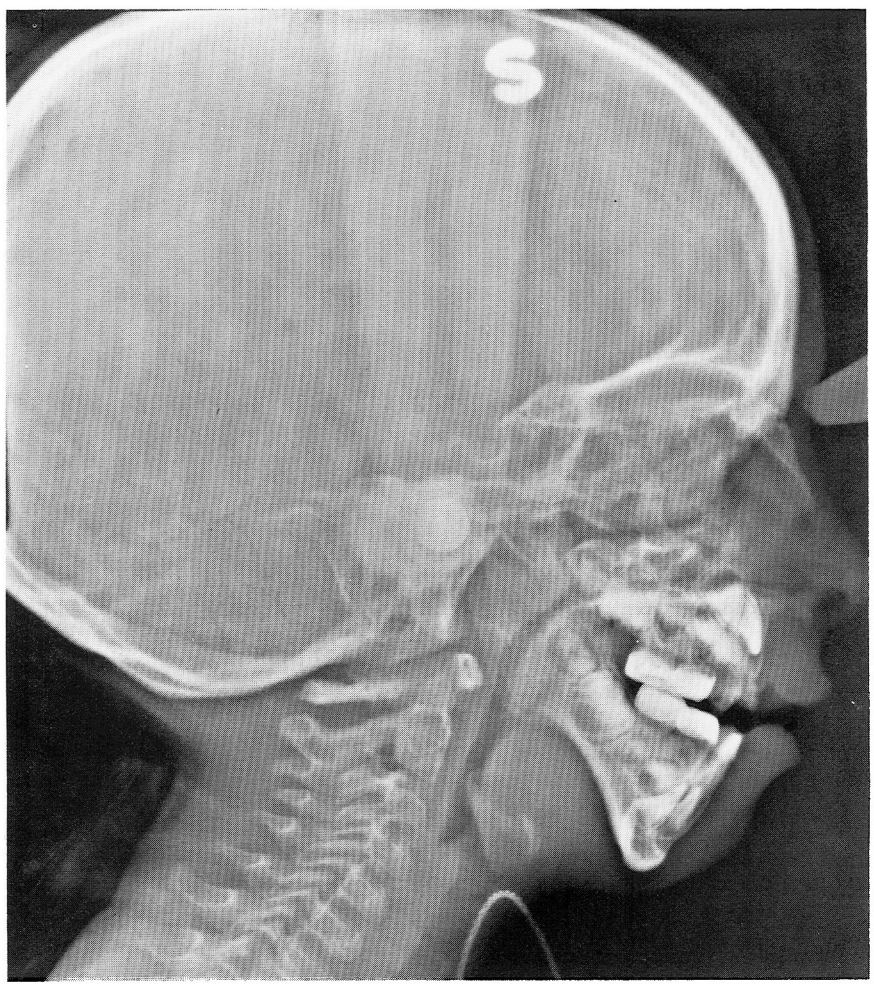

FIGURE 3. MFD patient with adequate palatal length but inconsistent elevation during speech. This lateral cephalometric roentgenogram was taken during sustained production of /s/. Note raised dorsum of tongue, obstructing oral emission of the airstream.

2. Constriction of the airway (from the mandibular and maxillary hypoplasia) may complicate anesthesia and pose the threat of respiratory embarrassment. This particular risk has delayed closure of the palate in several cases in our own clinic and has been the determining factor in the decision *not* to interpose a pharyngeal flap in other cases. Naturally, a restricted airway can also cause problems in the preparation and insertion of a prosthesis. Patients in our series who have undergone facial reconstructive surgery have required preoperative tracheostomy.

3. The narrow, anomalous shape of the nasopharynx and palatal vault further complicates the fitting of a prosthesis.

4. In cases of inadequate or inconsistent elevation of the soft palate, whether in CPI or post-surgical cleft, the prosthesis must often work against the "dead weight" of the velum, at least in the initial phases of treatment. Under such circumstances, the potential hazards to the dentition must be weighed carefully.

Patients with MFD present complex, interrelated treatment needs in the areas of cosmetic appearance, respiratory function, speech, and hearing. The more severely affected may present a number of features which adversely affect speech: (1) significant hearing loss, (2) malocclusion, and (3) abnormal tongue posture. In the presence of such features, palatopharyngeal incompetency may go undetected whether such incompetency is congenital or follows surgical repair of a cleft. In our clinical experience, the presence of hypernasality and nasal emission has been masked in several cases by the peculiar "muffled" voice quality which appears to be related to the position of the body of the tongue in these patients. The curved body of the mandible resulting from hypoplasia of the ramus leads to abnormal height of the dorsum of the tongue at rest and in function (Figure 3). This aberration in tongue posture further impinges upon an already constricted oropharynx. We are currently studying acoustic and radiocephalometric data in an attempt to explain the relationship between the aberrant tongue posture and the abnormal voice quality in these patients.

It is the intent of this report to stimulate clinicians who see patients with MFD to be on the alert for problems of velopharyngeal function even in the absence of overt clefts. Hopefully, those who have had experience in the treatment of such problems will contribute commentary and advice.

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