Minor Oral and Facial Defects in Relatives of Oral Cleft Patients

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Introduction

Clefts of the lip and/or palate frequently show familial predisposition; however, the vast majority of cases do not show clear-cut evidence of a simple genetic mode of transmission.[1,2,3,4,5] The following orofacial morphological aberrations have been reported to occur with greater frequency and consistency in the near kindred of cleft propositi: (1) asymmetry of the nares; (2) high palatal vault; (3) micromaxilla; (4) "V" shaped maxillary arch; (5) supernumerary maxillary incisors; (6) peg-shaped maxillary lateral incisors; (7) congenitally missing anterior teeth; and (8) palatal tori. It has been suggested that these modifications may be a minor expression of congenital orofacial clefts.[1,3,4,6,7,8] If these aberrations do in fact represent a less severe expression of the defect, their inclusion in pedigree analysis might prove of considerable value in defining a simple genetic basis for a larger portion of oral clefts.

Evidence that these minor anomalies do occur more frequently in kindreds of cl/cp* propositi can also be surmised from the work of Mengel who studied seventeen pedigrees which included 1,222 persons. He found that bifid uvula, split gingivae, notched lip or absent teeth were present in thirteen of the seventeen pedigrees.[9] Other anomalies, such as vertical notching of the lip, have been suggested in many studies too numerous to mention.

The basic criticism of all of these studies is that control populations were not used. The prevalence of minor congenital malformations is not known for the population in general. The objective of this study was to determine if the frequencies of various dental and oral malformations were different in a population with a positive history of cp/cl when compared with a population with no known history of cp/cl.

The Sample

The cleft propositi were obtained from the patient files of the Lancaster Cleft Palate Clinic. All records of Caucasian patients residing within Lancaster County and the adjacent five counties were selected. The sample was further screened so that all probands would be between five and eleven years of age at the time of the study. The age restriction was set in an attempt to maximize the number of siblings available for examination. These procedures resulted in a potential sample of 110 study families. Of these sixteen could not be located and twenty refused to participate resulting in a total of seventy-four families. A variety of reasons were given for refusal but generally were related to the inconvenience

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*cl/cp—cleft lip and/or palate

attending participation. In an attempt to increase the sample size, the upper limit was extended to fourteen years and the files rechecked for additional cases. Also, several new families meeting the original criteria had applied to the clinic for care during the first year of the study. These were also added. Particular effort was made on this second selection to obtain multiple case families.

This second selection yielded an additional nineteen cases resulting in a final sample of ninety-three families for study with an average of 4.98 members per family unit. A comparison group of families was derived from patients seen at the clinic for problems other than congenital oral clefts. These included patients seen for speech problems, traumatic defects, Class I and II malocclusions, and general dental work. These index cases were selected from the same general geographic area and were within the same age range as the cleft group. None of these families had a known history of cleft. The final sample of control families numbered eighty-two with an average of 4.85 members per family unit.

Methods

Data from three sources are reported in this paper. These include clinical examinations, color photographs, and dental casts obtained from mother, father, and normal siblings of families which had one or more children with oral clefts, and the same from unaffected families.

The dental casts were measured with three-dimensional orthodontic calipers to determine arch breadth in the first molar and second premolar region, arch length, and palatal height. This technique has been described elsewhere.[10] The casts of the maxillary arches were classified into three categories according to their shape: (1) "U", (2) "V", and (3) square. The presence of palatal tori and prominent midmaxillary sutures was also recorded.

The data from the clinical examinations are based on visual inspection and palpation. Frontal laminagraphs were taken for the evaluation of bony defects in the nasal septum and floor of the nose. These will be the subject of a later report. The figures for congenitally missing and supernumerary teeth are based on clinical examination and observation only. Since both study families and comparison families were examined by the same methods, the biases introduced should be comparable in the two groups.

Results

Data obtained from the study of the dental casts are presented in Tables I and II. Since there were no differences in the prevalences of these variables between male and female, the sexes were pooled for the presentation of the data. The average measurements for maxillary arch breadth and length are shown (Table II). More palatal tori occurred in the control families ($p. < .01$).

Because of the difficulty of establishing objective criteria for the presence of asymmetry of the nares, the color photographs were evaluated by six observers (independently) and a composite evaluation derived. Figure 1 is an example of the type of photograph taken for the evaluation of nasal symmetry. Table III presents frequencies derived in two ways. Row 1 gives the results of requiring agreement among four or more observers and Row 2, the more stringent, required agreement among five or more observers. In neither case is there any appreciable difference between the study and control group.

TABLE I

PALATAL AND JAW CONFIGURATIONS FOR CLEFT AND CONTROL FAMILIES, LANCASTER, PENNSYLVANIA, 1966

	Cleft Families* Total 363	Control Families Total 394	Per Cent of Total Cleft 363	Control 394
Palatal Vault				
Flat	16	22	4.4	5.6
Medium	197	234	54.3	59.4
High	149	136	41.0	34.5
Unclassified	1	1	0.3	0.3
Palatal Suture	167	178	46.0	45.2
Arch Shape				
U	246	275	67.8	69.8
V	67	54	18.5	13.7
Square	48	59	13.2	15.0
Unclassified	2	6	0.6	1.5
Palatal Tori	166	219	45.4	53.3

* excludes clefts

TABLE II

MAXILLARY ARCH MEASUREMENTS FOR CLEFT AND CONTROL POPULATION, LANCASTER, PENNSYLVANIA, 1966

FAMILY	ARCH LENGTH		ARCH BREADTH PM_2		ARCH BREADTH M_1	
	N	MEAN*	N	MEAN	N	MEAN
CLEFT						
Male	60	33.03	56	50.98	60	56.14
Female	72	32.15	72	47.88	72	53.87
CONTROL						
Male	71	33.06	67	50.28	71	56.35
Female	84	33.77	78	47.32	83	53.51

* All measurements are to the nearest 0.1 mm.

TABLE III

ASYMMETRY OF THE NARES, LANCASTER, PENNSYLVANIA, 1966

	Cleft Families 363	Control Families 396	Per Cent of Total Cleft	Control
4-6 Observers Agree	78	83	21.3	21.0
5-6 Observers Agree	49	48	13.4	12.1

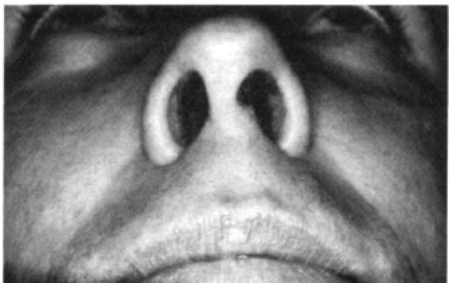

Fig. 1 Sample photograph used for evaluation of nasal symmetry.

Results obtained from clinical examination are shown in Table IV. The frequency of supernumerary, congenitally missing, and hypoplastic teeth did not differ between the two groups. No significant differences were noted between the two groups in the prevalence of alveolar notching and frenum abnormalities (item 3, Table IV). These anomalies occurred in 5.5 per cent of the near kindred of cleft persons and 5.8 per cent of the controls.

Palatal defects, on the other hand, showed considerable differences between the two groups. Included in these defects are submucous clefts of the hard and/or soft palate, bifid uvula, and velopharyngeal inadequacy. Among the cleft group, twenty-two persons (6.0 per cent) exhibited one or more of these defects, whereas six persons (1.5 per cent) of the control group were affected (Table IV).

In addition to the foregoing defects, three individuals with slight notches of the upper lip were seen among the near relatives of the cleft persons. No defects of this nature were noted among the control individuals. Thirteen individuals among five cleft families were found to have bilateral fistulae of the lower lip (Van der Woude syndrome). This syndrome has previously been reported to be due to an autosomal dominant gene.[11,13] The occurrence of this defect in over 5.0 per cent of the cleft families represents a somewhat higher frequency than has previously been reported. It is at present not possible to determine if this represents sampling error or a high gene frequency in the Lancaster sample. Three individuals with submucous clefts of the hard palate were also observed in this group.

TABLE IV

SELECTED ORAL ANOMALIES FROM CLEFT AND CONTROL FAMILIES, LANCASTER, PENNSYLVANIA, 1966

	Cleft Families* Totals 366	Control Families Totals 396	Per Cent of Total Cleft	Per Cent of Total Control
Supernumerary & Congenitally Missing Teeth	15	16	4.1	4.0
Fused, Hypo, or Dysplastic Teeth	13	16	3.6	4.0
Alveolar Notching and Frenum Abnormalities	20	23	5.5	5.8
Defects of the Palate	22	6	6.0	1.5

* excludes clefts

Dicussion

The purpose of this study was to determine if the selected morphological aberrations occurred with greater frequency in the near kindred of cl/cp individuals when compared with a control population. Numerous sources have indicated that malformed palate, micromaxilla, palatal tori, bifid uvula, submucous clefts, notched upper lip, and asymmetry of the nares occurred with greater frequency in parents and siblings of cl/cp offspring.[7,11,12,13,14,15,16,17] If differences could be established, the minor anomalies might be useful in pedigree analysis, completion of the clinical picture of cl/cp, and eventually might be used for the detection of the abnormality in apparently normal individuals with no known history of clefts. With the exception of palatal defects and notches of the lip, our results indicate no significant differences in the prevalence of these aberrations between the two populations. The findings on lateral incisors are similar to those of Woolf and co-workers.[18]

For palatal defects there were striking differences between the two populations. However, there are two major problems. (1) The diagnosis of these defects is not clear-cut. With visual examination and palpation of the palate it is very difficult to make a positive diagnosis. There is also a wide range of normal palatal closure through all stages of submucous cleft to frank cleft palate. (2) These minor palatal defects did occur, with lower frequency, in the control population. Therefore, they are not limited solely to cleft families.

From an epidemiological point of view, if the siblings of the cleft children were intermingled with the siblings of the noncleft children (control), their detection and separation into two distinct populations, with the diagnostic criteria employed, would be impossible without prior knowledge of the cleft history of the family.

Summary and Conclusion

The objectives of this study were to determine if the frequencies of various dental and oral malformations were different in families with one or more children with oral clefts when compared with noncleft families. The data gathered from the plaster casts and color photographs demonstrated that these morphological aberrations occurred as frequently in noncleft families as in cleft families. Therefore, these items would not be useful as diagnostic aids for the stated objectives.

However, there are certain defects (palatal) that might represent minor manifestations of the malformation. The apparent increase in lip and palatal defects associated with oral clefts did not prove consistent or definitive enough to be of much value in pedigree analysis in the relatively small sample of families. Rather careful and detailed clinical study is needed before too much stock is placed in them. At the present time the usefulness of these defects in epidemiological and genetic surveys would appear quite limited.

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