Congenital Malformations and Cleft Lip and Palate

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There is general agreement among authorities (*8, 10, 14, 16*) that a higher incidence of additional congenital malformations exists in the cleft palate population than in the noncleft palate population. However, reports of the incidence of associated congenital malformations in the cleft palate population vary widely. The reported incidence of congenital malformations among cleft palate groups has ranged from 3% to 33% (*3, 4, 11, 12, 15, 16*). These percentages are in marked contrast to the reported incidence of congenital malformations in the general population: approximately 1%.

In a study of 1013 patients with cleft lip and/or cleft palate, Fogh-Andersen (*2*) reported associated structural malformations in more than 10%, with three or more abnormalities occurring occasionally. Loretz, Westmoreland, and Richards (*9*) reported that of 368 cases of cleft palate, 67 had a total of 127 malformations in addition to cleft lip and palate. Kitamura and Kraus (*6*) found that in thirty fetuses with clefts there were 306 anomalies in addition to the cleft.

All types of cleft-associated anomalies do not occur with equal frequency. Loretz, Westmoreland, and Richards (*9*) found that malformations of the bones and joints are the most commonly found malformations in conjunction with cleft lip and/or palate. According to Ivy (*5*) and McKeown and Record (*13*), anomalies of the nervous system occurred most frequently, while Kraus, Kitamura, and Ooe (*7*) reported that brachdactyly and syndactyly were the malformations most frequently seen in cleft lip and/or palate fetuses. Other malformations frequently accompanying cleft palate are: eye abnormalities; supernumerary fingers, toes, and teeth; malformed ears; clubbed hands or feet; tongue abnormalities; microglossia and mandibular micrognathia (*1*).

The sex difference in the occurrence of additional anomalies is unsettled; however, on the basis of current published research it would appear that slightly more girls than boys are born with malformations in addition to cleft palate defects. According to Lutz and Moor (*11*) 62% of the cleft associated defects were in females, while Ivy (*5*) found that slightly more than 50% of the associated anomalies were observed in cleft palate males.

The type of cleft palate defect and frequency with which associated

malformations occur are other variables. In a study of 2,003 cleft patients, Greene and his associates (*3*) reported a total incidence of associated anomalies of 16.5%. With regard to type of cleft, the frequencies were as follows: cleft lip only 8.2%, cleft lip and palate 13.9%, and cleft palate only 27.3%.

Differences in the literature regarding cleft palate associated with other congenital malformations are numerous. Considerably more research is needed in all areas concerning this aspect of cleft palate.

Purpose

The purpose of this survey was to describe the incidence of congenital malformations among individuals with clefts in Oklahoma. Subjects with congenital insufficiency of the palate were also considered. In this report, the term palatal insufficiency (CPI) includes deficiencies in the anterior-posterior dimension of the velum and/or hard palate. Information for this survey was compiled by various members of the interdisciplinary Cleft Palate Team from case records of 100 patients seen at the University of Oklahoma Medical Center between September 5, 1963, and March 26, 1964. The inherent restrictions of birth certificates and clinical records as sources of data are recognized (*3, 5, 9*). However, it is also recognized that clinical records have been the major basic source for epidemiologic studies. Information used in this study includes age, sex, type of cleft palate defect, and associated congenital anomalies. At examination, subjects ranged in age from 3 to 38 years; the median age was 9.06 years.

Results

A summary of the various types of cleft defects found in the subjects is shown in Tables 1, 2, and 3. Cleft of the lip alone occurred in only seven cases (7%) and cleft of the palate alone in 27 (27%). Nine (9%) presented congenital insufficiencies of the palate. Clefts affecting both the lip and palate were found in 57 subjects (57%).

TABLE 1. Distribution of the subjects according to type of cleft or congenital palatal insufficiency (CPI) and age.

age	*CPI*	*type of cleft*			*total*
		lip only	*palate only*	*lip & palate*	
3-0 to 5-11	0	1	6	17	24
6-0 to 8-11	1	3	8	11	23
9-0 to 11-11	3	0	3	10	16
12-0 to 14-11	4	0	3	9	16
15-0 or more	1	3	7	10	21
total	9	7	27	57	100

TABLE 2. Number of subjects presenting associated malformations, according to type of cleft.

type of cleft	*no assoc. malform.*		*with assoc. malform.*		*total*
	N	%	*N*	%	
CPI	3	4.4	6	19.4	9
lip only	5	7.2	2	6.5	7
palate only	19	27.5	8	25.7	27
lip and palate	42	60.9	15	48.4	57
total	69	100.0	31	100.0	100

A summary of the various types of cleft palate defect with and without associated malformations is presented in Table 2. The incidence of cleft palate with associated malformations is 31%. Of the 31 subjects with associated malformations studied, two (6.5%) had cleft lip only. There were only slight differences in the occurrence of associated malformations between the congenital palatal insufficiency and cleft palate only groups; 19.4% and 25.7%, respectively. Associated malformations were most frequently observed in the lip and palate group: 48.4%.

Of the 100 subjects studied, 58 (58%) were males, and 42 (42%) were females. There were only slight sex differences in the occurrence of cleft lip and palate. However, there were twice as many males as females in the sample with bilateral cleft lip and palate.

TABLE 3. Distribution of associated malformations according to type of cleft and sex.

type of cleft	*male*		*female*		*total*	*% with assoc. malform.*
	no assoc. malform.	*with assoc. malform.*	*no assoc. malform.*	*with assoc. malform.*		
CPI	2	3	1	3	9	66.6
lip only					7	28.5
right	1	0	0	0		
left	1	0	1	1		
bilateral	1	0	1	1		
palate only	8	7	11	1	27	29.6
lip and palate					57	26.3
right	4	4	3	3		
left	12	3	9	1		
bilateral	9	3	5	1		
total	38	20	31	11	100	31.0

As shown in Table 3, of the 31 subjects with associated malformations, 20 (64.5%) were males, and 11 (35.5%) were females. There were no sex differences in the occurrence of congenital palatal insufficiency with associated malformations. Cleft lip alone with associated malformations occurred in only two subjects (6.5%): both were females. There were twice as many males in the sample with cleft lip and palate with associated malformations and seven times as many males in the cleft palate group.

The 31 subjects with congenital malformations in addition to cleft lip and palate had a total of 54 malformations. Malformations of the musculoskeletal system were most common, comprising 68% of the 54, followed by cardiac anomalies, which occurred in 11%. The types of malformations appear in Table 4.

Of the 31 cleft palate subjects with associated malformations, 16 (51%) had only one associated malformation, while 15 (49%) had two or more malformations. There were no sex differences in the group of cleft subjects with one associated malformation, half were males and

TABLE 4. Number and types of other malformations in 31 cleft individuals, whether occurring singly or in combination.

malformation	*subtotal*	*total*
musculoskeletal system:		
mandibular micrognathia	11	37
malformed ears	8	
glandular	5	
hypertelorism	4	
skeletal dysplasias	4	
clubfoot	2	
respiratory	2	
syndactylism	1	
cardiac anomalies		6
central nervous system:		3
cerebral palsy	2	
hydrocephaly	1	
genitourinary tract:		4
cryptorchism	2	
hypospadias	1	
hypogenetic nephritis	1	
gastrointestinal tract:		1
imperforate anus	1	
ranula		3
total		54

half were females. However, there was a sex difference in the cleft group presenting two or more malformations. Twelve (80%) of the subjects with two or more malformations were males, and three were females (20%). In the group with three or more associated malformations, all were males.

In the group of 16 subjects with only one additional malformation, three had cleft palate only, two had congenital insufficiency of the palate, and nine had cleft lip and palate. None of the subjects with cleft lip only presented more than one associated malformation as judged by the examiner. There were 10 subjects with two additional malformations; four had cleft lip and palate, three had cleft palate alone, and three had congenital palatal insufficiency. Three subjects presented three additional malformations; two had cleft lip and palate, and one had congenital insufficiency of the palate. There was one subject with four additional malformations and one with five; both subjects had cleft palate only.

With respect to the type of associated malformation, it is notable that the three subjects with central nervous system involvement had at least one other associated malformation. In the group of four with malformations of the genitourinary tract only one had one additional malformation while the remaining three had at least two other additional malformations.

Summary

During a six-month period at the University of Oklahoma Medical Center, 31% of a group of 100 patients were found to have congenital malformations in addition to cleft palate and/or lip. These 31 subjects had a total of 54 other malformations. Of the group, 15 (49%) had two or more associated malformations. Malformations of the musculoskeletal system were most common, comprising 68% of the 54, followed by cardiac anomalies. There were almost twice as many males as females with associated malformations; 74.7% of the associated malformations were in the males. Associated malformations were found among all types of clefts. Some of the subjects in all cleft categories, except that of lip only, presented more than one associated malformation. This survery suggests the need for further research; considerably more information is needed concerning the incidence, including interpopulation differences, and the type of additional congenital malformations in cleft palate.

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