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Original Research Article

Association of other congenital anomalies in children with cleft lip and palate: a prospective hospital based observational study

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ABSTRACT

Background: Cleft lip and palate is a common congenital anomaly affecting approximately 1 in 700 live births in south Asia. It is often associated with syndromes and other malformations but the exact incidence of these in Asians is not known. The present study was carried out to determine the association of other congenital anomalies in children with cleft.
Methods: The study was carried out in the patients attending the Cleft centre of our Hospital. They were examined for other major external congenital malformations and syndrome association. Where ever relevant, appropriate investigations were done.
Results: Of the total of 2367 children examined, 262 (11.06%) had congenital malformations. Among the non-syndromic children, 9% had associated malformations. The commonest was congenital heart disease (1.4%) following by genitourinary and skeletal anomalies. The highest number of anomalies was seen in patients with cleft palate alone (24.89%). 1.4% patients had identifiable syndromes.
Conclusions: The study emphasizes the need for a thorough examination of all children with cleft. The overall lower incidence of syndromic clefts and associated anomalies in present study suggests that other etiological factors may be involved in our country.

Keywords: Anomaly, Cleft, Orofacial, Syndromes

INTRODUCTION

Cleft lip and palate is a common congenital anomaly affecting approximately 1 in 700 - 1000 live births.[1,2] Cleft is believed to be caused by the interaction between genetic and environmental factors.

When environmental factors interact with a genetically susceptible genotype during the early stage of embryonic development clefts occur.[3] Micronutrient deficiency has also been implicated in the causation of cleft in our country.The condition is often associated with other malformations. According to Western literature, 10-20% of children with cleft have additional malformations and approximately 3% are associated with known syndromes. (4) There are over 200 syndromes associated with clefts. The incidence and types of malformations associated vary between studies.

Identification of the associated anomalies is an important aspect of counseling and management.

There is a paucity of data on this from Southeast Asia where the prevalence of clefts is high. The objective of the present study was describing the prevalence and types of other congenital anomalies in children with cleft lip and palate presenting to the Cleft and Craniofacial Centre of a large teaching hospital in South India

METHODS

Study design

This hospital based descriptive study was carried out in a tertiary care teaching hospital in South India.

Inclusion criteria

- Children aged 0-18 years with oro-facial clefts coming to the cleft clinic during the study period were included in the study.

Study procedure

After obtaining informed consent from the parents, the demographic details and cleft description was noted down by the nurse in the cleft clinic. The children were clinically examined by a pediatrician and the presence of external congenital malformations was noted. The children were categorized into known syndromes if they had typical features.

The others were described as non-syndromic. The cardiovascular system was examined for signs of congenital heart disease. Wherever relevant, appropriate investigations were done. ECHO was done by the pediatric cardiologist using Vivid - 7.

Median facial dysplasia syndrome: It is a disorder of the craniofacial region that is characteristic of deficient mid facial structures with the addition of a unilateral or bilateral cleft lip with or without cleft palate.[5]

Velocardiofacial syndrome: is a genetic condition characterized by abnormal pharyngeal arch development that results in defective development of parathyroid glands, thymus and conotruncal region of the heart.[6]

Pierre Robin syndrome: is a clinical trial of micrognathia, glossoptosis, airway obstruction and clefting of the palate.[7]

Crouzon syndrome: is a genetic disorder characterized by the premature fusion of certain skull bones affecting the shape of head and face associated with cleft lip / cleft palate.[8]

Van der Woude syndrome: is a condition that affects the development of the face associated with cleft lip / palate, depressions near the center of the lower lip.[9]

Seckel syndrome: is a autosomal recessive disorder characterized by proportionate dwarfism, bird headed appearance, mental retardation and occasionally cleft palate.[10]

Treacher Collins syndrome: is a condition that affects the development of bones and other tissues of the face associated with cleft palate, eyelid coloboma, small deformed ears and hearing loss.[11]

Statistical analysis

Data was analyzed for the distribution of each abnormality and its association with the type of cleft. To standardize the assessment, authors segregated the anomalies by the affected system

RESULTS

During the study period 2367 cleft patients were examined over four years. Of these 171 (7.2%) had cleft lip alone, 282 (11.9%) had cleft palate alone and 1914 (80.8%) had cleft lip and palate. Overall, 262 (11.06%) had other congenital malformations and 34 patients had syndromic association (1.4%) (Table 1). 13 children had Pierre Robin Syndrome, 9 children had Median Dysplasia Syndrome and two Velocardiaofacial Syndrome.

Associated malformations

On excluding the syndromic patients, authors noted that 228/2333 patients (9.7% of non-syndromic patients) had associated anomalies (Table 2).

Table 1: Various syndromes identified in the children.

Syndrome	Cleft lip (171)	Cleft palate (282)	Cleft lip and palate (1914)	Total (2367)
Media dysplasia syndrome	2	2	5	9
Velocardiofacial syndrome	0	2	0	2
Pierre Robin syndrome	0	13	0	13
Crouzon syndrome	0	0	1	1
Ectodermal dydsplasia	0	2	2	4
Popliteal pterygium syndrome	0	0	1	1
Amniotic constriction ring	1	0	0	1
Wander Wood syndrome	0	0	1	1
Seckel syndrome	0	0	1	1
Treacher Collin syndrome	0	1	0	1
Total	3	20	11	34

Table 2: Types of congenital anomalies and their distribution.

	Cleft lip 171	Cleft palate 282	Cleft lip and palate 1914	Tessier cleft type-IV	Tessier cleft type-VII	Tessier cleft type - X	Total 2367
Cardiac	2 (1)	6 (3)	26 (2)	-	-	-	34 (6)
Genito urinary gus	6 (1)	4	14	-	-	-	24 (1)
Skeletal	7	6 (2)	17 (4)	-	-	-	30 (6)
Hands	0	0	6	-	-	-	6
Skull	4 (2)	4 (1)	5 (1)	-	-	-	13 (4)
Foot	0	0	1	-	-	-	1
Mandible	1	22 (13)	7	-	-	-	30 (13)
Ear	2	14 (3)	26 (1)	1	2	-	45 (4)
Eye	10	7 (1)	23 (3)	7	-	2	49 (4)
Skin	2 (1)	3 (1)	6	-	-	-	11 (2)
CNS	3	4 (3)	12 (2)	-	-	-	19 (5)
Total	37 (5)	70 (27)	143 (13)	8	2	2	262 (45)

Within brackets are the numbers of children with these defects who had identifiable syndromes.

34 (1.4%) patients had cardiac anomalies. 24 patients had anomalies of the genitourinary tract. Ear anomalies, although high in number were mostly pre-auricular tags. The highest number of anomalies was seen in patients with cleft palate alone 70/282 (24.89). Next in frequency were patients with cleft lip alone 70/282(24:8%).Anomalies of the mandible like retrognathia were commonly seen in patients with cleft palate alone. Tessier cleft type 4, 7 and 10 accounted for about 12 patients in the study. Cardiac anomalies in association with cleft: 1.4% of cleft patients had cardiac anomalies (Table 3). Ventricular septal defect was the commonest defect identified (10 patients). Atrial septal defect was seen in 6 patients. Here again the highest numbers were seen among patients with isolated cleft palate (2%).

Table 3: Different types of cardiac anomalies seen among the patients.

Cardiac disorder	Cleft lip (1710)	Cleft palate (282)	Cleft lip and palate (1914)	Total (2367)
VSD	2	0	8	10
ASD	0	2	4	6
VSD and ASD	0	0	1	1
Dextrocardia	0	0	2	2
TR	0	0	1	1
Tubercular constructive pericarditis	0	1	0	1
MVP	0	0	3	3
Dilated cardiomyopagthy	0	0	2	2
PDA	0	0	2	2
Asd and Ps	0	1	0	1
VSD and Ps	0	1	0	1
Bicuspid aortic valve	0	0	1	1
Cardiac surgery details not known	0	3	0	3
Total	2 (1%)	6 (2%)	26 (1.3%)	34

DISCUSSION

Authors studied the incidence and types of associated malformation in 2367 children with cleft lip and palate treated at our center. The commonest type was cleft lip and palate (80.8%) followed by cleft palate (11.9%). Overall, 1.4% of children with cleft had identifiable syndromes On excluding the 34 syndrome patients 228/2333 (9.7%) had congenital malformations.

Associated anomalies were seen more frequently in infants with isolated cleft palate (24%) than in the other types. 21% of children with cleft lip alone and 7% of

children with cleft lip and palate had associated anomalies. In present study the commonest associated anomaly involved the eye (18.7%) followed by preauricular tag (17.1%). Among the major congenital anomalies, congenital heart disease were the commonest (12.9% of the total anomalies) followed by genitourinary malformations (9.1%). The overall incidence of congenital heart disease in present study population was 1% and among the children with cleft palate alone it was 2% The relative incidence of the type of cleft varies in different studies.[2,4] The reported incidence of associated anomalies varies between different studies. In western literature the incidence reported is 20 to 36 %, but the number of cases studied has been much smaller in all these reports.[12-14]

The lower incidence in present study could be due the larger numbers studied and the higher prevalence of non-syndromic clefts. This, in turn, suggest that the etiological factors may be different in our country. The types of anomalies noted were comparable with those noted in other studies as in Liang et al and Milerad et al.[12,15] The incidence of congenital cardiac anomalies in the general population is less than 0.1%, while among our cleft children it was 10 times higher at 1%. Liang et al demonstrated an overall prevalence of congenital heart diseases to be 5.4% while Milerad et al noted an incidence of 3.75%.[16] The lower prevalence in present study is probably due to the higher prevalence of non-syndromic clefts in present study population

CONCLUSION

The prevalence of other malformations in children with cleft was 11.9% and it was highest is those with cleft palate alone (24.89%). Among the children studied 1.4% patients had identifiable syndromes. This emphasizes the need for a thorough physical examination and investigations of all children presenting with cleft lip and palate.

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