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Original Research Article

A cross-sectional study of cardiac anomalies among children with orofacial cleft - role of echocardiography

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ABSTRACT

Background: Orofacial cleft is one of the most common congenital facial abnormalities. Congenital heart disease (CHD) has been reported in up to 15% of the patients with orofacial clefts. Clinical cardiac examinations may sometime miss cardiac anomalies in children with orofacial clefts. The aim of our study is to find out CHD by echocardiography and to correlate this with clinical examination findings in children with orofacial clefts.
Methods: In this cross-sectional study, 510 patients with orofacial clefts aged 1 month to 18 years were enrolled. History and clinical examination were performed and data entered in a pre-validated proforma. The type of orofacial cleft and syndromic features was specifically noted. Echocardiography was performed for all patients by a paediatric cardiologist and the clinical cardiovascular findings were correlated with the echocardiogram.
Results: The commonest deformity was cleft lip and palate (69%), followed by isolated cleft palate (20.4%), and isolated cleft lip (10.6%). In our study population, 87.5% were non-syndromic and 12.5% were syndromic; 21.9% had cardiac anomaly, of which in 7.8% no cardiac defect was detected during systemic cardiovascular examination but their echocardiogram showed cardiac abnormality. This was statistically significant ($p<0.000$). Atrial Septal Defect was the commonest anomaly in both the groups. Pierre Robin Syndrome was found to be the commonest syndrome.
Conclusions: The high prevalence of CHD among children with orofacial clefts in this study justifies the need for screening echocardiography because many times cardiac anomalies might not be detected in routine systemic examination.

Keywords: Congenital heart disease, Echocardiography, Orofacial cleft

INTRODUCTION

Orofacial cleft is a common birth defect that occurs either isolated or in association with other malformations in approximately 1 in every 600 live births.[1,2] The distribution of orofacial clefts is as follows: 15% cleft lip, 45% cleft lip and palate, and 40% isolated cleft palate.[3] In India alone the number of infants with orofacial cleft is 28,600. This which means that 78 affected infants are born every day, or 3 infants with clefts are born every hour.[4] Orofacial clefts can be associated with other medical conditions like skeletal anomalies, central nervous system problems, respiratory problems, speech impairment, and congenital heart disease (CHD).[5-7] Children with orofacial clefts can be non-syndromic or syndromic. The majority of orofacial clefts are non-

syndromic. Approximately 36% of all cases with orofacial clefts are associated with known syndromes.[5] The high prevalence of malformations underlines the need for systematic examination and thorough investigation in these children.

Congenital heart disease (CHD) has been reported in up to 15% of the patients with orofacial clefts.[8] The association of orofacial cleft and CHD varies according to the study population. The prevalence of CHD varies depending on the duration and intensity of case finding and the sensitivity of the diagnostic technique used. The use of 2-D echocardiography has helped in diagnosing even very small defects and prevalence rates have increased as diagnosis has been enhanced. Even though cardiac examination of children with orofacial clefts is clinically insignificant echocardiography has picked up many cardiac anomalies.[9] This study was undertaken to estimate the incidence of CHD by echocardiography in all children with orofacial clefts and to verify clinical examination findings with echocardiography.

METHODS

After obtaining ethical committee approval, all cases enrolled in the outpatient and inpatient departments of Sri Ramachandra Medical College and Research Institute, Chennai, during the study period of November 2013 to October 2015, whose parents gave consent, were included in the study.

The number of children with orofacial clefts who were enrolled was 510 and the age group ranged from 1 month till 18 years of age. Detailed history, general, and systemic examination were performed and recorded in a predesigned proforma. Any child with dysmorphic features and associated malformations were checked for known syndromes using Smiths's Recognizable patterns of human malformation-7th edition. A thorough cardiovascular systemic examination was done for all the patients. All children with orofacial clefts, irrespective of whether they were clinically normal or abnormal, underwent 2-D echocardiography to detect cardiac defects by the same paediatric cardiologist. Echocardiography was done with a "GE-Vivid V6, echo machine using 6 Megahertz Sector Paediatric Probe along with Doppler imaging and M-Mode imaging". Trivial mitral regurgitation and tricuspid regurgitation (MR/TR) were ignored and any defect in the inter-atrial septum equal to or lower than 3mm was considered as normal. The data collected was tabulated and the results were analysed. Statistical analysis was done by SPSS version 17 software. Statistical test of significance was applied where ever indicated.

RESULTS

A total of 510 cases were studied. The male to female ratio was 1.3:1. The majority of children (38%) belonged to the age group of 1 to 11 months. The commonest deformity was cleft lip and palate (69%), followed by isolated cleft palate (20.4%), and isolated cleft lip (10.6%) (see Table 1). Of the total patients, 446 (87.5%) children were non-syndromic and 64 (12.5%) were syndromic.

Table 1: Types of orofacial clefts in the study group.

Types of orofacial clefts	Frequency	%
Cleft lip	54	10.6
Cleft palate	104	20.4
Cleft lip/palate	352	69
Total	510	100

In 112 children (21.9%) cardiac anomaly was picked up by echocardiography, of which 40 children (7.8%) had normal cardiovascular examination (Table 2).

Table 2: Correlation between systemic examination and echocardiography findings in the study group.

	SE normal, echo-normal	SE normal, echo-abnormal	SE abnormal, echo-abnormal	SE abnormal, echo-normal	Total
Non-syndromic	359 (80.5%)	27 (6.1%)	57 (12.8%)	3 (0.7%)	446 (87.4%)
Syndromic	29 (45.3%)	13 (20.4%)	22 (34.3%)	0 (0%)	64 (12.5%)
Total	388 (76.3%)	40 (7.8%)	79 (15.4%)	3 (0.5%)	510 (100%)

SE-Systemic examination

This was statistically significant (p<0.000). During systemic examination, 27 children (6.1%) in the non-syndromic group and 13 children (20.4%) in the syndromic group did not show any cardiac abnormality but the cardiac defect was picked up by echocardiography. In non-syndromic children with cardiac anomalies, 35 (7.8%) were Atrial Septal Defect (ASD), 26 (5.8%) Ventricular Septal Defect (VSD), 9 (2%) Patent Ductus Arteriosus (PDA), and 6 (1.3%) Tetralogy of Fallot (TOF). In syndromic children, there were 10 (15.6%) ASD, 6 (9.3%) MR, 5 (7.8%) VSD, and 4 (6.2%) PDA (Table 3). In present study, 41 children (64%) had Pierre Robin Syndrome which was the commonest, followed by 6 (9.3%) Velocardio facial

syndrome, 2 (3.1%) Craniofacial cleft, 2 (3.1%) Facial bipartition, 1 (1.5%) Binder's syndrome, 1 (1.5%) Crouzon Syndrome, and the rest were unspecified syndromes (Figure 1).

Table 3: Echocardiography findings in the study group.

Cardiac defect	Non-syndromic	Syndromic	Total
ASD	35 (7.8%)	10 (15.6%)	45 (8.8%)
VSD	26 (5.8%)	5 (7.8%)	31 (6%)
PDA	9 (2%)	4 (6.2%)	13 (2.5%)
TOF	6 (1.3%)	0 (0%)	6 (1.1%)
MR	2 (0.4%)	6 (9.3%)	8 (1.5%)
MV PROLAPSE	2 (0.4%)	1 (1.5%)	3 (0.5%)
COA	1 (0.2%)	1 (1.5%)	2 (0.3%)
Mild pulmonary	1 (0.2%)	0 (0%)	1 (0.2%)
ASD/VSD/PDA	0 (0%)	1 (1.5%)	1 (0.2%)
ASD/VSD/PS/PR	0 (0%)	1 (1.5%)	1 (0.2%)
TR/MR	1 (0.2%)	0 (0%)	1 (0.2%)
Total	83 (18.6%)	29 (45.3%)	112(21.9%)

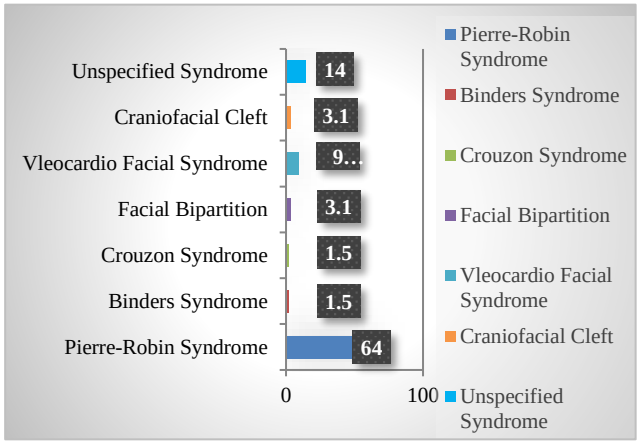

Figure 1: Syndromes in orofacial cleft patients in the study group.

DISCUSSION

In our study population, out of 510 cases of orofacial clefts, the common type of cleft deformity was cleft lip and palate (69%), followed by cleft palate (20.4%), and then cleft lip (10.6%). Other studies showed incidence of 24% of cleft lip and palate, 60% of cleft palate, and 16% of cleft lip.[10]

Out of 510 children, the majority, 446 (87.5%) were non-syndromic and 64 (12.5%) were syndromic. In comparison, the study done by Murthy et al showed that 70% of children were non-syndromic and 30% children were syndromic.[11] Out of 64 syndromic children with orofacial clefts the commonest syndrome in our study was Pierre Robin Syndrome which contributed to 41 (64%), followed by Velocardio facial syndrome 6 (9.3%), Craniofacial cleft 2 (3.1%), Facial bipartition 2(3.1%), Binder's syndrome 1 (1.5%), Crouzon Syndrome 1 (1.5%), and 10 (15.6%) unspecified syndrome. In a study done by Venkatesh, the common syndromes were Van der Woude syndrome, Median facial dysplasia syndrome, and Pierre Robin Syndrome.[5]

In our study 112 (21.9%) children had congenital heart disease whereas a study done by Nancy Geiss et al and Shafi et al showed the overall prevalence of congenital heart disease in children with orofacial cleft from 6.7% to 15%.[6,8,11] Present study showed a higher incidence of cardiac anomalies possibly because the study was conducted in a tertiary care referral hospital.

Out of 510 children, in 388 (76.3%) children, systemic examination as well as echocardiography findings were normal. In 40 (7.8%) children, systemic examination was normal whereas echocardiogram was abnormal, and in 79 (15.4%) children both systemic examination and echocardiogram were abnormal. The study shows that in some children (7.8%) systemic examination was normal, but cardiac anomaly was picked up by echocardiography. This shows that all children with orofacial clefts should undergo compulsory echocardiography to rule out cardiac anomaly even if the systemic examination is inconclusive. This difference was statistically significant ($p<0.000$).

An article by Roelandt evaluates the performance of the traditional physical examination, indicating that 30% major and 65% minor pathologies are missed.[9] Correct identification of heart sounds and murmurs ranged between 20% and 50%, respectively. A physical examination integrated with echocardiography increases its diagnostic yield by >50%, adds greater accuracy and unsuspected but clinically relevant abnormalities are

diagnosed in 20% of patients. The common cardiac anomaly in both the groups (non-syndromic and syndromic) in our study was ASD, 45 (23.4%). In the study by Sun et al the commonest cardiac defect was ASD which was found in around 39.7% patients.[12] Other studies done by Shafi et al and Liang et al also showed similar findings.[8,13]

CONCLUSION

Owing to the high incidence of cardiac anomalies in children with orofacial clefts, echocardiography is essential for evaluating all children with orofacial clefts because cardiac anomalies are highly likely to be missed by only systemic clinical examination. Since children with orofacial clefts require surgical correction, which involves general anaesthesia, it is clinically important to detect all cardiac defects.

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