Original Research Article

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Association of congenital heart diseases with extracardiac anomalies

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ABSTRACT

Background: Congenital heart diseases are one of the main causes of death among the congenital malformations. An extracardiac malformation further increases the risk of morbidity and mortality to these patients, in addition to risks of surgical correction. So here comes the importance of extracardiac malformation.

Methods: This prospective observational study was conducted at Neonatology Section, Department of Pediatrics, Jawaharlal Nehru Medical College, Aligarh Muslim University, Aligarh. The study was conducted over a period of 2 years (from January 2015 to December 2016). Newborns with congenital heart disease were examined for extracardiac defects.

Results: This hospital based cross-sectional study found that there were 121 cases of congenital heart diseases among 11030 live births making an incidence of 1.09% Out of these 73.5% were acyanotic heart diseases and 26.5% were cyanotic heart disease. Extracardiac anomalies were found in 38 (31.5%) cases.

Conclusions: Association of Congenital heart disease with extracardiac anomalies is well known. Syndromes are most commonly associated.

Keywords: Congenital cardiac malformation, Etiology, Extracardiac malformations, Lives infants, Syndromes

INTRODUCTION

Congenital heart disease is one of the most common types of birth defect, affecting 10-20 per 1,000 live births in the North India. The most common type of heart defect is a ventricular septal defect. Most of the congenital heart diseases are idiopathic. However, there are some known risk factors in mother such as rubella during pregnancy, poorly controlled diabetes, consanguineous marriage and drugs. Congenital heart diseases are one of the main causes of death among the congenital malformations. An extra-cardiac malformation further increases the risk of morbidity and mortality to these patients, in addition to risks of surgical correction. Further, such babies may need surgical procedures or

intensive care, whatever may be the congenital heart disease. So here comes the importance of extracardiac malformation. Thus, the aim and objective of this study was to the association between congenital heart disease and extracardiac malformation. Data for the prevalence and pattern of these defects in Indian children with congenital heart diseases are less.

METHODS

This prospective observational study was conducted at Neonatology Section, Department of Pediatrics, Jawaharlal Nehru Medical College, Aligarh Muslim University, Aligarh. The study was conducted over a period of 2 years (from January 2015 to December 2016).

Newborns with congenital heart disease were examined for extracardiac defects. The CHDs were diagnosed on basis of complete history, general physical and examination, cardiovascular chest X-ray, electrocardiography (ECG) and confirmed by 2Dechocardiography. The extracardiac anomalies were diagnosed and further investigations like ultrasound computed tomography (abdomen/skull), audiometry, ophthalmologic evaluation, thyroid hormone profile and karyotyping were done. A malformation or an anomaly was defined as a permanent change produced by an intrinsic abnormality of development in a body structure during prenatal life.

Malformations were subdivided into major and minor malformations according to the guidelines by EUROCAT (European Registers of Congenital Chromosomal Anomalies and Twins).⁴ Anomalies were classified as major and minor according to its medical or surgical or

serious cosmetic significance. Congenital heart diseases associated with extracardiac anomalies were included in the study. Extracardiac anomalies were grouped into 8 groups (identified syndromes and rest system wise group).

For the purposes of study, a syndrome was defined as a recognized pattern of congenital abnormalities whose unique combination of features sets it apart from other patterns. Statistical analysis was carried out by SPSS 23.

RESULTS

This hospital based cross-sectional study found that there were 121 cases of congenital heart diseases among 11030 live births making an incidence of 1.09% Out of these 73.5% were acyanotic heart diseases and 26.5% were cyanotic heart disease. Extracardiac anomalies were found in 38 (31.5%) cases.

Table 1: Distribution of congenital heart diseases.

Congenital Heart disease	Without extracardiac anomalies	With extracardiac anomalies	Total
Acyanotic	58 (65.1%)	31 (34.9%)	89 (73.5%)
Cyanotic	25 (78.1%)	7 (21.9%)	32 (26.5%)
Total	83 (68.5%)	38 (31.5%)	121 (100%)

Table 2: Distribution of extracardiac anomalies in congenital heart diseases.

	Congenital heart disease			
Extracardiac anomaly	Total	Present	Absent	P value
Identified syndrome	36	20 (55.5%)	16 (44.5%)	0.0001
Musculoskeletal	35	6 (17.1%)	29 (82.8%)	0.087
Central nervous system	24	4 (16.6%)	20 (83.4%)	0.161
Genitourinary system	17	3 (17.64%)	14 (82.36%)	0.294
Gastrointestinal system	8	2 (25%)	6 (75%)	0.828
Respiratory system	3	1 (33%)	2 (67%)	0.847
Other (craniofacial, eyes etc)	11	2 (18%)	9 (82%)	0.434

Table 3: Distribution of congenital heart diseases in identified syndromes.

Syndromes	Total number	Extracardiac anomalies
Downs syndrome	23	10 (43.4%)
Edward syndrome	4	3 (75%)
Patau syndrome	2	2 (100%)
Cornelia de lange syndrome	2	2 (100%)
Pierre robin syndrome	3	1 (33%)
Holt oram syndrome	1	1 (100%)
Treacher collins syndrome	1	1 (100%)

Table 2 shows that extracardiac anomalies system wise and identified syndromes. Identified syndromes were present in 20 (55%) cases of congenital heart disease which was a statistically significant association. (P <0.0001). Musculoskeletal, central nervous system,

genitourinary system, gastrointestinal system and respiratory system anomalies were present in 6 (17.1%), 4 (16.6%), 3 (17.64%), 2 (25%) and 1 (33%) cases of congenital heart diseases. But these are not statistically significant.

Table 3 shows distribution of syndromes in congenital heart diseases. 43.4% cases of Down syndrome are associated with congenital heart disease. The most common single cardiac lesion was ventricular septal defect and patent ductus arteriosus. 75% cases of Edward syndrome and all cases of Patau syndrome associated with congenital heart diseases. Other syndrome like Cornelia de Lange, Holt Oram, Treacher-Collins and Pierre Robin are also associated with heart diseases.

DISCUSSION

In this study, we found the prevalence of congenital heart disease 10.9 per 1000 live births. Bhat et al conducted study in Uttarakhand found prevalence 8.54 per 1000 live birth similar to our study. Other studies in north India like Kapoor and Bharadwaj et al found higher prevalence 26.4 and 19.14 per 1000 live birth.^{2,3} In the present study incidence of extracardiac birth defect was 31.5%. Identified syndromes were most common significantly associated with congenital heart diseases. The most common anomalies were seen central musculoskeletal, nervous system genitourinary, however we did not find any significant association with congenital heart diseases. Abdullah et al in Saudi Arabia found incidence of 28.5% extracardiac anomalies.5 A significant association with congenital cardiac malformation was found between identified syndrome, central nervous system, genitourinary and musculoskeletal system and extracardiac anomalies. In a study by Julian and Farrú in Chile found that extracardiac malformation children with congenital heart disease was 31.9% of the patients, 22.7% of which constituted part of some syndrome.⁶ The authors did not identify any specific association except in classic syndromes. The most frequent anomalies were observed in the gastrointestinal, musculoskeletal and genitourinary tracts. This high number of cases is due to high pick up rate in our setting. Ferencz et al conducted study of CHD among live births in the US.7 Extracardiac anomalies were observed among 26.8% of the individuals with CHD, 93% of which were associated with chromosomal anomalies or other syndromes. Central nervous system malformations, gastrointestinal and urinary tract defects were more frequent among the patients with CHD. Pradat retrospectively observed the data of children with major CHD in Sweden.⁷ Extracardiac malformation was identified in 25.7% patients after excluding 15% of the children with chromosomal anomalies. Bosi et al retrospectively evaluated CHD in Italy and found 24% of patients had extracardiac malformations.9 Chromosomal anomalies were found in 9.1% of the newborns. The genitourinary system was the most frequently affected. Karande et al, Miller et al and Dilber and Malcic found incidence of extracardiac anomalies 17.5%, 13.5% and 14.5% respectively which was lower than our study. 10-12 Down syndrome is the most common identified syndrome in our study. The prevalence of congenital heart disease in down syndrome was similar to the findings from other studies, which have been between 44% and 62%. 13-16 The actual incidence of congenital heart disease with or without extracardiac anomalies, exactly can be determined if we have included all live births, fetal deaths and spontaneous and induced abortions. However, in this study, fetal deaths and abortions were not studied. Association of Congenital heart disease with extracardiac anomalies is well known. Syndromes are most commonly associated. The pediatrician must be aware that extracardiac anomalies are common with congenital heart defects. Association of congenital heart diseases with chromosomal anomalies helps in surgical management of anomalies, early intervention for developmental delay, rehabilitation and genetic counselling for future pregnancies. Therefore, it is necessary to perform routine radiological and biochemical screening in such cases.

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