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Original Research Article

A Study of Prevalence of Dysmorphic Features in Children with Congenital Heart Disease and their Association with type of CHD

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Abstract:

Introduction: Congenital heart disease (CHD) accounts to 28 percent of major congenital anomalies, accounting it to be the most common among them. Children with Congenital heart disease often have various dysmorphic features.

Objectives: To study the prevalence of dysmorphology in children with CHD and their association between cyanotic and acyanotic types of CHD.

Methods: A cross sectional observational study was conducted on 127 children with congenital heart disease admitted in JLN Medical College during the study period. Various dysmorphologies were noted. Prevalence of dysmorphic features and if any dysmorphic feature is significantly associated with the type of CHD (cyanotic or acyanotic) were statistically determined.

Result: 75.5% of the sample had some form of dysmorphism, of which the most common was face dysmorphism. Hypertelorism and total face dysmorphism were significantly associated with cyanotic CHD.

Conclusion: Dysmorphisms when found in any child warrants the clinician to look for any congenital cardiac anomalies. This helps in early diagnosis and thus, better survival of such children. Hypertelorism and face dysmorphism if present, can indicate a higher probability of cyanotic CHD in a child.

Keywords: Congenital heart disease, Dysmorphism, Hypertelorism.

Introduction

Congenital heart disease is defined as gross structural abnormality of the heart or intra thoracic great vessels. (1) CHD accounts to 28 percent of major congenital anomalies, accounting it to be the most

Table 1: Proportion of various dysmorphisms

Dysmorphic feature	Frequency	Percentage	95% CI
Skull shape anomaly	18	14.17%	8.84 – 21.74
Skull size anomaly	12	9.45%	5.2 – 16.26
Frontal bossing	3	2.36%	0.61 – 7.27
Total Skull anomaly	**29**	**22.83%**	**16.05 – 31.29**
Depressed nasal bridge	35	27.56%	20.19 – 36.32
Hypertelorism	22	17.32%	11.4 – 25.27
Eye anomaly	19	14.96%	9.47 – 22.63
Ear anomaly	37	29.13%	21.58 – 37.96
Cleft lip	5	3.94%	1.46 – 9.41
Cleft palate	4	3.15%	1.01 – 8.36
Retrognathia	20	15.75%	10.11 – 23.52
Micrognathia	15	11.81%	6.98 – 19.03
Total Face dysmorphism	**79**	**62.2%**	**53.13 – 70.52**
Neck dysmorphism	**19**	**14.96%**	**9.47 – 22.63**
Genito-urinary dysmorphism	**7**	**5.51%**	**2.44 – 11.44**
Limb dysmorphism	**38**	**29.92%**	**22.29 – 38.79**

common among them. [2] congenital heart disease has a prevalence of 5-10 per 1000 live births.[3] Prevalence of CHD in India as per recent studies is 8.5-13.6 per 1000 children [4][5]. Extracardiac malformations are associated with 30% of all congenital heart diseases. David W. Smith, a pediatrician and clinical geneticist, coined the term dysmorphology in the 1960s.The Greek words “dys” (disordered/abnormal) and “morph” (shape, form) were combined to term dysmorphism. Neural crest cells play pivotal role in formation of the craniofacial region and also conotruncal endocardial cushions, which septate the outflow tract into pulmonary and aortic channels. Hence any abnormality in functions of neural crest cell can results in craniofacial anomalies along with cardiac defects [1].

Objectives:

- To study the prevalence of dysmorphology in children with CHD
- To study the association between dysmorphology and type of CHD (cyanotic and acyanotic)

Methods: A cross-sectional observational study was done on 127 cases diagnosed with congenital heart disease in age group newborn to 18 years, who were admitted in NICU, PICU and general ward of JLN Medical college Ajmer from December 1st 2021 to November 31st, 2022.All cases were examined from head to toe to check if any dysmorphic features were present.

Inclusion Criteria

Patients aged between 0-18years, with CHD, proven by 2D Echo ,admitted to the department of paediatrics, JLN Medical College, Ajmer.

Exclusion Criteria

- Children whose parents did not give consent were excluded from the study.

Sample Size

Sampling was done as per convenience.127 children admitted with congenital heart disease during the study period were enrolled into the study.

Data Analysis: The data collected were analyzed using proportion and 95% CI of proportion, chi-square test, two-tailed Fisher’s exact test and t test. P >0.05 was considered statistically insignificant and P<0.05 statistically significant.

Table 2: Association of dysmorphic features and type of CHD

ASSOCIATION OF DYSMORPHIC FEATURES AND TYPE OF CHD					
Dysmorphic feature		**Cyanotic CHD**	**Acyanotic CHD**	**Chi square value**	**'p' value**
Skull shape anomaly	Yes	4 (22.2%)	14 (77.8%)	--*	0.741
	No	19 (17.4%)	90 (82.6%)		
Skull size anomaly	Yes	0	12 (100%)	--*	0.122
	No	23 (20%)	92 (80%)		
Frontal bossing	Yes	1 (33.3%)	2 (66.7%)	--*	0.454
	No	22 (17.7%)	102 (82.3%)		
Total skull anomaly	Yes	4 (13.8%)	25 (86.2%)	0.472	0.492
	No	19 (19.4%)	79 (80.6%)		
Depressed nasal bridge	Yes	8 (22.9%)	27 (77.1%)	0.734	0.392
	No	15 (16.3%)	77 (83.7%)		
Hypertelorism	Yes	8 (36.4%)	14 (63.6%)	--*	**0.028**
	No	15 (14.3%)	90 (85.7%)		
Eye anomaly	Yes	3 (15.8%)	16 (84.2%)	--*	1.000
	No	20 (18.5%)	88 (81.5%)		
Ear anomaly	Yes	7 (18.9%)	30 (81.1%)	0.023	0.879
	No	16 (17.8%)	74 (82.2%)		
Cleft lip	Yes	1 (20%)	4 (80%)	--*	1.000
	No	22 (18%)	100 (82%)		
Cleft palate	Yes	2 (50%)	2 (50%)	--*	0.150
	No	21 (17.1%)	102 (82.9%)		
Retrognathia	Yes	3 (15%)	17 (85%)	--*	1.000
	No	20 (18.7%)	87 (81.3%)		
Micrognathia	Yes	4 (26.7%)	11 (73.3%)	--*	0.472
	No	19 (17%)	93 (83%)		
Total face dysmorphism	Yes	19 (24.1%)	60 (75.9%)	4.974	**0.026**
	No	4 (8.3%)	44 (91.7%)		
Neck dysmorphism	Yes	1 (5.3%)	18 (94.7%)	--*	0.194
	No	22 (20.4%)	86 (79.6%)		
Genital dysmorphism	Yes	1 (14.3%)	6 (85.7%)	--*	1.000
	No	22 (18.3%)	98 (81.7%)		
Limb deformity	Yes	5 (13.2%)	33 (86.8%)	0.897	0.344
	No	18 (20.2%)	71 (79.8%)		
{*Fisher's exact test}					

Result

In our study, out of 127 children with CHD, 96(75.5%) had some form of dysmorphism, out of which face dysmorphisms were the most frequent dysmorphism (62.2%), followed by limb dysmorphisms (29.92 %). 22.83 % of the children had skull anomalies,14.96% had neck dysmorphisms and 5.51% had Genito-urinary dysmorphisms. In our study, hypertelorism and total face dysmorphisms were found significantly higher in cyanotic CHD compared to acyanotic CHD with p values 0.028 and 0.026 respectively. No other significant association was noticed between dysmorphic features in children with CHD and type of CHD.

Discussion

This study was conducted to study the prevalence of dysmorphic features in children with CHD and to study association

of dysmorphology with the type of CHD. In the current study a high prevalence (75.5%) of dysmorphology was seen in the children with CHD. Face dysmorphisms were the most frequent dysmorphism (62.2%), followed by limb dysmorphisms (29.92 %). 22.83% of the children had skull anomalies, 14.96% had neck dysmorphisms and 5.51% had Genito-urinary dysmorphisms.

In the study by Settin A et al it was found that 34.8% of cases with CHD had dysmorphic features (OR= 2.6), most common of which were ear anomalies. Others were cleft lip, polydactyly and cleft palate .[6]

In a case control study, Egbe et al found significant association between CHD and extra cardiac congenital anomaly (OR= .01, CI: 1.97-2.14). Malformations seen associated with CHD in the study were craniofacial, respiratory, and genitourinary malformations and dysmorphisms found associated were cleft lip/palate, micrognathia, ear anomalies and hypospadias. But the study didn't find any association between CHD and limb anomalies.[7] Prasad C et al found that about 30% of congenital heart diseases were associated with extracardiac malformations.[8]

In a study conducted by Dharmedar et al, 2016), external dysmorphic features were seen in 79.4% of the cases and main dysmorphic features associated with CHDs were eye anomalies, ear anomalies, upturned nose, clinodactyly, and chest deformity- eye anomalies (long eyelashes, wide set eyes, and hypertelorism) (OR=176.6, p<0.0001), ear anomalies (OR=217.6, p<0.0001), upturned nose (OR=68.7, p=0.0002), clinodactyly (OR=58.7, p=0.0015), and chest deformity (OR=37.07, p=0.0145). [9]

Gucer S et al in their study found out that extra cardiac anomalies occur in 15-45% of cases with CHD. In 45.9% of cases, one or more extracardiac malformations were present. The most common extra cardiac malformation seen was craniofacial malformation.[10]

In our study, hypertelorism and total face dysmorphisms were found significantly higher in cyanotic CHD compared to acyanotic CHD, with p values 0.028 and 0.026 respectively. In a study conducted by Sumi Ghorai et al in West Bengal (2012-2013), on association of hypertelorism in children with congenital acyanotic heart disease, no statistical significance was revealed, but VSD was the most common congenital heart disease found in children with hypertelorism [11].

Conclusions

Dysmorphisms are very frequently seen in children with congenital heart diseases. Hypertelorism and face dysmorphisms are significantly high in cyanotic heart disease. Dysmorphisms should, hence warrant clinician to look for congenital heart diseases in children, as early diagnosis improves quality of life of these children.

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