



STUDY OF CONSANGUINITY AS A RISK FACTOR FOR CONGENITAL HEART DISEASE IN PEDIATRIC AGE GROUP.

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ABSTRACT

Introduction: Congenital heart diseases (CHDs) are multifactorial in etiology. Consanguinity could increase the likelihood of Congenital Heart Disease. So, we studied correlation between parental consanguineous marriages with CHDs in children aged 0-12 years.

Material and method: Confirmed cases of CHDs diagnosed by clinical examination and echocardiography in age group of 0-12 years were enrolled in study group. Children without any CHDs matched for age but not for gender, ethnicity or social class were enrolled in control group.

Results: During study period, 50 patients (male 26, female 24) were enrolled in study group and 50 patients (male 27, female 23) in control group. In study group, 43 (86%) parents of patients found to have consanguinity while in the control group, 20 (40%) parents of patients were found to have consanguinity. Most common congenital heart disease seen was Ventricular Septal Defect in 19 (38%) patients. Mortality rate was 8% in study group and 2% in control group.

Conclusion: Most common age group in patients with CHD was 0-1 years. Male to female ratio was equal in (1.08:1) in patients with CHD. Commonest symptom in patients with CHD was respiratory distress and fever. Maximum CHD patient's parent had 3rd degree consanguineous marriage. We found Consanguinity as a risk factor for congenital heart disease in Hindus and Muslims. Most common congenital heart disease was Ventricular Septal Defect. In the present study majority of the patients of CHD survived.

KEYWORDS :

INTRODUCTION:

Congenital heart disease (CHD) is defined as a malformation of the heart structure and or cardiac great vessels that happen during intrauterine life and present from birth.^(1,2) CHD is the most common defect among all birth defects representing a major global health problem.

Approximately about 6-8 infants per 1000 live births have cardiovascular malformations. With a believed incidence rate of 8/1000 live birth, nearly 180,000 children are born with CHDs each year in India. Of these nearly 60,000 to 90,000 suffer from critical CHDs per year, requiring early intervention.⁽³⁾

CHDs have multifactorial etiology, involving interaction between genetic and environmental factors. Hence, we planned to study correlation between parental consanguineous marriages with CHDs in a representative sample of pediatric population.⁽⁴⁾

MATERIAL AND METHOD:

This hospital based case control observational study was conducted in department of Pediatric, Government Medical College, Aurangabad Maharashtra from November 2017 to November 2019 after approval from Institutional Ethical Committee.

After written informed consent, all pediatric patients from 0 to 12 years attending pediatric OPD and IPD were enrolled. 50 cases were enrolled in study group who had CHD confirmed by clinical examination and echocardiography. 50 cases without CHD were enrolled in control group. Cases in control group were matched for age with cases in study group but not for gender, ethnicity or social class. A predesigned proforma

was used to collect information. After enrollment, detailed history and physical examination were done. All necessary investigations like CBC, chest X ray, ECG, 2DECHO and other required investigations were done in both study and control group. Treatment with oxygen, IV fluid, IV antibiotics, inotropes, diuretics, nebulization were given as per need. Complications were noted and treated accordingly. Outcome was noted as 'Discharge' or 'Death'.

RESULTS:

During study period, 50 patients were enrolled in each study and control group.

Table 1 -Demographic profile of patients enrolled

Parameter	Study group	Control group	P value
Age distribution			
0-1 year	37 (74%)	37 (74%)	0.07 ⁺
1-5 year	07 (14%)	07 (14%)	
5- 12 year	06 (12%)	06 (12%)	
Mean age (months)	15.46 ± 24	14.1 ± 23.36	
Sex distribution			
Male	26 (52%)	27 (54%)	0.99 ^s
Female	24 (48%)	23 (46%)	

Value: Mean ± SD (Otherwise mentioned) + Unpaired t test; two tailed p value >0.05 not significant (@95 %CL)

\$ Chi square test for Trend; p value >0.05 not significant (@ 95 CL)

Table 1 shows demographic profile of patients enrolled. Most common age group having CHD was 0- 1 year [37(74%)]. There was male preponderance with 26 (52%) cases being male in study group and 27 (54%) in control group as shown in table 1.

The mean age and sex in both groups were comparable and no statistically significant difference was found. (p value for age 0.07⁺, p value for sex 0.99).

Table 2 -Symptomatology in Patients

Symptoms	Study group (N=50)	Control group (N=50)
Fever	29 (58%)	27 (54%)
Cold	22 (44%)	26 (52%)
Cough	11 (22%)	15 (30%)
Respiratory distress	29 (58%)	26 (52%)
Feeding difficulty	05 (10%)	05 (10%)
Vomiting	04 (8%)	05 (10%)
Loose motions	02 (4%)	04 (8%)
Headache	01 (2%)	00 (0%)
Convulsions	02 (4%)	01 (2%)
TOTAL	105(100%)	109(100%)

In the present study, most common symptoms were fever [29 (58%)] and respiratory distress [29 (58%)] in study group while fever [27 (54%)] and cold [26 (52%)] were most common symptom in control group as shown in table 2.

Table 3 -Distribution of patients according to Consanguinity in parents

Consanguinity Status	Study group (N=50)	Control group (N=50)	Total No (%)	P value
Present	43 (86%)	20 (40%)	63 (63%)	0.001
Absent	07 (14%)	30 (60%)	37 (37%)	
Total	50 (100%)	50 (100%)	100 (100%)	

Chi square value- 22.47, df-1, p value- 0.001 Chi square test for Trend; p value < 0.05 significant (@ 95% CL)

In present study, consanguinity was seen in 43 (86%) cases in study group and 20 (40%) cases in control group as shown in table 3.

There was statistically significant difference for presence of consanguinity in study and control group (p value 0.001).

Table 4 - Distribution of patients according to Degree of Consanguinity in parental marriage.

Degree of Consanguinity	Study group (N=50)	Control group (N=50)	Total No (%)
2 nd degree	2 (4%)	1 (2%)	3 (03%)
3 rd degree	41 (82%)	19 (38%)	60 (60%)
No	7 (14%)	30 (60%)	37 (37%)
Total	50 (100%)	50 (100%)	100 (100%)

In present study, most parents had 3rd degree consanguineous marriage in study group seen in 41 (82%) cases while in the control group non consanguineous marriage was common and was seen in 30 (60%) cases as shown in table 4.

Table 5 -Distribution of Hindu parents according to Consanguinity Status

Consanguineous Marriage	Study group (N=50)	Control group (N=50)	Total No (%)	P value
Present	16 (72.72%)	7 (24.13%)	23 (45.09%)	0.02
Absent	6 (27.27%)	22 (75.86%)	28 (54.90%)	
Total	22 (100%)	29 (100%)	51 (100%)	

In present study, consanguinity seen in 16 (72.72%) cases in study group and 7 (24.13%) cases in control group in Hindu parents as shown in table 5.

There was statistically significant difference for presence of consanguinity in Hindu parents in study group as compared to control group (p value 0.02).

Table 6- Distribution of Muslim parents according to Consanguinity status.

Consanguineous Marriage	Study group (N=50)	Control group (N=50)	Total No (%)	P value
Present	27 (96.42%)	13 (61.90%)	40 (81.63%)	0.001
Absent	1(03.57%)	8 (38.09%)	9 (18.36%)	
Total	28 (100%)	21 (100%)	49 (100%)	

In present study, consanguinity was seen in 27 (96.42%) cases in study group and 13(61.90%) cases in control group in Muslim parents as shown in table 6.

There was statistically significant difference for presence of consanguinity in Muslim parents in study as compared to control group (p value 0.001).

Table 7- 2 D ECHOCARDIOGRAPHY findings in patients with congenital heart disease.

Findings	(N=50)	Percentage (%)
Ventricular Septal defect (VSD)	19	38
Atrial Septal Defect (ASD)	18	36
Tetralogy of Fallot(TOF)	3	6
Transposition of great arteries(TGA)	4	8
Patent ductus arteriosus(PDA)	5	10
Patent foramen ovale (PFO)	1	2
Total	50	100

In present study, the most common congenital heart disease found was Ventricular Septal Defect in 19 (38%) patients ,followed by Atrial Septal defect 18 (36%) as shown in table 7.

Table 8 - Outcome of patients.

Outcome	Study group (N=50)	Control group (N=50)	P value
Survival	46(92%)	49(98%)	0.08
Death	4(8%)	1(2%)	
Total	50 (100%)	50 (100%)	

Chi square value- 1.87, df-1, p value- 0.08 Chi square test for Trend; p value < 0.05 significant (@ 95% CL)

In the study group,46 (92%) patients survived while in control group, 49 (98%) patients survived as shown in table 8.

In present study mortality in both the groups were comparable and no statistically significant difference was found (p value 0.08).

DISCUSSION:

The most common age group of presentation was 0-1 years in both study and control group and was similar in many studies like Golmei et al (2018)⁽⁶⁾, Bhushan deo et al (2015)⁽⁶⁾. But Vinod Jacob et al(2009)⁽⁷⁾ reported most common age group 0-1 weeks in 23 (46%) patients. The reason for this might be due to study was conducted on neonates.

In the study group ,there was male preponderance with male to female ratio 1.08:1 while in control group male to female ratio 1.17:1 and was similar in studies like Ariz Fazeriandy et al (2018)⁽⁸⁾, Pedram Nazari et al (2016)⁽⁹⁾. Deveshwar Dev et al(2016)⁽⁴⁾ shows male to female ratio of 2.2:1 in study group and 1:1 in control group. This variation might be because large number of patients were enrolled in study group (n=518).

In present study, common symptoms in study group were respiratory distress 29 (58%) and fever 29 (58%) while in control group were fever 27 (54%). This findings were similar in studies like Karthiga et al (2017)⁽¹⁰⁾, Redddy et al (2016)⁽¹¹⁾. Ashok kumar meena et al(2016)⁽¹²⁾ reported cough as the

most common symptom in 219 (56.2%) patients. The reason might be because this study was conducted in a different geographical location i.e Bikaner, Rajasthan.

In present study, consanguinity was seen in 43 (86%) cases in study group and 20 (40%) cases in control group and was similar in studies like **Zaid R et al (2010)**⁽¹³⁾, **Abbas Muhammed Hussain et al (2010)**⁽¹⁴⁾. **Deveshwar Dev et al (2016)**⁽⁴⁾ found consanguinity in 34 (6.56%) cases in study group while 7 (2.9%) cases in control group. This difference might be due to difference in study population as large no of cases were enrolled (n=518) and it was conducted in children between 0 to 5 years of age.

In present study, most parents in study group had 3rd degree consanguineous marriage [41 (82%)]. While in the control group non consanguineous marriage was common and was seen in 30 (60%) cases. This finding was similar in studies like **Abbas Muhammed Hussain et al (2010)**⁽¹⁴⁾. But study done by **Ramegowda et al (2006)**⁽¹⁶⁾ found most parents in study group had 2nd degree consanguineous marriage [68 (46.81%)]. While in control group 3rd degree consanguineous marriage was seen in 31 (15.5%) cases. This study was conducted in South India (Mysore) and more number of patients were studied (n=144). This may explain difference in degree of consanguinity from present study.

In present study, consanguinity seen in 16 (72.72%) cases in study group and 7 (24.13%) cases in control group among Hindu community and was similar in study done by **Bittles et al (1991)**⁽¹⁵⁾. But **Ramachandra Barik et al (2016)**⁽¹⁶⁾ reported 29 (27.88%) cases had consanguinity in Hindu parents. This difference might be because the study was carried out in a different geographical location i.e Andhra Pradesh.

In present study, consanguinity seen in 27 (96.42%) cases in study group and 13 (61.90%) cases in control group among Muslims. Similar findings was seen in study done by **Deveshwar Dev et al (2016)**. But **Badaruddoza et al (1994)**⁽¹²⁸⁾ found 25 (3.37%) cases had consanguinity in Muslim parents. This difference might be because the study was carried out in Muslims of different geographical location like Aligarh city, North India and more cases were enrolled (n=1721) in this study.

In present study, the most common congenital heart disease was Ventricular Septal Defect [19 (38%)], followed by atrial Septal defect [18 (36%)] and was similar in studies like **Fuad I. Abbag et al (2006)**⁽¹⁷⁾, **Ashok kumar Meena et al (2016)**⁽¹²⁾. But **Begum et al (2017)**⁽¹³⁾ conducted a study and found ASD as the most common congenital heart disease [45 (72.6%)]. This difference might be explained because study was carried out in neonates in their study.

In the study group, 46 (92%) patients survived while in control group 49 (98%) patients survived. This finding was similar in study like **Golmei et al (2018)**⁽⁵⁾. On contrary, **Vinod et al (2009)**⁽⁷⁾ reported 28 (56%) patient's survival. This less survival might be because these patients had complex congenital heart diseases.

CONCLUSION:

In our study, most common age group in patients with CHD was between 0-1 years. Male to female ratio was almost equal in (1.08:1) in patients with CHD. Commonest symptom in patients with CHD was respiratory distress and fever. We found Consanguinity as a risk factor for congenital heart disease in Hindus and Muslims. Maximum patient's parent had 3rd degree consanguineous marriage. Most common congenital heart disease was Ventricular Septal Defect. In the present study majority of the patients survived in both study & control group.

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