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RESEARCH ARTICLE
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EVALUATION OF CONSANGUINITY AS A RISK FACTOR FOR CONGENITAL HEART DISEASE IN PATIENTS WITH ADULT CONGENITAL HEART DISEASE AT TERTIARY CARE HOSPITAL IN PAKISTAN

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Abstract:

Objectives: To assess consanguinity as a potential risk factor for congenital heart disease in patients with adult congenital heart disease in a tertiary care hospital setting in Pakistan.

Materials and Methods: This is cross sectional study conducted in outpatient department of Pediatric Cardiology, National Institute of Cardiovascular Disease (NICVD), Karachi, Pakistan from November, 2022 to October, 2023. Patients with age equal and more than 18 years with CHD were enrolled. This study A predesign questionere were used to collect data. For statistical analysis we used SPSS Version 25.

Results: A total of 129 patients were enrolled. The mean age of the patients with congenital heart disease (CHD) was 25.74±9.77 years. Among the total enrolled CHD patients, 73 (56.6%) were male, and 65 (43.4%) were female. The history of CHD in the mother, father, and siblings was reported in 7 (5.4%), 5 (3.9%), and 7 (5.4%) cases, respectively. Additionally, 52 (40.3%) patients had a history of consanguinity in their parents' marriage.

Conclusion: The study's conclusion indicates that there is a heightened risk of Congenital Heart Diseases (CHDs) in adult patients associated with consanguinity in parental marriages.

Key words: Consanguinity, Congenital heart disease, Adult patients, Pakistan.

Introduction:

Consanguinity refers to the degree of kinship or blood relationship between individuals.(1) Consanguinity is derived from two Latin words: "con," signifying similarity, and "sanguineus," meaning blood.(2) It denotes a connection among individuals who share common ancestors or belong to the same bloodline.(3) Pakistan has one of the highest rates of consanguineous marriages (marriages between close relatives) in the world. (4) The practice of cousin marriage is deeply rooted in the country's cultural, social, and historical traditions. It is estimated that around 62% of marriages in Pakistan involve cousins marrying each other.(5) The prevalence of hereditary diseases in Pakistan is a significant public health concern, and it's associated with the high rate of consanguineous marriages in the country.(6, 7) Around 30 million individuals in Pakistan are afflicted with various forms of hereditary diseases.(8) Every year in Pakistan, approximately 40,000 children are born with congenital heart defects.(9) And in India, approximately 180,000 children are born with congenital heart defects each year.(10, 11) Consanguineous marriages, have been associated with an increased risk of certain genetic disorders and congenital anomalies, including congenital heart defects.(12) in the present research studying consanguinity as a potential risk factor for adult congenital heart disease (ACHD) involves investigating the possible connection between the degree of blood relationship between parents (consanguinity) and the likelihood of their offspring developing congenital heart defects that persist into adulthood.

Objective:

To assess consanguinity as a potential risk factor for congenital heart disease in patients with adult congenital heart disease in a tertiary care hospital setting in Pakistan.

Methodology:

This cross sectional study has been conducted at Outpatient Department of Paediatric Cardiology, NICVD, Karachi, Pakistan for the period of 1 year from (Nov, 2022 to Oct, 2023)**.** An approval was obtained from the hospital's ethical review committee. Patients of both genders with age more or equal than 18 years with CHD who presented first time were included in the study. Confirmation of CHD was done by Chest X ray, ECG, and 2D Echocardiography. A written informed consent was obtained from the enrolled patients/guardian after explaining the purpose and benefits of the study. A predesign questionere were used to collect data. Patients data like age, weight, gender, residence, history of consanguinity of marriage in parents and history of CHD in parents and siblings were obtained. For statistical analysis we used SPSS Version 25.

Results:

Total 129 patients were enrolled. The mean age of the patients with CHD was 25.74±9.77 years (Table 1). The mean weight of the patients was 48.76±10.60. Out of total enrolled CHD patients 73(56.6%) patients were male and 65(43.4%) were female (Table 2, Fig 3). (Table 2, Fig 2). History of CHD in mother, father and in sibling were 7(5.4%), 5(3.9%) and 7(5.4%) respectively. History of consanguinity of marriage in parents were 52(40.3%). In our study patients 120(93.0%) patients were of 18-40 years of age group, 8(6.2%) patients were of 41-60 years of age group and the remaining 1(0.8%) were of >60 year of age group. The patients were the residence of Sindh 97(75.2%), Kpk 6(4.7%), Baluchistan 11(8.5%), Punjab 7(5.4%), Azad Kashmir 6(4.7%), Gilgit 1(0.8%) and Afghanistan 1(0.8%) (Table 2, Fig 2).

Table 1: Mean age of all enrolled Patient *(n=129)*

Variables	**Mean±SD**
Age (Years)	25.74±9.77
Weight (kg)	48.76±10.60

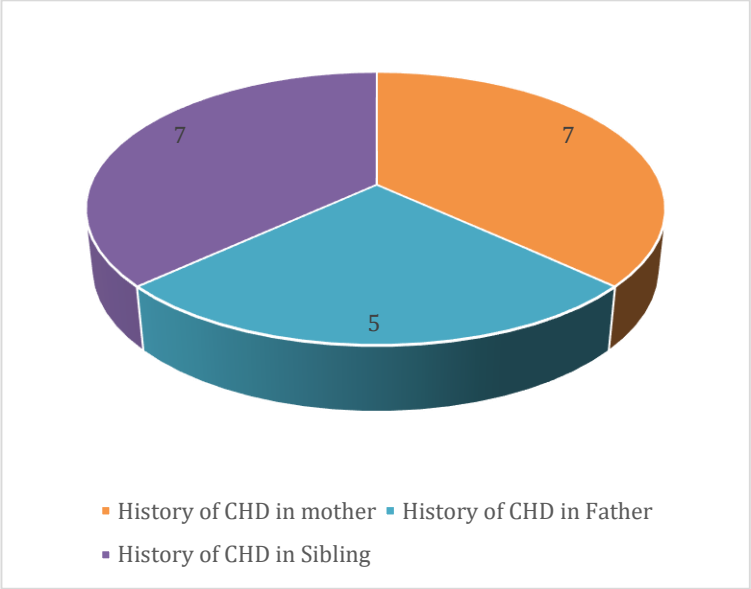

Fig 1: Frequency of CHD in mother, father and sibling.

Table 2: characteristic of all the enrolled patients *(n=129)*

Variables	Frequency	Percentage
Gender		
Male	73	56.6
Female	56	43.4
History of CHD in mother	7	5.4
History of CHD in Father	5	3.9
History of CHD in Sibling	7	5.4
History of consanguinity of marriage in parents	52	40.3
Age groups		
18-40 years	120	93.0
41-60 years	8	6.2
>60 years	1	0.8
Residence		
Sindh	97	75.2
Kpk	6	4.7
Baluchistan	11	8.5
Punjab	7	5.4
Azad Kashmir	6	4.7
Gilgit	1	0.8
Afghanistan	1	0.8

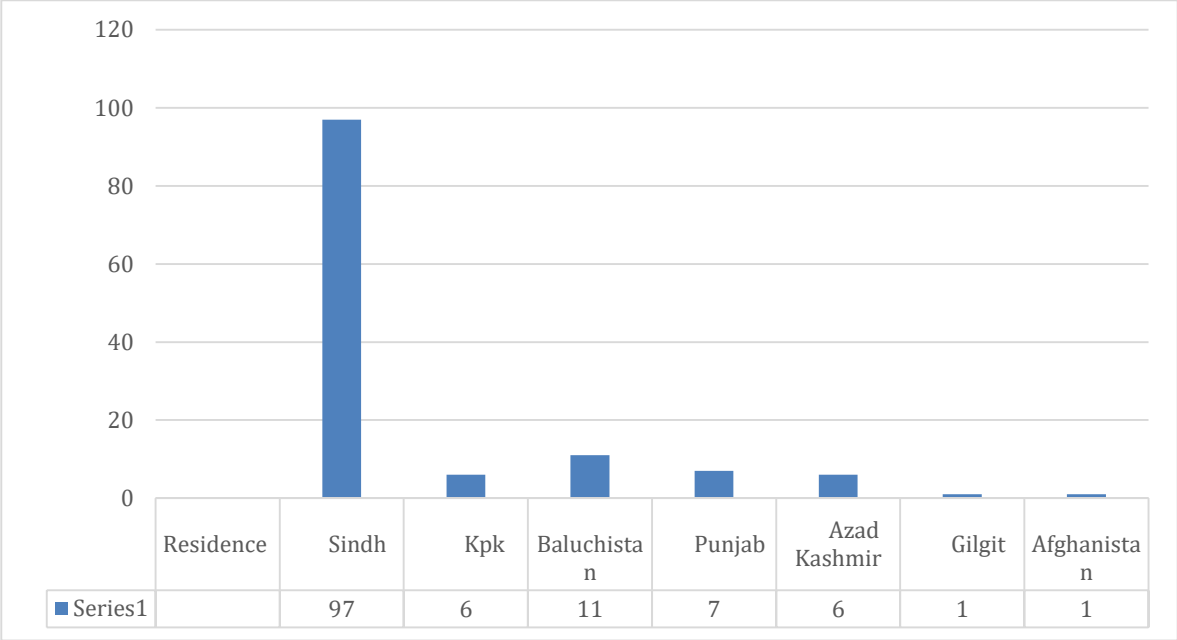

Fig 2: Frequency of CHD patients on the basis of residence

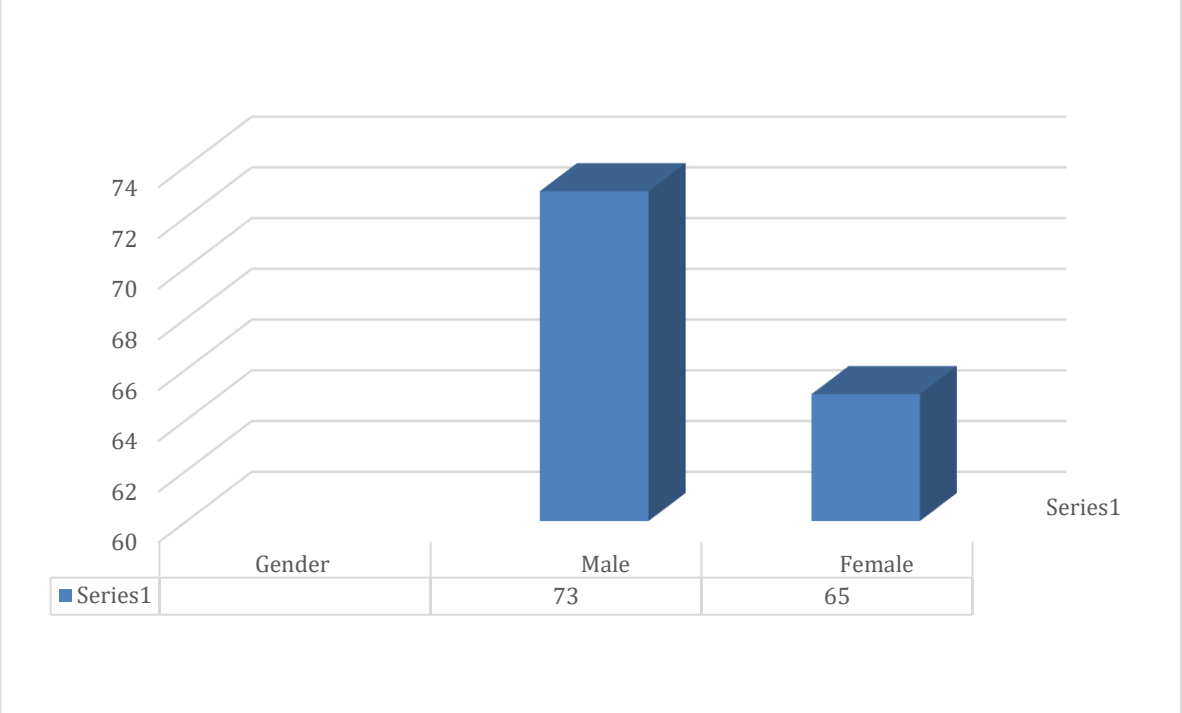

Fig 2: Frequency of gender.

Discussion:

Consanguinity has been suggested as a potential risk factor for congenital heart disease (CHD) in adults. Studies have explored the association between consanguineous marriages and the prevalence of congenital heart defects. Consanguinity can increase the risk of certain genetic disorders, including those affecting the heart. When close relatives have shared genetic material, there is a higher likelihood of passing on recessive genetic mutations associated with congenital heart diseases.

The aim of the present study was to assess consanguinity as a potential risk factor for CHD in adult congenital heart disease patients in a tertiary care hospital setting in Pakistan. In our study all the enrolled patients were suffering from CHD and the frequency of consanguinity of marriage in parents were 52(40.3%). This is clear from the frequency that there is strong association between CHD and

consanguinity. A study conducted by Becker et al on 1013 patients with Congenital Heart Diseases (CHDs) and compared the data to rates of consanguinity from an earlier study by ElHazmi et al, involving 3212 Saudi families. The comparison showed a statistically significant association ($p < 0.001$) between first-cousin marriage and CHDs in the study population. This suggests that there was a notable correlation between consanguineous marriages, particularly first-cousin marriages, and the prevalence of Congenital Heart Diseases in the studied population.(13, 14) our study finding was supported by the study of Deveshwar Dev et al.(15) in which they stated that among the 758 participants in the study, 41 individuals (5.41%) had parents who were in a consanguineous marriage. This suggests that the occurrence of congenital heart diseases (CHDs) is associated with a higher risk when parents have consanguineous marriages. An another study conducted by Yunis et al.[16] revealed that consanguinity was identified in 17.9% of cases with Congenital Heart Diseases (CHDs) compared to 9% in the control group. The observed results were found to be statistically significant with a p-value of <0.001. This suggests a notable association between consanguinity and the occurrence of CHDs in the studied population. A study conducted by Haq et al.[17] demonstrated that among 250 cases of Congenital Heart Diseases (CHDs), 122 patients (49%) were born to consanguineous marriages. In contrast, among the control group, only 72 patients (29%) had parents with consanguinity. Upon multivariate analysis, consanguinity was identified as an independent risk factor for CHDs in children, with a statistically significant p-value of <0.001. This indicates that consanguineous marriages were associated with a higher risk of CHDs in the study population, even after accounting for other variables. The present study shows a history of CHD in mother, father and in sibling were 7(5.4%), 5(3.9%) and 7(5.4%) respectively. Our study was supported by Maryam Naveed et al.(18) stated that 14% of mothers in the study had congenital abnormalities resulting from consanguineous marriages. This suggests a correlation between consanguinity in marriages and the occurrence of congenital abnormalities in mothers within the examined population. In our study the enrolled patients belong to all over Pakistan, in which most of the patients were from Sindh.

Conclusion:

It was concluded that consanguinity in parental marriages is associated with an elevated risk of Congenital Heart Diseases (CHDs) in patients.

Study Limitation: This is single centre study comprising majority of population from one region (sindh) does not represent the whole population of pakistan.

Conflict of interest: Authors declare no conflict of interest.

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