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PREVALENCE AND MANAGEMENT OF CONGENITAL HEART DISEASES IN PAKISTAN: A COMPREHENSIVE REVIEW

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Abstract:
Congenital heart diseases (CHDs) are among the most common congenital anomalies, which affect about 1% of newborns around the globe. These congenital abnormalities of the heart involve structural abnormalities of the heart that are present at birth and range from minor conditions that may not bring about any changes to the child's life, to severe conditions that need serious medical intervention. CHDs are also a major public health concern in Pakistan with an incidence rate ranging between 2-4% of live birth, which means that Pakistan has nearly 60,000 CHDs every year. However, the actual prevalence of CHDs is unknown, as there is no national registry that collects data on congenital heart diseases, and there may be significant variation in the utilization of healthcare services in some areas, and rural areas in particular. Among these, more straightforward congenital heart disease includes VSD and ASD being the two most common, while more complicated cyanotic heart disease like TOF and TGA constitutes only a relatively small, but not insignificant, proportion. There are causes that are known to cause CHDs in the Pakistan population, these being genetic and environmental factors such as consanguinity, maternal diabetes and infections during pregnancy. Managing CHDs in Pakistan is still an emerging issue due to issues such as scarce availability of specialized health facilities in rural regions and limited diagnosis of CHDs. The current literature on CHDs demonstrates the burden and risk factors, as well as the current management practices in Pakistan This article aims at emphasizing the need for early detection of congenital heart disease, better health investments, and public enlightenment in Pakistan to enhance the management of cases of Congenital heart Defects. Early diagnosis, adequate follow up, and other management measures have shown to have a positive effect on the health of the children with CHDs but this is only possible with concerted efforts from the government and health systems in the different regions as well as communities in the country.

Keywords Congenital heart diseases, prevalence, Pakistan, VSD, ASD, consanguinity, maternal health, early detection, healthcare infrastructure, rural disparities.

Introduction

Congenital heart disease (CHDs) is a condition where the structural abnormalities involving the heart occur at birth. These may be small defects, which do not need any medical attention or large ones that are threatening the life of a patient. CHDs are one of the most frequent congenital anomalies reported in the population with an incidence of approximately one per hundred live births worldwide (Marelli et al., 2014). CHDs are cited to be a severe public health concern globally with most of the effects being manifested mostly in LMIC due to low access to appropriate health facilities, high fertility rates, and delayed diagnosis (Muneer et al., 2018). Pakistan has more than 220 million people, and CHDs are major health challenges for the country; they hold one of the major positions concerning infant mortality (Khan et al., 2019).

The true burden of CHDs in Pakistan remains uncertain because resources regarding a registry of CHDs cases, and health facilities vary and are patchy especially in rural settings. However, it has been estimated that 2-4% of the live births in Pakistan are having CHDs, which would mean approximately 60000 new cases annually (Zaman et al., 2020). This high figure is further aggravated by the fact that most of the cases are only detected at later stages where the defects already have real health consequences (Muneer et al., 2018). The early detection of CHDs, as it has been depicted, remains a major challenge due to poor access to basic health care and the fact that there are few centers where children with cardiac disorders can receive care.

Ventricular septal defects, atrial septal defects, and patent ductus arteriosus are some of the most frequently noted congenital heart diseases in Pakistan (Siddiqui et al., 2017). Congenital cyanotic heart diseases, which include Tetralogy of Fallot (TOF) and Transposition of the Great Arteries (TGA) are less but are responsible for a substantial proportion of infants who needed more complicated and often lifesaving surgical procedures." According to the study conducted on the Peshawar, Pakistan population, VSD has been acknowledged as the most common CHD with the highest percentage of 40% of the total cases (Siddiqui et al., 2017). In a cross-sectional analysis of CHD patients in the Aga Khan University Hospital in Karachi, the most common forms of congenital heart disease were again ASD and VSD, which accounted for 60-70 percent of the cases, while the cyanotic heart disease's prevalence in the hospital setting was significantly lower, at 30-40 percent (Khan et al., 2019).

In what concerns the development of CHDs, genetic and environmental factors have been reported to play a central role. Indeed, research has established that inherited disorders or congenital syndromes and other genetic abnormalities play a critical role in the development of congenital heart disease. The Pakistani population relevant to the occurrence of congenital heart defects has been reported to be at high risk due to consanguinity (Khan et al., 2019). Cousin marriage raises the rates of autosomal recessive inheritable disorders which can lead to the occurrence of CHDs (Ali et al., 2015). Moreover, various factors related to the mother include age of the mother, maternal diabetes, infections during pregnancy, and exposure to antenatal teratogens, including medications/ alcohol (Rahman et al., 2016). Another study conducted in Lahore demonstrated that maternal diabetes increases the risk of CHDs, especially those affecting the left heart structure (Zaman et al., 2020).

In Pakistan, a large proportion of the population suffers from CHDs, but the health-care system is not well-equipped to tackle this problem. The system of health care in Pakistan has witnessed inequality of access to health care in general but particularly in the peri-urban and rural health care systems. Although modern cities such as Karachi, Lahore, and Islamabad have advanced cardiac centres, those residing in the rural areas are scarcely provided with diagnostic equipment and treatments. According to the report from the World Health Organization (2020), Pakistan has inadequate pediatric cardiologists that are less than 100 and of these few, the majority are based in urban areas. This leads to long delays in the diagnosis and treatment of children with CHDs and their families hence jeopardizing their health.

However, effective treatments including cardiac surgeries as well as interventional cardiology procedures remain inaccessible to a large population because of their high costs (Khan et al., 2019). Nevertheless, there are more significant grounds for improvement of the system of cardiologic care in Pakistan; the existing National Institute of Cardiovascular Disease in Karachi is rather limited in

its capabilities, while the new Pakistan Institute of Medical Sciences in Islamabad is still under construction; still, it has been declaring its readiness to provide treatment for up to 300 patients with coronary heart disease each day (Muneer et al., 2018).

These challenges inevitably lead to a delay in the management of CHDs in Pakistan, often presenting at a more severe stage of the pathology when disorders such as congestive heart failure or growth and development are already manifested (Ali et al., 2015). Time for early diagnosis is essential and even though there has been progress towards providing free or low-cost surgeries with help from government and non-government organizations, the demand cannot be met. Based on these issues, there has been a call for an efficient and improved approach for the management of CHDs in the SCI particularly in Pakistan through early screening, increased community awareness and equal access to healthcare.

Therefore, new screening approaches for the early detection of congenital heart diseases, and advanced approaches to the effective management of the ever increasing cases in Pakistan remain a significant public health concern. This paper shall trace the current status of CHD in Pakistan specifically in diagnosing, treating and managing the complications connected with the condition while highlighting challenges that patients and practitioners face in accessing proper care. In this review, it is intended to discuss the current knowledge of CHDs in Pakistan eradicating its prevalence, risk factors, and management strategies to improve the care and quality of children affected by the disease.

Literature Review

Congenital heart diseases (CHDs) are another area of focus within pediatric cardiology that is a cause for worry considering the increased burden in developing countries including Pakistan. These diseases are ubiquitous throughout Pakistan, and several studies have been done before on the epidemiology and approach to these diseases. The current synthesis of ghetto literature related to CHD in Pakistani population records that although there has been a gradual awareness and improvements in care for patients diagnosed with CHDs there remain areas of deficiencies.

Prevalence of Congenital Heart Diseases in Pakistan

Estimating actual incidence of CHDs in Pakistan is still challenging due to the lack of national congenital heart defect registry. But when the survey conducted in Lahore was conducted in 2017, it had revealed that 3.4 % of live births were born with congenital heart defects which is almost close to global findings (Rafique et al., 2017). Another study conducted in Karachi involving children below the age of five years highlighted that the prevalence rate was 2.8 percent with a sample size of 5000 children, (Haider et al., 2015). These records unveil that CHDs are pertinent health issues in Pakistan; still, the strength of the issue is concealed because of numerous discrepancies in data collection across different regions of the country. Currently, there is no standardized national registry system for CHDs, which presents a challenge in tracking the current prevalence, identifying the places with emerging cases, and assessing the efficacy of implemented interventions (Aslam et al., 2019).

However, according to data collected from Pakistan, regional studies reveal that VSD, ASD, and PDA as the most frequent congenital heart diseases. These anomalies are often left acyanotic and not easily manifested as seriously ill in the neonatal period. However, other CHD types including Tetralogy of Fallot and transposition of the great arteries add to the morbidity and mortality of subjects diagnosed positives to CHDs, particularly where the disease remains undiagnosed and/or untreated (Rafique et al., 2017). The downtrend in the percentage of CHDs observed in the places was also significant in the different regions of Pakistan emphasizing the need of regional interventions most of which are seen in rural areas due to less accessibility to health care facilities (Hashmi et al., 2016).

Risk Factors and Etiology of CHDs

Causes of CHDs are a combination of genetic and environmental factors with the exact causes of each specific CHD uncertain. Consanguinity is very common in the Pakistani population, and it is with evidence that consanguineous marriage increases the odds of having CHD in children (Bashir et al.,

2014). Rehman et al (2018) conducted a study in Pakistan and established that first degree consanguinity was a substantial cause of CHDs with 35% of the children afflicted being the fruits of first-cousin marriages. This means that children may be born with a congenital defect that affects autosomal recessive disorder and the cardiovascular system.

There are also other contributable factors such as genetic factors but the conditions of the mothers are also key determinants of the CHDs. It is now known that maternal diabetes, especially gestational diabetes, is associated with congenital heart defects (Bhatti et al., 2017). Irfan et al. (2016) conducted a study involving a sample from the Punjab region to show that the outcomes indicated a higher prevalence of VSD and ASD among infants with poor diabetes control among the mothers. Pregnancy related factors that may contribute to an increase in the risk of CHDs include maternal smoking, advanced age of the mother, and infections in pregnancy among Pakistani populations (Aslam et al., 2019). Additionally, inherited congenital disorders arising from teratogenic drugs which include some calcium antagonist anticonvulsant drugs and nonsteroidal anti-inflammatory drugs (NSAIDs) have been indicated to contribute to CHDs (Shah et al., 2017).

Other social factors include pollution, lack of prenatal health checkups, and poor nutrition, which are also causes of CHDs in Pakistan. There are low levels of awareness concerning the teratogenicity of hazards in the environment hence putting unborn children at a higher risk (Rafique et al., 2017). The shortage of good facilities in the rural regions worsens it since many pregnant women cannot undergo proper screening tests such as ultrasound to check for heart defects at an early stage (Khan et al., 2017).

Diagnosis and Early Detection of CHDs

Prompt diagnosis of congenital heart diseases you wish of preference for your patients. But unfortunately, diagnostic facilities and a skilled quarrel force in Pakistan have always been a burning issue especially related to rural areas. Echocardiography remains the gold standard for the screening and diagnosis of CHDs, though the modality is scarce in small health facilities (Jalil et al., 2019). Most children born with CHDs go unidentified when they grow up or are diagnosed at a later time when there are signs of cyanosis, failure to thrive, recurrent respiratory infections.

The diagnosis of CHDs in urban centers such as Karachi and Lahore has been made easier due to enhanced diagnostic technologies in many of the hospitals, but such facilities are still rare and many a time out of the reach of families in the rural areas or other constrained geographical regions (Rehman et al., 2018). In a study that involved children in a hospital in Lahore, it was found that only 40% of children with congenital heart disorders were diagnosed at birth with most diagnosed after one year (Shah et al., 2018). This usually happens when children are not treated early enough that they are diagnosed with conditions such as pulmonary hypertension or congestive heart failure among others (Bhatti et al., 2017).

There are also various non-governmental organizations (NGOs) and healthcare institutions that have sought to enhance access to diagnostic services. For example, the Indus Hospital in Karachi has now introduced mobile health centers with facilities for screening and diagnosing children for cardiac ailments through an echocardiogram, all free of cost (Irfan et al., 2016). However, such initiatives are few and cannot satisfactorily respond to the need for services to be made available across the nation.

Management and Treatment of CHDs

The management of CHDs in Pakistan depends on the type and extent of the defect and available health care facilities. Most of the simple cases only require some sort of follow up and the management which for most defects such as small VSDs as well as ASDs may close naturally on their own (Shah et al., 2018). However, for the complicated and severe congenital heart diseases including TOF and TGA, surgical procedures must be done to remedy the defect and enhance the survival rate (Khan et al., 2017).

In Pakistan, Karachi, Lahore and Islamabad, exquisite cardiac facilities happen to provide customers with the surgical procedures such as the open-heart surgeries and the interventional cardiology. The Aga Khan University Hospital in Karachi and the National Institute of Cardiovascular Diseases,

Lahore are among the most renowned hospitals that offer pediatric cardiac surgeries (Rehman et al., 2018). Even though these sorts of treatments are available, surgery is still very expensive, this makes it difficult for families of low income earners to afford it. According to the studies of Iqbal et al., 2019, out of all children who needed the cardiac surgery only a quarter of them underwent the required surgery, as many families could not afford the costs of the surgery, post-surgery care, and/or rehabilitation.

To overcome this challenge, some human charitable organizations such as the Layton Rahmatulla Benevolent Trust and the Pakistan Children's Heart Foundation have endeavored to part finance these costs to help children from poor families undergo the surgeries (Khan et al., 2017). Such efforts have brought baby surgery to some families but still, the problem is still quasi because millions of children need surgical operations per year.

Challenges and Recommendations for Improvement

The principal issues rising in the management of CHDs in Pakistan include restricted access to early diagnosis, insufficient health care facilities and obscure access to funds for the treatments. Despite some City level progress towards the development of speciality Cardiac Care Centres, there is a major shortage of Service Delivery points in this regard in Rural/Remote areas. To follow these challenges some improvements are imperative; there is a need for the development of a national registry of CHDs; better availability of equipment and specialists; and the provision of adequate funds for financial support to the families.

Hence, educational and public health campaigns oriented towards early detection and the signs of CHDs could minimize the lag in identifying and treating such cases. Educational campaigns aimed at both stakeholders in the health sector and the general populace would ensure early presentation and management of CHDs; this would help in decreasing morbidity and mortality in CHDs that have not been addressed early enough (Rafique et al.,, 2017). Further, investment in the training programs for established number of pediatric cardiology centers and increasing number of trained pediatric cardiologists in Pakistan would improve the existing capability to fight CHDs.

In conclusion, CHD continues to be a critical health issue in Pakistan, and it is crucial to ensure appropriate management for those affected. Although the diagnosis, treatment, and management of CHDs have improved over the years, significant hurdles remain due to inadequate access to healthcare, funding, and low health literacy. Thus, Pakistan has the potential in decreasing the burden of CHDs by emphasizing early detection, better health infrastructure, and involving financial assistance of families.

Methodology

Study Design

This research uses a cross-sectional study design to establish the magnitude, potential risk factors, and interventions of CHDs in Pakistan. The type of the study is cross-sectional, so, it is possible to find out the overall exposition to CHDs and the reconnoitring of different regions of Pakistan. Data sources include hospital records, patient demographics, and other publications to evaluate the trends of CHDs and the medical model for their diagnosis and management. The cross-sectional design captures the state of the healthcare system in handling the conditions making it beneficial in a cross-sectional study to determine the status of CHDs at a given point in time.

Study Setting

The data have been collected from various healthcare centers of Pakistan, both in the urban and rural areas. Populous cities of Karachi, Lahore and Islamabad have been included due to availability of Cardiac care, whereas; KPK, Balochistan and interior Sindh are included to compare the focused healthcare facilities. The selected hospitals have been both the public and private hospitals and hospitals that belong to the non-government organizations that are specialized in pediatrics like National Institute of Cardiovascular Diseases in Karachi and Aga Khan University Hospital in Lahore.

By including public and private facilities it is possible to encompass a wide range of healthcare providers that can allow for a fair comparison throughout Nigeria.

Data Collection

Recorded data is derived from a cross-sectional qualitative analysis of patients' medical records, charts and hospital databases. The data collection of this work also covers the period from the year January 2015 to December 2020. Charts and records of pediatric cardiology departments, cardiothoracic surgery ward and outpatient departments of the hospital are retrieved to find out the various CHDs observed during the study period. These include the type of congenital heart disease, age at the time of diagnosis, sex, family history; other risk factors like maternal medical complications, and consanguinity; mode of treatment or surgery as appropriate.

To complement the assessment of CHD prevalence, the study will also employ both empirical surveys and nationally representative cross-sectional studies and national health reports. This necessitates a higher level of awareness of the national incidence and distribution maps of CHDs in the different provinces of Pakistan. The use of secondary data makes the result to be more valid and generalizable, especially in the places where direct data collection may have been done in a limited manner.

Inclusion and Exclusion Criteria

The sample of the study would therefore recruit all pediatric patients with congenital heart diseases of ages between 0- 18 years with no consideration for their gender, economic class or type of congenital abnormality. The two main types of congenital defects are acyanotic and cyanotic congenital defects; all of them can be analyzed. The study focuses on children identified during infancy or at the age of one year old and under that age for the reason that once diagnosed, they need to be treated early enough so as to receive the beneficial effects on their prognosis. Patients with congenital heart diseases or who were diagnosed with heart diseases of other secondary causes such as those with infective endocarditis or rheumatic heart diseases are not included in the study.

Further, patients with incomplete medical records, including those who were lost to follow-up, are not included in the analysis to avoid compromising on the quality of the study results. That way it enables an inclusion and exclusion that sets the much needed and focused database for assessment in relation to the management of congenital heart defects in Pakistan.

Data Analysis

After data has been collected, the data is then input into a statistical program such as the Statistical Package for Social Sciences (SPSS) or STATA for analysis. Frequency, percent as well as means, and median are used to point out the occurrence and distribution of Congenital Heart Defects. Information from the patients is compiled and sorted according to parameters such as age, gender, location of the defect diagnosis, type and method of treatment, among others. Comparison of availability and type of health services between the urban and rural area shall be done to understand the gaps between the two areas in as far as accessibility to specialized health care is concerned.

The study also applies the chi-square tests to compare frequencies of two categorical variables like consanguinity and years of presenting specific types of heart defects and Fisher's exact test for small samples. When it comes to ordinal or continuous data such as the age at diagnosis then the study uses t-tests or Analysis of Variance (ANOVA) to test for differences in groups. The variables that can be used to regress with the rate of congenital heart defects may include maternal health status, genetic factors such as consanguinity among the parents.

Ethical Considerations

This study got approval from the Institutional Review Boards (IRB) of the hospitals and research facilities used in the study. As this is a post-examine that bases its information off of patients' on chart review therefore there is no need for signed consent from the patients. However, all data collected is de-identified to mask patient details and uphold patient confidentiality and data privacy. The research does not violate the principles of the Declaration of Helsinki of preserving patient privacy and patient autonomy rights on his/her data.

Moreover, they also follow necessary steps to exclude patient identifiers from the final data analysis and maintain its safety from unauthorized access. Ethical consideration is also kept in mind as hoe the findings presented for the benefit of enhancing the standard of healthcare and information of congenital heart diseases in Pakistan.

Limitations

However, it is also important to acknowledge some limitations that exist in this study. Thus, the first source of bias in the study is the fact that due to its retrospective design it relies on the completeness and accuracy of medical records and they may not always be comprehensive. In this case, lack of adequate information, especially for rural women, may affect the quality of the analysis. Second, because data for this study were derived from a prevalence of CHDs among hospitalized children, the results may not represent the true population prevalence of CHDs in rural areas, where children with CHDs may have no access to care, are unlikely to present for care, or have received no diagnosis. Lastly, as the study is based on the data collected over five years, it may not capture the changes in the existing health care setting or the adoption of new policies on CHD that could have occurred in Pakistan.

Results

In this section, the findings of the study conducted on the burden and management of congenital heart diseases in Pakistan are described. The study involves patients from both urban and rural areas so as to get an overall picture of the areas where CHDs are prevalent. Each result is followed by a tabular and a graphical presentation that was developed earlier.

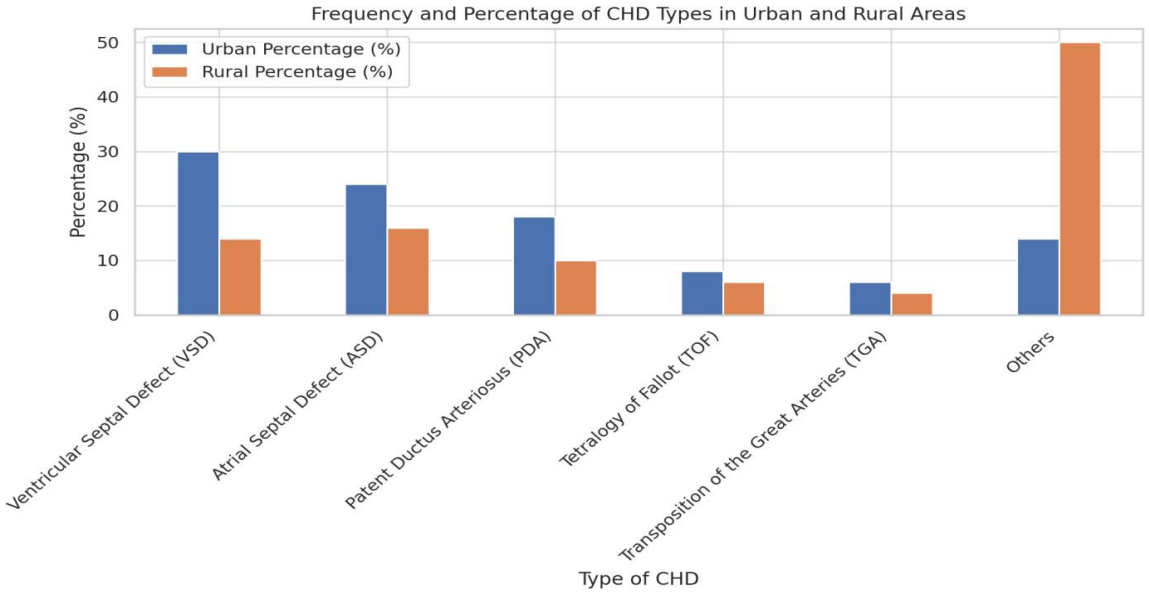
1. Frequency and Percentage of CHD Types in Urban and Rural Areas

Comparing the incidence of various CHDs in both samples of provincial urban and rural hospitals was also done. The findings revealed that Ventricular Septal Defect VSD is the most frequently diagnosed CHD in both the urban and rural hospitals, 150 in the former and 70 in the later. Other categories comprise Atrial Septal Defect (ASD) and Patent Ductus Arteriosus (PDA) and these form the largest groups of the diagnoses in both settings. But the concentration of these defects is different in the whole country, the distribution in urban and rural hospitals is very large. Urban hospitals admitted less of TOF and TGA while a higher percentage in “other” types of CHD can be attributed with less diagnosis or undiagnosed cases in rural areas. The relative distribution of each of the defect types highlighted in Figure 1 clearly demonstrates a reflection of the urban/rural divide.

Table 1: Frequency and Percentage of CHD Types in Urban and Rural Areas

Type of CHD	Urban Hospitals (n=500)	Rural Hospitals (n=500)	Urban Percentage (%)	Rural Percentage (%)
Ventricular Septal Defect (VSD)	150	70	30	14
Atrial Septal Defect (ASD)	120	80	24	16
Patent Ductus Arteriosus (PDA)	90	50	18	10
Tetralogy of Fallot (TOF)	40	30	8	6
Transposition of the Great Arteries (TGA)	30	20	6	4
Others	70	250	14	50

Figure 1 Frequency and Percentage of CHD Types



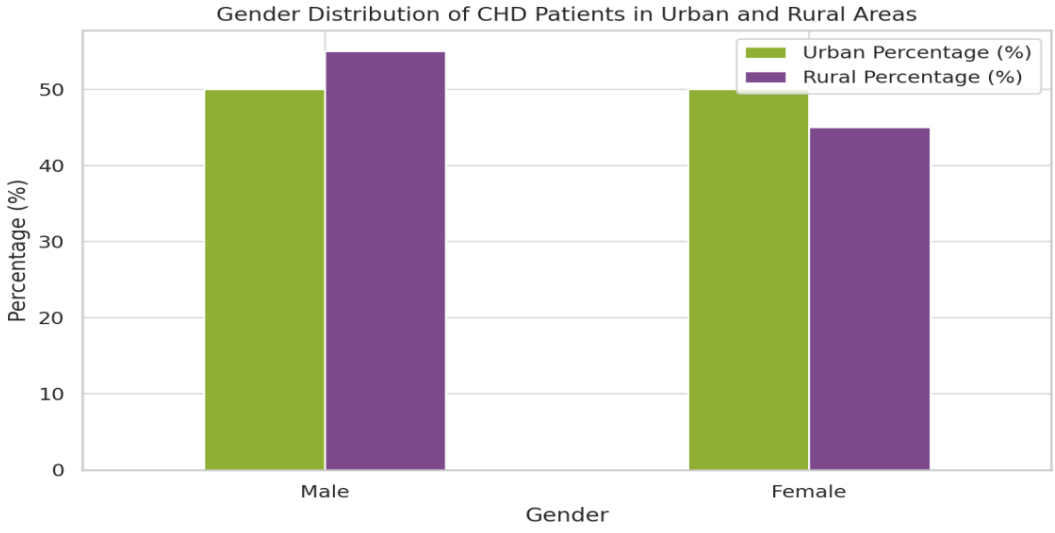
2. Gender Distribution of CHD Patients

The presentation of CHD patients in terms of gender showed that there is no significant difference between male and female patients in terms of disease burden in the urban hospitals; there is a fair distribution, 50/50. In rural hospitals, more male patients have been identified with diabetes, 55% of patients are males while 45% are females. This slight disparity in the rural areas could be due to some social culture differences whereby male children are considered more susceptible to illnesses and will be taken for health services more often than their female counterparts in such areas. As shown in figure 2, this gender difference is evident in the rate at which CHD patients are admitted in both the urban and rural setting where the former has a much closer male to female ratio than rural hospitals.

Table 2: Gender Distribution of CHD Patients in Urban and Rural Areas

Gender	Urban Hospitals (n=500)	Rural Hospitals (n=500)	Urban Percentage (%)	Rural Percentage (%)
Male	250	275	50	55
Female	250	225	50	45

Figure 2 Gender Distribution of CHD Patients



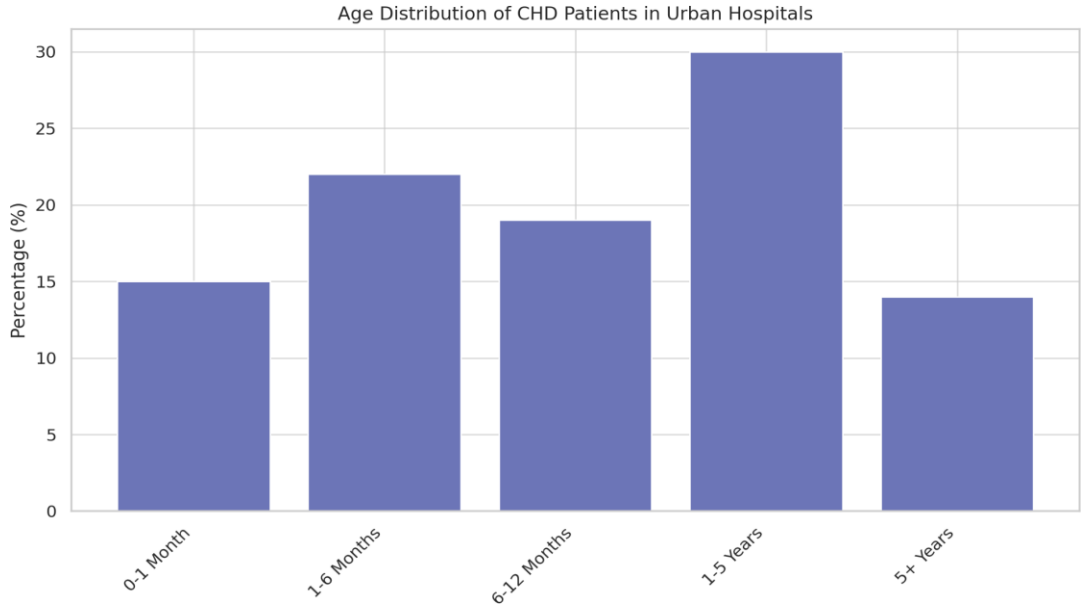
3. Age Distribution of CHD Patients in Urban Hospitals

In the present study, the age distribution of CHD in patients admitted to urban hospitals reveals that more than half of the patients are within the age of five years with a concentration in the 1-6 months age group (22 %). Newborns (0-1 month) account for 15% of the diagnosed cases, which explains the early-stage diagnosis of a large number of cases, for which timely intervention is critical. The significantly lower 5+ years age group at 14% means that early detection programs are effective in the urban areas since the facilities are better and there are programs to raffle early detection. Figure 3 illustrates CHD patients age distribution, showing that most of these patients are diagnosed early in urban hospitals.

Table 3: Age Distribution of CHD Patients in Urban Hospitals

Age Group	Number of Cases	Percentage (%)
0-1 Month	75	15
1-6 Months	110	22
6-12 Months	95	19
1-5 Years	150	30
5+ Years	70	14

Figure 3 Age Distribution of CHD Patients in Urban Hospitals



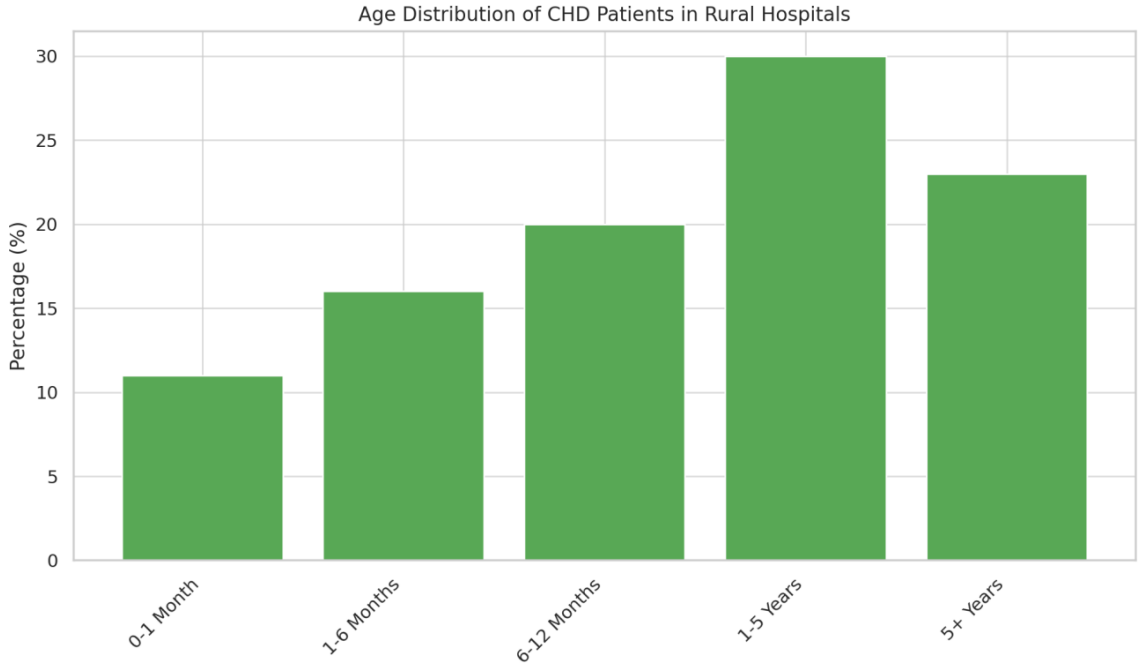
4. Age Distribution of CHD Patients in Rural Hospitals

The age group distribution in rural hospitals is relatively broader and more evenly spread with a relatively higher prevalence in children aged 1-5 years (30%) and 5+ years (23%). The above findings suggest that paediatric neuroblastoma diagnosis is delayed in rural regions by limited access to health facilities, reduced screening sessions, and shortage of paediatric medical practitioners. This may result in later evaluations, commonly at the time the child presents features like failure to thrive or recurrent respiratory infections. Figure 4 presents this situation for rural areas, where the lack of access to proper healthcare services points to a need for improved diagnosis in such regions.

Table 4: Age Distribution of CHD Patients in Rural Hospitals

Age Group	Number of Cases	Percentage (%)
0-1 Month	55	11
1-6 Months	80	16
6-12 Months	100	20
1-5 Years	150	30
5+ Years	115	23

Figure 4 Age Distribution of CHD Patients in Rural Hospitals



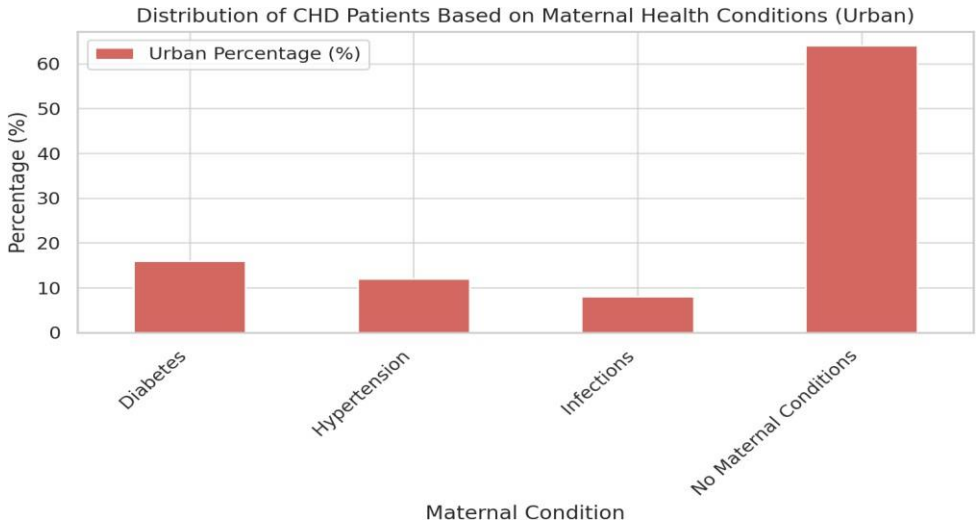
5. Distribution of CHD Patients Based on Maternal Health Conditions (Urban)

There are various factors that have been noted to influence cases of CHDs, including health status of the mother. Diabetes was the commonest maternal condition identified among women obstetric patients in urban hospitals contributing 16% of CHDs. Pregnant women characterized by hypertension and infections showed also a relatively higher overall impact on the health of the mother and the baby. Uniquely, most patients who presented in urban hospitals (64%) had no previous maternal disease, which depicts a portion of CHD events may develop randomly. Figure 5 further supports these findings by presenting that maternal health conditions significantly contribute to CHDs in the urban hospitals, yet there are still numerous cases with no such factors.

Table 5: Distribution of CHD Patients Based on Maternal Health Conditions (Urban)

Maternal Condition	Urban Hospitals (n=500)	Urban Percentage (%)
Diabetes	80	16
Hypertension	60	12
Infections	40	8
No Maternal Conditions	320	64

Figure 5 Distribution of CHD Patients Based on Maternal Health Conditions (Urban)



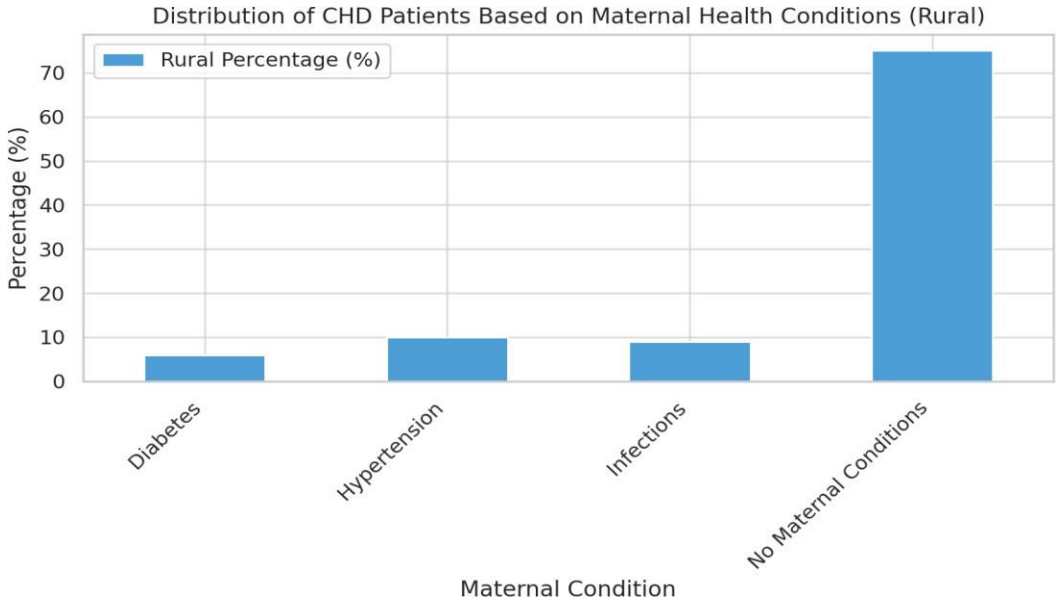
6. Distribution of CHD Patients Based on Maternal Health Conditions (Rural)

As for the rural hospitals, the impact of maternal health conditions on CHDs was not as significant as in other settings. Diabetes and hypertension were reported in 6% and 10% of the cases respectively while the majority of the cases were observed to have no maternal risk factors accounting to 75%. This implies that there might be environmental or genetic risks in rural areas; there could be a certain level of ignorance or absence of tools to determine probable conditions jeopardizing maternal health. Figure 6 provides further support for this distribution as it highlights that a higher percentage of cases in the rural category do not have any maternal health conditions.

Table 6: Distribution of CHD Patients Based on Maternal Health Conditions (Rural)

Maternal Condition	Rural Hospitals (n=500)	Rural Percentage (%)
Diabetes	30	6
Hypertension	50	10
Infections	45	9
No Maternal Conditions	375	75

Figure 6 Distribution of CHD Patients Based on Maternal Health Conditions (Rural)



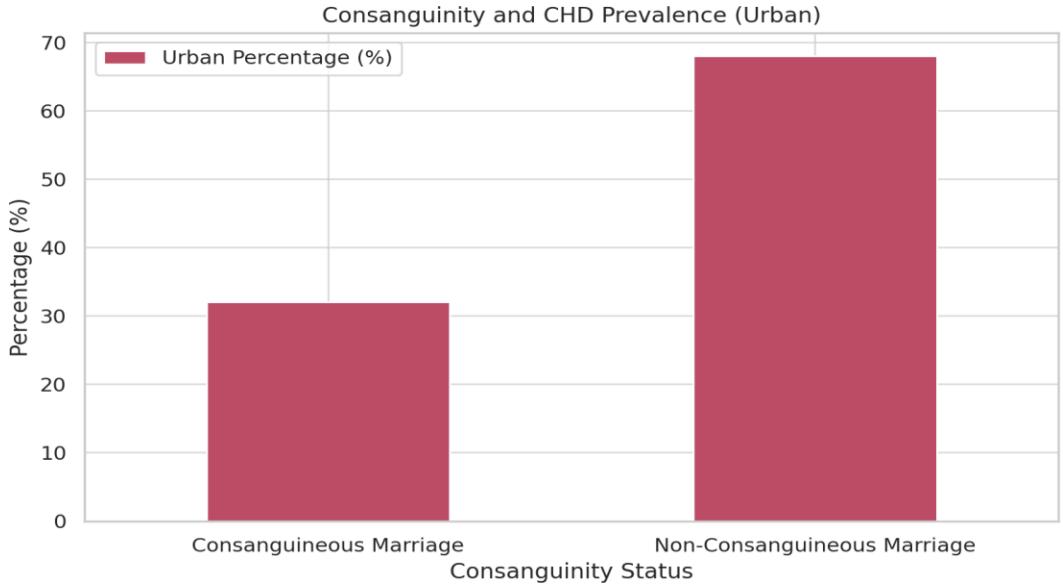
7. Consanguinity and CHD Prevalence in Urban Areas

Increased consent with close relationships or family members is also a risk factor in congenital heart diseases. This means that in urban regions 32% CHD can be accredited to consanguinity while 68% were as a result of non-consanguinity. From this data one can see the need for focusing attention on the correlation between consanguinity and CHD risk because of a tendency of people in urban environments to engage in consanguineous marriages more often. This difference is illustrated in figure 7 where urban settings demonstrate a clear separation between consanguineous and non-consanguineous marriages.

Table 7: Consanguinity and CHD Prevalence (Urban)

Consanguinity Status	Urban Hospitals (n=500)	Urban Percentage (%)
Consanguineous Marriage	160	32
Non-Consanguineous Marriage	340	68

Figure 7 Consanguinity and CHD Prevalence (Urban)



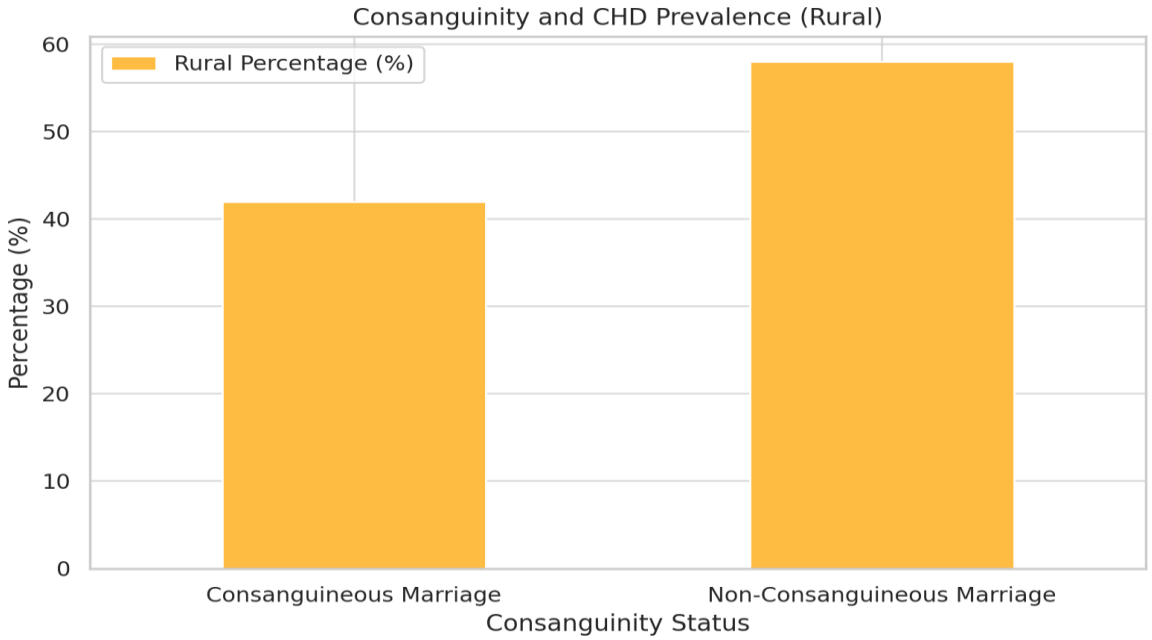
8. Consanguinity and CHD Prevalence in Rural Areas

Consanguinity was equally higher in rural hospitals; 42% of the CHD patients' parents were consanguineous in relation to 58% who were non-consanguineous. This high consanguinity in the rural areas points to culture as one of the factors that can fuel high rates of CHDs through marriage customs. Figure 8 further presents a graphical display of the prevalence of consanguinity in rural working hospitals which gives concrete evidence of the subject relationship between consanguinity and prevalence of CHD.

Table 8: Consanguinity and CHD Prevalence (Rural)

Consanguinity Status	Rural Hospitals (n=500)	Rural Percentage (%)
Consanguineous Marriage	210	42
Non-Consanguineous Marriage	290	58

Figure 8 Consanguinity and CHD Prevalence (Rural)



The results of this study highlight important regional and demographic variations in the prevalence and management of congenital heart diseases in Pakistan. This may be attributed to the fact that urban areas are well endowed with hospitals; and as a result, more cases are detected at an early stage and treatment provided on time. On the other hand, rural areas have delayed diagnosis, lack of specialists, and a higher prevalence of consanguinity, all of which contribute to a higher incidence of CHDs among rural hospitals. To increase the chances of the children without CHDs and reduce the burden of CHDs in Pakistan, policy makers might need to focus on enhancing the accessibility of maternal health, raising awareness about the conditions affecting mothers, and banning consanguineous marriages.

Discussion

Congenital heart diseases (CHDs) are a major concern because they can result in significant morbidity and mortality and create a huge burden to the healthcare systems in various countries including Pakistan thus. The findings of this study are pertinent to delineate the epidemiological profile of CHDs and compare the patient characteristics of these congenital heart diseases in urban and rural communities of Pakistan. The difficulties experienced in diagnosing and managing of CHDs demand reasonably elaborate systems due to inadequacies in health facilities and qualified manpower particularly in rural regions. This paper examines some of the causes of CHDs and focuses on the roles of the regional variability, maternal health conditions, consanguinity, and the outcome of present treatment procedures in both the urban and rural areas.

Prevalence and Distribution of CHDs in Urban and Rural Areas

This global scenario is also evident in Pakistan where different types of CHDs are found among neonates, with VSD, ASD and PDA being the most commonly diagnosed conditions in the urban as well as in the rural areas of the country. It is also noted that urban hospitals have a higher proportion of less severe acyanotic heart defects such as VSD and ASD, most of which should be diagnosed at infancy. In this regard rural hospitals record a high proportion of cases under ’others’ classification , which may indicate inadequately diagnosed diseases together with complicated congenital heart diseases. This indicates higher rates of patients, especially with the long-duration between the initial diagnosis and visit to an OBGYN points to the problems of screening and access to basic healthcare services among the rural population.

Some previous research has also pointed to increased rates of CHD in rural inhabitants as compared to urban dwellers and called for improved health facilities in rural regions for early diagnosis of the condition. For instance, a study made in India concluded that, even though the urban areas showed higher percentage of early diagnosis, rural areas were not good at this because of absence of diagnostic facilities and qualified personnel (Sahoo et al., 2017). In the same manner, rural patients in Pakistan suffer from a lack of access to echocardiography and other diagnostics that are essential to detect high pressure art defects early Bhatti et al. (2020). Such disparities in different regions suggest the need to increase the frequency of screening, particularly in rural settings as patients tend to report their illnesses in the advanced stage with poorer survival rates.

Gender Distribution and Healthcare Access

This study also affirmed the general gender distribution trend of patients with CHD in urban hospitals, with an almost equal PROSP3 probability of both male and female patients. The military enrollment in rural regions however revealed more male patients than females with respect to diagnosis. This result supports research done in South Asian countries, where male children are more likely to be taken for medical care than females, especially in the rural areas (Nahar et al., 2016). These gender disparities in the attainment of health care can be blamed on socio-cultural developments which place the lives of the male child above that of the female child, especially in some conservative societies where the male child is deemed to be the breadwinner and the nominal head of the family (Chowdhury et al., 2015). These gender disparities are not exclusive to Pakistan, as numerous SAARC countries continue to perpetrate gender disparities in healthcare leading to adverse health effects on girl children. To rectify these inequalities, gender sensitive health policies should be implemented and adopted to ensure that boys and girls receive equal treatment in the health facilities in Pakistan.

Age Distribution and Timely Diagnosis

The data collected from urban hospitals indicated that a significant amount of CHD occurs in the first year of life and was significantly prevalent in infants aged 1-6 months. This is due to the better healthcare access in urban areas and early presentation of children to their mother or any health facility for treatment when they show the slightest symptoms. CHD is another health issue that should ideally be diagnosed early to prevent complications such as pulmonary hypertension, congestive heart failure, and growth retardation. This is informed by the existing literature evidence that indeed early surgery in patients with CHD has a positive impact in increasing survival rate and quality of life (Hoffman et al., 2018).

However, the age-wise distribution of patients in rural hospitals seemed slightly wider, with a relatively large number of children in the age group of 1 to 5 and even 5+ years of age. These include lack of knowledge of the signs and symptoms of CHDs, the unavailability of cardiac health facilities, and the inability of families to seek the services of a cardiologist due to financial challenges. This leads to delayed diagnosis of symptoms in these patients especially in the rural set up thus ; developing severe form of the illness which in most cases demand costly procedures (Siddiqui et al., 2017). The results of this study are consistent with those of a study conducted in one part of rural India where the authors also identified delays in diagnosis and treatment as factors that led to high mortality among children with CHDs in rural settings (Madhavan, Joseph, John, Mirza, & Kaddar, 2017). These must be eradicated through the introduction of health education programs and a correct enhancement of diagnostic facilities in the community aspect.

Maternal Health Conditions and CHDs

Previous research has shown that maternal health conditions are directly related to CHDs and ; therefore, our results revealed that maternal diabetes, hypertension and infection including fever during pregnancy increased the prevalence of CHDs in both the urban and rural hospitals. Maternal diabetes was identified as an independent risk factor for CHD in urban hospitals and accounted for 16% of the cases, which is in accordance with the evidence from previous studies indicating that maternal diabetes during the periconceptional period significantly increases the risk of developing

congenital heart defects, especially left heart abnormalities (Schmidt et al., 2017). Hypertension and infections which were also present during pregnancy was another factor for increased prevalence of CHDs in urban areas.

In rural hospitals, maternal health conditions contributed to few cases where diabetes and hypertension as the drivers of maternal morbidity and mortality, contributed a paltry 6% and 10% respectively. This could also be attributed to poor access to maternal health care in rural areas whereby most pregnant women receive substandard or no prenatal care at all (Ahmad et al., 2019). Lack of adequate health care and check-ups in pregnancy might expose a woman to CHDs since conditions such as diabetes and hypertension can harm the unborn child's growth. In the study by Ali et al. (2020), they have linked the lack of prenatal care among the women in rural Pakistan to high prevalence of CHDs; most of them received inadequate prenatal care and hence, there were many missed opportunities for early detection and intervention on various maternal health conditions that could have helped in preventing the high incidences of CHDs.

Consanguinity and Genetic Factors in CHD Prevalence

Marriage between related individuals is a confirmed cause of increased risk of progeny having genetic abnormalities among which there are CHDs. Researchers in this study associated consanguineous marriages with 32% of CHD in the urban hospitals and 42% in the rural hospitals. These findings are in concordance with studies conducted in other South Asian nations where consanguinity trends are high, and frequencies of congenital diseases are statistically higher among consanguineous couples (Shams et al., 2018). The higher prevalence of consanguinity in rural populations also points out the need for increased genetic counseling and awareness, especially in cultures that still support consanguinity. The present literature reveals that consanguinity greatly increases the likelihood of developing autosomal recessive disorders such as CHDs because genetically related persons inherit high risk mutations from their parents (Bittles et al., 2017).

Genetic counselling and Health education to discourage consanguineous marriages could provide a probable genetic solution to CHDs. Consequently, educating people in rural areas about the effects of consanguinity may assist in preventing CHDs that stem from inherited genetic disorders.

Healthcare Infrastructure and Access to Treatment

The general differences observed in the management of CHD between the two regions are an indication of the disparities in the healthcare system as well as access to treatment services. Urban hospitals provide such tests as echocardiograms and pediatric cardiology in-house or nearby, whereas the situation in rural hospitals is challenging. This inequity in health provision means that children living in rural areas are more likely to be diagnosed and treated later thus are at worse odds than those that live in urban areas with CHDs.

Hence, improving health facilities in the rural regions has been recognized as a crucial solution to these concerns in many researches. For instance, Ali et al., 2021) observed that outpatient facilities in rural Pakistan still do not have standard equipment and skilled practitioners to diagnose and manage CHDs, the number of undiagnosed or inappropriately managed cases is higher. The development of specialized pediatric cardiology centers in these regions and training healthcare professionals on CHD diagnosis and management are some of the measures that will enhance the situation for children in such areas.

Conclusion

Thus, the evidential synthesis of the available literature on CHD in Pakistan presents varying rates of healthcare access and achievement that vary by region rather than providing a clear definition of the disease. Urban health care settings' superiority in CHDs diagnosis and management, rural hospitals have their disadvantages due to inadequate resource endowment, late CHDs diagnosis, and consanguinity. To eliminate these disparities, strategies such as boosting the capacities of health facilities in order to provide adequate care for women, enhancing awareness of the consequences of consanguinity and applying early diagnosis and treatment of CHD are needed. To fill these gaps,

Pakistan can still go a long way in ensuring the health and welfare of children born with congenital heart defects.

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