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Article

Prevalence of Congenital Heart Diseases Among Infants

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Abstract: Background: Congenital Heart Diseases (CHD) is the most common type of major congenital malformations number among newborns as well as the leading cause of morbidity and mortality in infancy especially in low-developed countries. Studies across different regions highlight the interplay of multiple social and environmental risk factors, such as high prevalence of consanguineous marriages, weak pre- and post-natal screening programs and the need for reliable local epidemiological data in certain provinces in Iraq, including Kirkuk9 volume up. **AIM**: To identify the frequency of congenital heart diseases in infants admitted to Kirkuk General Hospital during the three years period (2023 -2026), and the clinical presentation and risk factors associated with it. **Materials and Methods**: A retrospective cross-sectional study was performed including a review of medical records of the patients with congenital heart diseases underwent two-dimensional echocardiography in the Maternity, Gynecology, and Pediatrics Hospital for the period from January 2023 to June 2026. The demographic, clinical and parental risk variables were taken and the data was analyzed using SPSS version 26. **Results** :Between 1982 and 2020, there were 26,450 live births in our study, and diagnosed 312 cases of congenital heart disease, with a prevalence of 11.8 per 1,000 live births. The m:f ratio was 53.8-46.2% (1.17:1) The most frequent type was ventricular septal defect (VSD) (31.4%), followed by atrial septal defect (18.3%) and patent ductus arteriosus (14.1%). Non-cyanotic forms were (77.9%) more frequent than cyanotic ones (22.1%). Consanguinity was present and the non-use of folic acid in the pre-pregnancy period was found in 57.1% and 46.2% respectively Cases detected annually demonstrated a gradual increase, starting at 89 and ending at 119 cases between 2023 and 2026. **Conclusion**: The rate of congenital heart diseases in Kirkuk Governorate is significantly in a high level, and this rate is in agreement with the pattern detail in other Iraq governorates, and in consanguineous marriages play important role for as a major risk factor. Too Many Heart Defects Go Unnoticed, Highlights Need for Registry and Early Screening Efforts The findings highlight the urgent need for national cardiac screening programs and a unified registry for congenital heart defects.

Keywords Congenital heart diseases; infants; prevalence; Kirkuk; Iraq; consanguineous marriage; echocardiography.

1. Introduction

Congenital heart diseases are proposed to be defined as structural or functional abnormalities that appear in the heart or great vessels connected to it during the embryonic development period. Most important are the most frequent type of major congenital defects in newborns, with an incidence of 8 to 12 cases per 1,000 births based on the majority of international epidemiology studies [1,2].

A global systematic overview pooling > 24 million live births showed that the reported prevalence of congenital heart diseases has steadily increased over decades [1].

Furthermore, this can largely be explained by improvements in ultrasound diagnostic techniques as well as improvement in clinical awareness, more than by a true biological increase in incidence alone [2]. This upward gravitas was reasserted in a recent 260-study review of 20 countries from some of the continents where health care systems have excelled, once again with very different continent-to-continent relationships reflecting differences in diagnostic capabilities [3]. The Global Burden of Disease Study 2017 also reported that congenital heart diseases are still one of the top morbidities and mortalities attributable to chronic diseases in infants and children aged younger than five years, even though advances in cardiac surgery and catheter interventions have helped improve therapeutic results substantially [4].

Due to advances in surgical and interventional care, the vast majority of children with mild to moderate CHDs survive into adulthood, creating a new population of adults with CHDs who require lifelong, specialized follow-up care [5]. Nevertheless, while major improvements in survival after diagnosis have occurred, these have been very limited by late presentation, and, in most low- and middle-income countries, the absence of specialist services for cardiac surgery provide limited benefit from this advance in many developing regions.

At the embryological stage, the primitive heart is formed during the third week of pregnancy, initiated by the fusion of embryonic heart streamers from the mesodermal layer, with the chamber, valve, and septation formation almost complete by the eighth week of pregnancy, and any genetic, epigenetic, or environmental disturbance occurring in this critical time window may be sufficient to cause a structural congenital defect [6] The orchestrating of this process by a network of transcription factors and signaling molecules highlights the complexity of the developmental genetic control, disruption of which results in a spectrum of cardiac defects as evidenced by molecular studies [6], [7].

Congenital heart diseases can be broadly classed into two categories: acyanotic forms, such as ventricular and atrial septal defects, patent ductus arteriosus, and aortic coarctation; and cyanotic forms, such as tetralogy of Fallot, truncus arteriosus, and transposition of the great vessels. In addition, a distinct classification by level of clinical complexity (simple, moderate, complex) is used to guide treatment options and facilitate triage to cardiac surgical centers [8]. The causes are many, including purely genetic and chromosomal (as seen in Down syndrome) and the multifactorial causes due to the interplay of genetic predisposition with environmental factors. In fact, about 10% of cases are linked to already identified single genetic mutations [7].

Among the most heavily researched categories of modern preventive research are the environmental modifiers of CHD risk, given that maternal exposure to viral infections such as rubella in the first trimester of pregnancy, pre-pregnancy diabetes, obesity, and the use of selected teratogenic medications, are all strongly associated with increased risk of congenital heart defects in the fetus [9]. There has also been marked geographical and racial variation in recorded prevalence rates. A large American study made it possible to quantify the relative differences both between holistic groups and between consecutive time periods in the recording of specific patterns of congenital heart defects [10]. In fact, the finding corroborates data from what is regarded as one of the oldest and largest epidemiological studies of congenital heart diseases in newborns, the Baltimore-Washington reference study [8].

Such matrimoniFon represents the mid properties of risks in Middle East and North Africa region, with rates going above 30-50% of the tandem in some communities. A large Saudi study reported an increase prevalence of congenital heart diseases compared to other communities in the Middle East and Europe with a clear link with first-degree consanguinity [11]. Another Lebanese study confirmed that cousin marriages are an independent risk factor for congenital heart defects in less developed countries [12] while a relatively recent genetic review from Saudi Arabia observed that parental consanguinity

is associated with an increased prevalence of certain congenital heart defects such as ventricular and atrial septal defects, common atrioventricular canal defect, and pulmonary artery stenosis [13], which are consistent with the recessive gene hypothesis. Another systematic review is in agreement with these findings, finding that first-degree consanguinity confers a twofold risk for congenital heart disease among offspring in populations with high rates of consanguinity [14]. Ordinary prevalence rates of congenital heart diseases range between neighboring nations on the regional level; from an Iranian study documenting rates approaching the global average [15] to a study from the central Anatolia region in Turkey showing a qualitative dispersion akin to World Health Organization classification, with a predominance of diaphragmatic defects [16]. A large school study in Alexandria, Egypt, recorded the prevalence and relative distribution of cardiac forms within the anticipated range in Middle East countries [17], whilst a report from the Sultanate of Oman noted an incidence almost 7 per 1000 live births [18]. The study highlighted different prevalence rates based on adopted methodology and examination tools in the Indian city of Mysore as well [19] which too shed light on the need for standardization of detection and diagnosis protocols across different studies.

In spite of the significance of this public health problem, this bibliometric review of scientific research outputs in the field of congenital heart diseases in the Arab world over the last 25 years indicates that regional research productivity is still extremely low compared to that of the developed countries, and focused mostly on clinical research at the expense of epidemiological and basic genetic studies. Moreover, countries motivated by armed conflict, e.g. Iraq and Libya, have significant losses in productivity in relation to their gross national income levels [20]. In fact, another bibliometric review had also confirmed this huge gap between developing Arab Arabic countries and Deviloped Countries concerning the area of congenital heart disease research [21]. In our opinion, this research gap comes hand in hand with genuine operational challenges, as to-date, pediatric cardiac surgery in many low- and middle-income countries continues to be limited by insufficient specialized human resources, and inadequate infrastructure, to address the actual demand [22]. In this context, a different strategy is mandatory to better address the issue of access in pediatric cardiac care in these countries [23].

Multiple local studies have been conducted recently on the monitoring of congenital heart diseases in the context of an environment burdened by an accumulated social, health and environmental load in Iraq. In a five-year hospital-based retrospective study from a private hospital in Baghdad 847 children with congenital heart disease were included with drastic differences in absolute rates and types of heart defects, the commonest being ventricular septal defect and the male/female ratio is 7.6/4.4 [24]. Similar demographic and clinical patterns were established with very high retrospective rates of congenital heart diseases among the children surveyed [25,26]. Similar results were also reported from the Ramadi Teaching Hospital for Obstetrics and Pediatrics, which showed that about 78% of the affected children with congenital heart disease was in consanguineous marriages Compared to 43.3% in non-affected controls, and both are statistically significant high difference [27]. Among children with congenital heart diseases who underwent surgical intervention, a study from the Kurdistan Region of Iraq reported relatively favourable surgical outcomes with a success rate of 95.5%, but with some postoperative complications reported in Duhok [28].

Conclusion: Early diagnosis through transthoracic echocardiography (TTE) is related to improvement of outcome in children with congenital heart diseases. This objective is made possible by accurate differentiation between simple forms that only need surveillance from complex forms which need urgent surgery. Even highly developed countries still show delays in diagnosis preceding birth and postnatal confirmation, highlighting the need to improve routine fetal and neonatal screening programs, as confirmed in a Norwegian study [29].

With the exception of the nationwide data set, there is an increasing local literature on congenital heart diseases, particularly from several Iraqi governorates, but there are no data to date regarding this important medical sector in Kirkuk a diverse governorate in terms of socio-ethnic composition with data poorly documented in a world of published studies. Because of the very high rates of consanguineous marriages in Kirkuk and the essential differences in availability of advanced cardiac screening services, an accurate mapping of the local epidemiology is crucial to better direct health policies and cardial screening programs at the local level.

The aim of this study is to: (1) determine the prevalence rate of congenital heart diseases among infants registered at the Obstetrics and Gynecology Hospital during the years 2023-2026; (2) describe the qualitative and demographics distribution of these cases by gender, age at diagnosis, type, and complexity of heart defect; (3) identify whether there are associations between the occurrence of these cases and parental risk factors (consanguineous marriages, maternal age, gestational diabetes, and non-used of folic acid); and (4) analyze the local results in comparison with those described in similar Iraqi, regional and global studies.

2. Materials and Methods

Study Design: A retrospective cross-sectional study design was used (by reviewing electronic and paper medical records)

Setting: The Pediatric Cardiology Unit and the Neonatology Division at the Maternity and Women's Hospital, between 16 January 2023 and 5 June 2026.

Study population and sample size: All infants (from one day old to the end of the sixth month) registered in the hospital database between September 2008 and April 2014 and the diagnosis of congenital heart defect was confirmed through two-dimensional echocardiography by pediatric cardiology specialists. The total number of live births during the study period was 26 450 births, which consisted of 312 cases of congenital heart defects, the final study sample.

Inclusion criteria were: (1) Diagnosis confirmed by echocardiogram by a specialist doctor (cardiologist); (2) Diagnosis of an infant less than 12 months; (3) A complete medical record; (4) Availability of all the required demographic and clinical characteristics (baseline and follow-up).

Exclusion criteria: (1) patients with non-congenital heart disease (e.g., rheumatic heart disease and endocarditis); (2) missing records or key outcome data; (3) patients referred from outside Kirkuk Governorate without a documented birth within the governorate.

Aggregated variables: The we prepared a questionnaire for data collection that comprised of (a) Demographic variables which consisted of: gender, age at diagnosis, place of residence (urban/rural); (b)Parental variables which contained: mother's age at birth, the degree of kinship between parents (first-degree or second-degree relatives or no kinship), gestational diabetes, a history of rheumatic disease or suspected viral infection during pregnancy, use of folic acid around the time of pregnancy, positive family history of congenital heart diseases; (c)Clinical variables which included: the type of heart defect based on the standard anatomical classification (ventricular septal defect, atrial septal defect, patent ductus arteriosus, tetralogy of Fallot, pulmonary artery stenosis, atrioventricular septal defect, aortic coarctation, transposition of the great vessels, and other complex forms), the degree of clinical complexity (simple/moderate/complex), and the outcome of the case (medical follow-up, surgical or catheter referral, death during the study period).

Statistical analysis: Data were entered and analyzed statistically by the SPSS statistical software version 26. Frequencies and percentages were described for descriptive variables; the Chi-square test was used to test relationships between categorical variables and a p-value less than 0.05 was considered as the level of significance.

3. Results

Out of a total of 26,450 live births recorded at Kirkuk General Hospital during the study period from January 2023 to June 2026, 312 confirmed cases of congenital heart diseases were diagnosed thru echocardiography, equivalent to an overall prevalence rate of 11.8 cases per 1,000 live births (1.18%).

Demographic distribution of the studied cases

The percentage of males among the affected cases was 53.8% (168 cases) compared to 46.2% for females (144 cases), with a sex ratio of 1.17:1 in favor of males. As for the age at diagnosis, the highest percentage of cases (41.0%) fell within the newborn category (0-28 days), followed by the 1-6 months age group (33.0%), and then the 7-12 months age group (17.9%). Only 8.0% of the cases were diagnosed after the age of 12 months, which reflects a relative effectiveness in the early detection programs within the hospital (Table 1, Figure1).

Table 1. The demographic distribution of congenital heart disease cases by gender and age at diagnosis (n = 312).

Variable	Number (n)	Percentage (%)
Sex		
Male	168	53.8
Female	144	46.2
Age at Diagnosis		
Neonatal period (0–28 days)	128	41.0
1–6 months	103	33.0
7–12 months	56	17.9
>12 months	25	8.0
Total	**312**	**100.0**

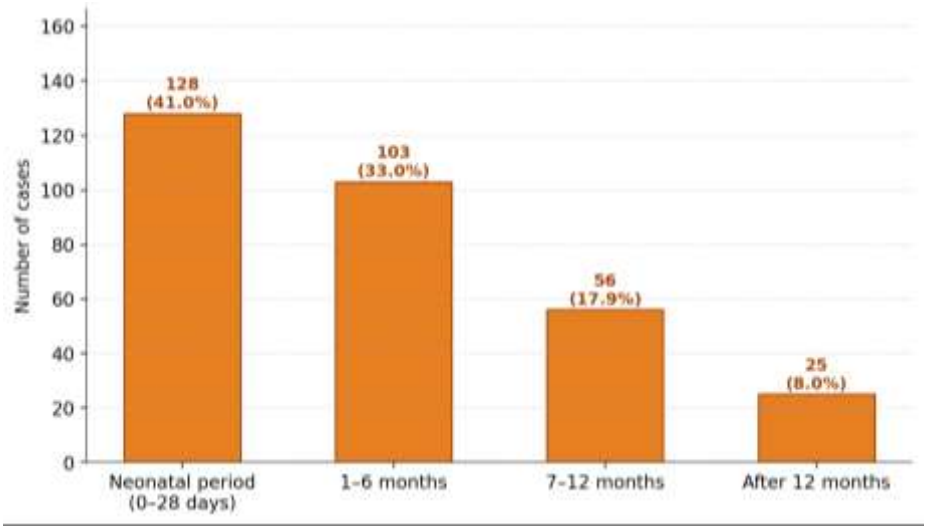

Figure 5. Age at diagnosis of congenital heart disease (n = 312).

Distribution of Congenital Heart diseases Qualitatively

The most common types were ventricular septal defect (VSD) at 31.4% (98 cases), atrial septal defect (ASD) 18.3% (57 cases), patent ductus arteriosus (PDA) 14.1% (44 cases), tetralogy of Fallot 9.0% (28 cases), pulmonary artery stenosis 7.7% (24 cases), atrioventricular septal defect (AVSD) 6.1% (19 cases), aortic stenosis 4.8% (15 cases), transposition of the great arteries (TGA) 3.2% (10 cases), and other complex forms 5.4% (17 cases) (Table 2, Figure2). In the general characteristics of the classification, 77.9% (243 cases) were identified as non-cyanotic forms, and 22.1% (69 cases) as cyanotic forms (Figure 2).

Table 2. Distribution of Congenital Heart Disease Cases According to Anatomical Classification .

Type of Cardiac Defect	Number (n)	Percentage (%)
Ventricular Septal Defect (VSD)	98	31.4
Atrial Septal Defect (ASD)	57	18.3
Patent Ductus Arteriosus (PDA)	44	14.1
Tetralogy of Fallot (TOF)	28	9.0
Pulmonary Stenosis (PS)	24	7.7
Atrioventricular Septal Defect (AVSD)	19	6.1
Coarctation of the Aorta (CoA)	15	4.8
Transposition of the Great Arteries (TGA)	10	3.2
Other Complex Congenital Heart Defects	17	5.4
Acyanotic CHD (Subtotal)	**243**	**77.9**
Cyanotic CHD (Subtotal)	**69**	**22.1**
Total	**312**	**100.0**

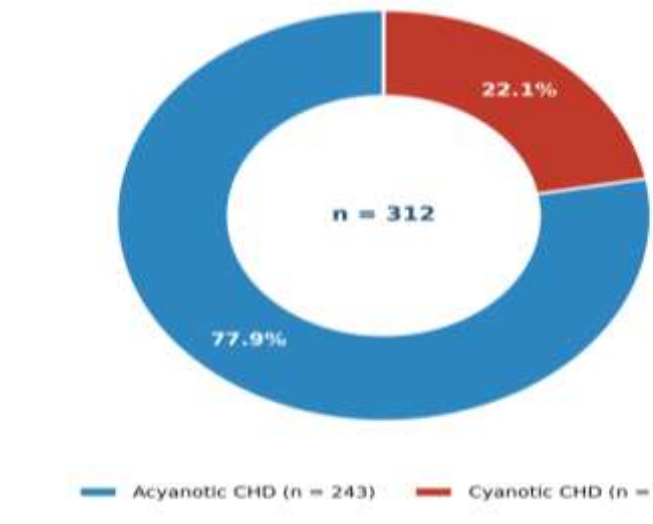

Figure 2. The ratio of non-cyanotic to cyanotic congenital heart disease forms (n = 312).

Parental and Maternal Risk Factors

The analysis of accompanying risk factors showed that there was a consanguineous marriage between the parents in 57.1% of the cases (178 cases), the majority of which were first-degree relatives (cousins). It was also found that 46.2% of the mothers (144 cases) did not use folic acid regularly during the period surrounding the pregnancy, and that 24.0% of the cases (75 cases) were born to mothers aged 35 years or older. Meanwhile, gestational diabetes was recorded in 14.1% (44 cases), and a history of suspected viral infection during pregnancy in 9.0% (28 cases), and a positive family history of congenital heart diseases in 11.9% (37 cases) (Table 3, Figure3). The difference in the rate of consanguineous marriages among mothers of the affected cases (57.1%) compared to the estimated rate of consanguineous marriages in the general population of the province (approximately 38% according to unpublished local estimates) was statistically significant ($p < 0.001$).

Table 3. Parental and maternal risk factors associated with congenital heart disease cases.

Risk Factor	Number (n)	Percentage (%)
Consanguineous marriage (any degree)	178	57.1
Irregular or no folic acid use before/during pregnancy	144	46.2
Maternal age ≥35 years	75	24.0
Gestational diabetes mellitus (GDM)	44	14.1
Febrile illness or suspected viral infection during pregnancy	28	9.0
Positive family history of congenital heart disease	37	11.9

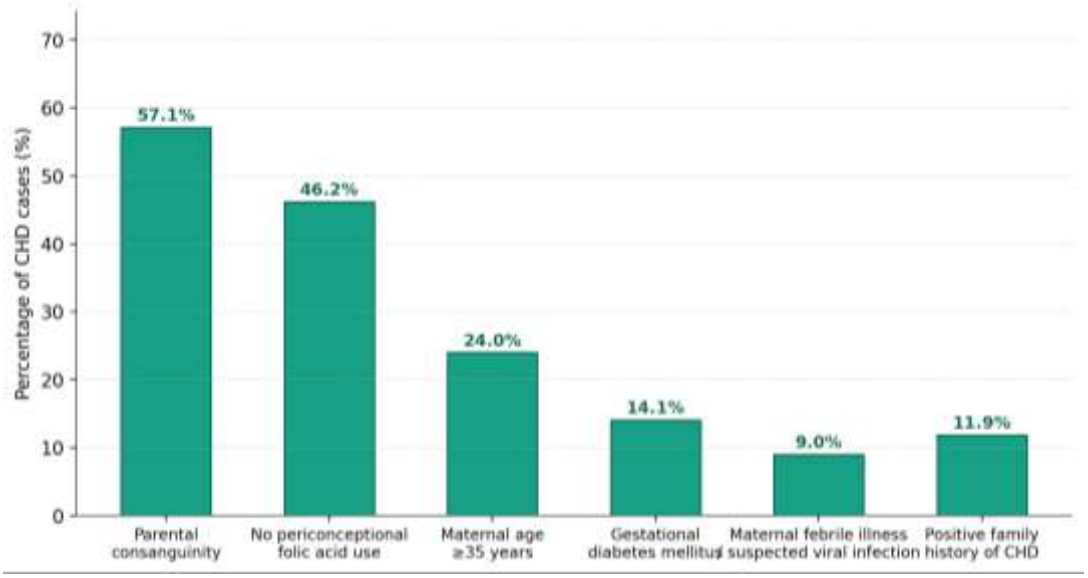

Figure 4. Prevalence of paternal and maternal risk factors in congenital heart disease cases (n = 312).

Clinical complexity and therapeutic outcome

A total of 190 cases (61.0%) were assigned to simple forms not requiring or after urgent surgical or catheter-based intervention, 87 cases (28.0%) to moderately complex forms, and 35 cases (11.0%) to complex forms requiring urgent surgical or catheter-based intervention. Regarding the therapeutic outcome, 39.0% of cases (122 cases) were referred for surgical/herapeutic interventions or catheter intervention, while 53.0% (165 cases) were

followed periodically by medical–clinical surveillance without any direct surgical intervention. During the study period, 8·0% of cases (25 cases) were reported dead, mainly in subjects with complex forms of cardiac defects (table 4).

Table 4. Case complexity and therapeutic outcome for congenital heart disease.

Variable	Number (n)	Percentage (%)
Clinical Complexity		
Simple	190	61.0
Moderate	87	28.0
Complex	35	11.0
Clinical Outcome / Management		
Medical and clinical follow-up only	165	53.0
Referred for surgical or catheter-based intervention	122	39.0
Death during the study period	25	8.0

4. Discussion

The present study found that the prevalence of congenital heart diseases was 11.8 per 1000 live births among infants in Kirkuk Governorate which is on the high side of values range found globally (8–12 per 1000) [1,2] and exceeded the lower limits from a recent comprehensive global review showing a sustainable gradual increase in detection rates in many countries over time leaving marked continental differences [3]. This is in line with several other comparable regional and Iraqi studies with notably high retrospective rates in Fallujeh [25,26] and a large range within a single hospital in Baghdad [14]. On the other hand, the rate of this study was approximately similar to what some Iranian studies have reported [15] and furthermore higher than the rate of congenital heart disease prevalence in Iran estimated to be approximately 2.5 per 1000 in a recent Bayesian review [30]. Importantly, such large discrepancies are usually a reflection of study type (hospital-based cross-sectional or more extensive community surveys) rather than a real biological difference in incidence.

In parallel with the findings of the Baghdad study [24] and the Duhok study in Kurdistan [28], the male-to-female ratio between the two groups in the current study (1.17:1) was also relatively high and consistent with the patterns witnessed in the Turkish and Egyptian studies [16,17]. The predominance of ventricular septal defect as the most common type (31.4%) is in accordance with previous findings which how these patterns have been described in the vast majority of regional and worldwide cross-sectional studies - Baghdad study [24], surgical Duhok study that also how isolated ventricular septal defect was one of the most common patterns in surgical referral contexts [28], Turkish Anatolia study [16], Egyptian Alexandria study [17]. This lends credence to the theory that this defect is the most frequently found pattern in the world regardless of geographic and racial influences.

The most important finding of this study in terms of prevention is the high percentage of consanguineous marriages among parents of affected cases (57.1%). It is close to the recorded high rate in the IraqiRamadi study (78% among cases compared to 43.3% among controls) [27], and it supports what Saudi and Lebanese studies confirmed about the close association between first-degree relatives and the incidence of congenital heart defects in particular [11]-[13]. The support for the Saudi genetic review hypothesis [4] that many cases of congenital heart disease in communities with high rates of consanguinity are exclusively monogenic recessive disorders [13], is also in agreement with another meta-analysis reporting similar conclusions but across heterogeneous communities with a range of consanguinity levels [14]. This limitation is perhaps reflected in the relative higher frequency of consanguineous marriages within the present study sample when compared to the estimated frequency for the general population of the

province, reinforcing the need for improved genetic counseling programs for consanguineous couples pre-marital and pre-pregnancy.

Excluding consanguinity, not using folic acid regularly in peri-conceptional period was a further identified key risk factor (46.2% of cases) also supported by global literature which suggests that deficiency of folic acid around the time of the embryonic period where cellular pathways for normal heart development occur represents a significant risk factor [9]. The finding of 24.0% of mothers being aged 35 years and older is in accordance with the known association between advanced maternal age being an independent risk factor for congenital heart disease, especially when accompanied with other comorbidity such as gestational diabetes as was encountered in 14.1% of cases of the current study. This is in accordance with the well-known effect of gestational and pre-gestational diabetes on the embryonic heart development process, which occurs primarily due to cellular oxidative stress mechanisms [9].

In the present study, clinically the diagnosis of 74.0% of cases before 6 months had been indeed a good reflection related to the efficiency of the screening programs in Kirkuk General Hospital in comparison to the ground data from other centers and areas, where many cases are being diagnosed in the late phase after childhood; 41.0% in the neonatal period and 33.0% during the first 6 months. However, the 8.0% of cases diagnosed after the age of 12 months remain a concern, largely since late diagnosis is associated with an increased risk of irreversible cardiac-pulmonary complications [11] and also as previously highlighted by a Norwegian study, on deficiencies in pre- and postnatal screening programs even in modern health care systems.

An 8.0% mortality rate was noted throughout the study period, the majority of these cases however involved complex forms of cardiac defects which often involved surgical management not always available at local healthcare facilities. This is a common reality among several Middle Eastern countries with limited availability for advanced pediatric heart surgery [22,23]. The surgical success rate for cases managed non-operatively was low (40.6%), with a relatively high surgical success rate of 95.5% reported for cases undergoing surgical intervention in a nearby study in Duhok in the Kurdistan Region of Iraq [28].

These results correlate with the findings of a systematic bibliometric review of congenital heart disease research in the Arab world, which showed a clear or contrasting decline in research productivity in countries afflicted by armed conflicts such as Iraq, compared to the levels dictated by their national income, with published research focused on clinical and surgical elements to the detriment of basic epidemiology studies [20,21]. This study, in producing local epidemiologic data from Kirkuk Governorate, attempts to assist in closing this regional research gap and serves as an introduction to multi-center collaborative studies in the region.

5. Conclusions

These discordant findings suggest that the prevalence rate of congenital heart diseases among infants in Kirkuk Governorate (11.8 per 1000 live births) is at the higher end of previously described global and regional estimates, while also revealing a relatively clear predominance of ventricular septal defect type, strong association with consanguineous marriages and clear absence of folic acid use as the most apparent modifiable parental risk factors. These data confirm the importance of congenital heart diseases as a public health problem in Kirkuk Governorate, so a strategic and periodic coordination intervention at the level of local health polices is needed.

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