



Prevalence of Critical Congenital Heart Disease (CCHD) in Qatar and implementation of Universal Newborn Pulse Oximetry Screening

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Abstract

Background: *Approximately 25% of congenital heart diseases (CHD) are critical that require intervention within the first year of life. There are population differences, and the prevalence is reported to vary between various geographic regions. Universal newborn Critical Congenital Heart Disease (CCHD) screening program is increasingly used worldwide and has a potential to reduce morbidity and mortality. This however puts additional burden on resources and cardiology services. The objective of this study was to establish the prevalence of CCHD in Qatar before considering implementation of CCHD pulse oximetry screening program.*

Methods: *Qatar has only one pediatric cardiac tertiary center which receives all referrals for assessment and management of CHD. Cases diagnosed with CHD are documented on Electronic Medical Record (EMR) system. For data collection, demographic parameters and cardiac diagnoses were extracted from the EMR. The CHD diagnosis was cross checked using the Echocardiography report of each patient. The live births (LB) data were acquired from the National Birth Registry database. The study was conducted at Sidra Medicine & Hamad General Hospital, in Doha, Qatar and study period was between Jan 2014 and Dec 2016.*

Results: *During the study period, there were 78,881 LB in Qatar and 961 babies were diagnosed with a CHD (12.1 per 1000 LB). Of these CHDs, 262 cases (27%) were diagnosed with a CCHD (3.3 per 1,000 LB). The prevalence of CCHD in 2014, 2015 and 2016 was 2.8, 3.1 and 3.9 per 1,000 LB respectively. After birth, most of the D-transposition of great arteries (D-TGA) and hypoplastic left heart syndrome (HLHS) cases were diagnosed before discharge (92% and 93% respectively). On the other hand, only 76% and 50% of the Tetralogy of Fallot (TOF) and Aortic coarctation (COA) cases, respectively, had early diagnosis.*

Conclusion: *The CCHD prevalence in Qatar is comparable to other western countries with highest prevalence. However, many cases could be missed before hospital discharge. These data would allow resource planning and implementation of universal CCHD screening in Qatar.*

Keywords: *Congenital heart disease, Pulse oximetry, Screening, Infant, CCHD.*

Introduction

CHD is the most common congenital disorder in newborn infants which occurs in ~1 in every 110 births in the United States[1].CCHD refers to lesions requiring surgery and/or catheter-based intervention in the first year of life. This category includes infants with ductus-dependent CHD and other cyanotic lesions that require immediate intervention in the neonatal period. Additionally, other forms of CHD that still require intervention in the first year of life, such as a large VSD or an atrioventricular (AV) canal defect (or AV septal defect) are included in CCHD. CCHD accounts for approximately 25 percent of all CHD.[2,3,4,5,6,7]

Although a proportion of newborn infants with CCHD are symptomatic soon after birth, others are not diagnosed until after discharge from the hospital[8,9,10,11]. In infants with critical cardiac lesions, the risk of morbidity and mortality increases when there is a delay in diagnosis and timely referral to a cardiac center with expertise in treating these patients.[12,13]

Infants with CCHD might present during immediate postnatal period, often with serious and life-threatening clinical findings that require immediate intervention.[14] However, some infants with CHD could appear normal on routine examination and signs of CCHD might not be apparent until after discharge. [14] The timing of presentation varies with the underlying lesion and its dependence upon a patent ductus arteriosus (PDA). Diagnosis of CCHD before acute cardiovascular collapse has a potential for improving health outcomes. Usually, the screening tests for early detection of CCHD include prenatal ultrasonography and postnatal clinical examination, but current screening methods may miss up to 50% of affected newborn infants before birth, and those sent home before diagnosis frequently die or endure major morbidity.

Routine pulse oximetry has been reported as an additional screening test that can potentially improve detection of CCHD. In a recent Cochrane review, the overall sensitivity of pulse oximetry for detection of CCHD was reported to be 76.3% (95% confidence interval [CI] 69.5 to 82.0) and specificity 99.9% (95% CI 99.7 to 99.9), with a false-positive rate of 0.14% (95% CI 0.07 to 0.22).[15]

There are population differences in CCHD and the prevalence is reported to vary between geographic regions. Prior to the routine use of pulse oximetry screening, approximately 30 percent of infants with CCHD were discharged home undiagnosed.[16] Universal Newborn CCHD screening program has a potential to reduce morbidity and mortality by early diagnosis, but it puts additional burden on resources and cardiology services. The objective of this study was to establish the prevalence of CCHD in Qatar before implementing CCHD pulse oximetry screening program.

Materials and Methods

This retrospective observational study was conducted at Sidra Medicine and Hamad General Hospital in Doha, Qatar from Jan 2014 to Dec 2016. Qatar has only one pediatric cardiac tertiary center which receives all referrals for intervention and assessment of CHD. Cases diagnosed with CHD are documented on Electronic Medical Record (EMR) system. For data collection, demographic parameters and cardiac diagnoses were extracted from the EMR. The CHD diagnosis was then cross checked using the Echocardiography report of each patient. The live births (LB) data were acquired from the National Birth Registry database.

The data was anonymised and all patient identifiable information was removed. The study did not require IRB approval. The prevalence of CCHD for each year as well as an average for the study period of 3 years were computed. Individual CHD prevalence was also computed.

Results

During the study period, there were 78,881 LB in Qatar. 961 babies (12 per 1000 livebirth (LB) were diagnosed with a CHD (table 1). Of these CHD, 262 cases (27%) were diagnosed with CCHD (3.3 per 1,000 LB). The prevalence of CCHD in 2014, 2015 and 2016 was 2.8, 3.1 and 3.9 per 1,000 LB respectively. The prevalence of specific CCHD over the study period is provided in table 2. After birth, 92% ,93%, 82% ,76% and 50% of the D-TGA, HLHS, TAPVC, TOF and the COA cases, respectively, were diagnosed before hospital discharge (Table 3). The data are comparable to the US states with highest prevalence of CCHD.

Congenital Heart Disease	Year		
	2014	2015	2016
Ventricular septal defect (VSD)	139	94	140
Atrial septal defect (ASD)	102	48	67
Pulmonary stenosis (PS)	16	10	14
Patent ductus arteriosus (PDA)	21	30	42
Tetralogy of Fallot (TOF)	10	11	8
Aortic coarctation (CoA)	3	7	10
D - transposition of great arteries (D-TGA)	8	8	9
Complete atrioventricular canal (CAVC)	5	4	11
Aortic stenosis (AS)	2	4	9
Double outlet right ventricle (DORV)	4	8	9
Partial atrioventricular canal (PAVC)	6	1	4

Hypoplastic left heart syndrome (HLHS)	3	4	8	
Total anomalous pulmonary venous connection (TAPVC)	3	4	4	
Tetralogy of Fallot with pulmonary atresia (TOF/PA)	3	2	3	
Congenitally corrected transposition of great arteries (ccTGA)	2	2	1	
Subaortic membrane	2	1	1	
Partial anomalous pulmonary venous connection (PAPVC)	0	1	1	
Ebstein anomaly	1	0	0	
Pulmonary atresia (PA)	3	2	3	
Interrupted Aortic arch (IAA)	0	3	3	
Hypoplastic Aortic arch	0	2	2	
Single ventricle	2	1	1	
Tricuspid atresia (TA)	1	1	2	
Hypoplastic Tricuspid valve	0	0	2	
Peripheral pulmonary stenosis	0	0	2	
Truncus Arteriosus	0	0	2	
Ventricular inversion	0	1	1	
Aorto Pulmonary Window	1	0	0	
Arterial Tortuosity syndrome (ATS)	1	0	0	
Anomalous origin of left coronary artery from pulmonary artery (ALCAPA)	1	0	0	
Coronary fistula	1	0	0	
Cleft Mitral valve	0	0	1	
Atrioventricular discordance/Ventriculoarterial concordance	0	1	1	
Double inlet left ventricle (DILV)	0	2	1	
Double inlet right ventricle (DIRV)	0	1	0	
Double outlet left ventricle (DOLV)	0	1	0	
Marfan Syndrome	1	0	0	
Scimitar Syndrome	0	1	0	
Situs inversus	0	1	0	
Mitral Stenosis	0	0	1	
Congenital heart block (CHB)	1	0	0	
Annual and total CHD cases during the study period	342	256	363	961
Annual and total Live Births over the study period	25443	26622	26816	78881
Annual and total CHD cases per 1000 live births	13.4	9.6	13.5	12.1

Table 1: Prevalence of congenital heart diseases by lesion during the study period

CCHD	2014		2015		2016		Total	
	No. of cases	Prevalence/1000 LB	No. of cases	Prevalence/1000 LB	No. of cases	Prevalence/1000 LB	No. of cases	Prevalence/1000 LB
Ventricular septal defect	18	7.1	13	4.9	15	5.6	46	5.8
Pulmonary Stenosis	3	1.2	5	1.9	3	1.1	11	1.4
Tetralogy of Fallot	10	3.9	11	4.1	8	3.0	29	3.7
Aortic Coarctation	3	1.2	7	2.6	10	3.7	20	2.5
D-Transposition of Great Arteries	8	3.1	8	3.0	9	3.4	25	3.2
Complete Atrioventricular Canal	5	2.0	4	1.5	11	4.1	20	2.5
Aortic stenosis	2	0.8	4	1.5	5	1.9	11	1.4
Double outlet right ventricle	4	1.6	8	3.0	9	3.4	21	2.7
Hypoplastic left heart syndrome	3	1.2	4	1.5	8	3.0	15	1.9
Total anomalous pulmonary venous connection	3	1.2	4	1.5	4	1.5	11	1.4
Tetralogy of Fallot with pulmonary atresia	3	1.2	2	0.8	3	1.1	8	1.0
Ebstein anomaly	1	0.4	0	0.0	1	0.0	2	0.3
Pulmonary atresia	3	1.2	2	0.8	3	1.1	8	1.0
Interrupted Aortic arch	0	0.0	3	1.1	3	1.1	6	0.8
Hypoplastic Aortic arch	0	0.0	2	0.8	2	0.7	4	0.5
Single ventricle	2	0.8	1	0.4	1	0.4	4	0.5
Tricuspid atresia	1	0.4	1	0.4	2	0.7	4	0.5
Hypoplastic Tricuspid valve	0	0.0	0	0.0	2	0.7	2	0.3
Truncus Arteriosus	0	0.0	0	0.0	2	0.7	2	0.3
Ventricular inversion	0	0.0	1	0.4	1	0.4	2	0.3
Aortopulmonary window	1	0.4	0	0.0	0	0.0	1	0.1
Anomalous origin of left coronary	1	0.4	0	0.0	0	0.0	1	0.1

artery from pulmonary artery								
Atrioventricular discordance with ventriculoarterial concordance	0	0.0	1	0.4	1	0.4	2	0.3
Double inlet left ventricle	0	0.0	2	0.8	1	0.4	3	0.4
Double inlet right ventricle	0	0.0	1	0.4	0	0.0	1	0.1
Double outlet left ventricle	0	0.0	1	0.4	0	0.0	1	0.1
Scimitar Syndrome	0	0.0	1	0.4	0	0.0	1	0.1
Congenital Heart Block	1	0.4	0	0.0	0	0.0	1	0.1
All CCHD	72	2.83	86	3.23	104	3.88	262	3.32
LIVE BIRTH	25443		26622		26816		78881	

Table 2: Prevalence of Critical Congenital Heart Diseases during the study period

Critical Congenital Heart Disease	Cases Diagnosed Before Discharge from Hospital/year			Cumulative pre-discharge diagnosis	Total cases	Diagnosed pre-discharge /total cases
	2014	2015	2016			
Tetralogy of Fallot	6	8	8	22	29	76%
Aortic coarctation	2	3	5	10	20	50%
D-transposition of great arteries	7	7	9	23	25	92%
Complete atrioventricular canal	2	4	4	10	20	50%
Double outlet right ventricle	3	7	8	18	21	86%
Hypoplastic left heart syndrome	2	4	8	14	15	93%
Total anomalous pulmonary venous connection	2	4	3	9	11	82%
Tetralogy of Fallot with pulmonary atresia	2	2	3	7	8	88%
Pulmonary atresia	3	2	3	8	8	100%
Interrupted Aortic arch	0	2	3	5	6	83%
Single ventricle	2	1	1	4	4	100%
Tricuspid atresia	1	1	2	4	4	100%
Truncus arteriosus	0	0	2	2	2	100%

Table 3: Rate of pre discharge diagnosis of Critical congenital heart disease (CCHD)

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Discussion

Congenital Heart Diseases (CHD) account for nearly one third of babies with major congenital anomalies diagnosed prenatally or during infancy in Europe [7] and are the most common congenital disorder in new born infants in the United States.[1]

Advances in management have led to a decrease in infants' mortality and an increase in survival of children and adults with CHD. There is thus growing pressure on pediatric and adult services for CHD survivors and the need for special management when CHD survivors themselves become pregnant. There is also increasing recognition of neurodevelopmental problems in childhood among CHD survivor which could be due to underlying disease or delay in diagnosis. Documenting the prevalence and trends of congenital heart defects provide useful data for pediatric practice, health-care planning, and causal research.[7,8,9,10]

This is the first report of CHD prevalence in the state of Qatar. We found the prevalence of CHD in Qatar during 2014 to 2016 to be 12 per 1,000 live births. These figures are higher than the prevalence in USA, where it occurs ~1 in every 110 births and in Europe where the prevalence of CHD between 2000 and 2005 was 8 per 1,000 births, with some variation between countries and different registries.[17]

The variation between the reported studies was mainly attributed to the differences in inclusion criteria (for example; some people included small VSDs and ASDs in their reports) and the use of different methods to detect CHD.[13] Among patients with congenital heart diseases, 25% of cases comprise the group of CCHDs.[6] If not detected promptly, CCHDs may have catastrophic consequences.[18,19]

The risk of morbidity and mortality increases when there is a delay in diagnosis and timely referral to a cardiac center with expertise in treating these patients.[12] The incidence of CCHD in Qatar was estimated to be 27% of all CHDs (3.3/1,000 LB). The data are comparable to the US states with highest prevalence of CCHD where it constitutes 25% of the reported cases with congenital heart diseases. These data would allow resource planning and implementation of universal CCHD screening in Qatar.

After birth, most of the D-TGA and Hypoplastic left heart syndrome (HLHS) cases were diagnosed before discharge (92% and 93% respectively). On other hand only 76% and 50% of the TOF and COA cases, respectively, had early diagnosis. Thus, with high prevalence of CCHD in Qatar and many of cases that could be missed before hospital discharge, there could be significant disease implications

associated with long term morbidity. Early diagnosis of these conditions has the potential for improving morbidity and possibly mortality.

Pulse oximetry is a simple, non-invasive bedside test that can accurately detect the percentage of hemoglobin saturated with oxygen; infants with CCHDs typically have a low percentage of saturation even before the onset of symptoms.[20] In a recent review of 13 studies using pulse oximetry to screen for CCHDs, pulse oximetry screening (POS) was shown to have a sensitivity of 76.5% and a specificity of 99.9%.[21] When screening is performed after 24 hours of age, there is an estimated very low false-positive rate of 0.05%. As such, POS is already in use in many parts of Europe[22,23,24] and has recently been recommended as Universal Screening Panel for newborns in the United States.[23,24]

There are limitations to this retrospective observational study. The data of antenatal diagnosis of CHD cases is not available. There could be cases with CHD that might have been missed if not referred from primary or secondary care as well as deaths due to CHD, and thus the true prevalence could be marginally more.

There are strengths of this study. As there is a single cardiac centre in Qatar and hence the heterogeneity of diagnostic pathway is limited. Majority of cases with CHD will be included in this study. The disease specific prevalence reported in this study would also allow appropriate planning of resources.

This is the first study that provides the prevalence of CCHD in Qatar. The case load and the spectrum of conditions presented in this paper and the high prevalence rates compared to many developed nations and the delay in diagnosis necessitate implementation of CCHD pulse oximetry screening program. After implementation of POS program, a prospective study could overcome the weaknesses of this study and also assess the long-term morbidity with various CCHD.

Conclusion

The CCHD prevalence in Qatar is comparable to other western countries with highest prevalence. However, many cases could be missed before hospital discharge. These data would allow resource planning and implementation of universal CCHD screening in Qatar.

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