ORIGINAL ARTICLE

SPECTRUM OF CONGENITAL HEART DISEASE IN FULL TERM NEONATES

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Background: Congenital heart disease is a significant problem world over especially in neonates. Early diagnosis and prompt interventions in neonatal period precludes the mortality associated with this disorder. The objective of this study was to highlight the diversity of congenital cardiac defects in our region so that appropriate interventions are devised to minimize significant morbidity and mortality associated with this disorder. **Methods:** This descriptive cross-sectional study was conducted at the Neonatology Unit of Department of Paediatrics, Ayub Teaching Hospital from January 2015 to December 2016. Approval of ethical committee was taken. All full-term neonates of either gender who presented in department of neonatology including those delivered in hospital or received from other sources (private settings, home deliveries), diagnosed as having congenital heart disease on echocardiography were included in the study. Preterm neonates of either gender were excluded from the study. Patient characteristics were recorded in a designed proforma. Data was entered in SPSS version 20 and analysed. **Results:** A total of 89 neonates were included in the study. Mean age of presentation was 6.34±7.058 days and range of 1–28 days. There was a male preponderance with 57 (64%) male patients as compared to 32 (36%) female patients. Ventricular septal defect (VSD) was the commonest cardiac lesion being present in 34 (38.2%) patients. Other defects included complex congenital heart disease in 8 (9%), atrial septal defect (ASD) and transposition of great arteries (TGA) in 7 (7.9%) each, atrioventricular septal defect (AVSD) in 6 (6.7%) and Fallots's tetralogy (TOF) and hypoplastic left heart syndrome in 5 (5.6%) each. **Conclusion:** Congenital heart disease is a problem of profound importance. It constitutes approximately one third of the total major congenital malformations. There is a diversity of cardiac lesions in our region that warrant early and prompt interventions so that the disease is recognized and treated at the earliest to reduce morbidity and mortality.

Keywords: Congenital heart disease; Ventricular septal defect; Cyanotic

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INTRODUCTION

Congenital heart disease (CHD) encompasses a group of cardiac anatomic and functional defects that are present at birth. It is considered to be one of the commonest birth defects.[1] The prevalence of CHD has shown a significant rise being identified as 0.6/1000 livebirths in 1930 to 1934 to 9.1/1000 livebirths in 1995. Since then, the prevalence showed a stabilization pattern. Asia has reportedly the highest birth prevalence of CHD estimated at 9.3/1000 live births.[2] The spectrum of disease ranges from simple defects to severe malformations which must be identified and treated at the earliest to avoid mortality.[3] Broadly the congenital heart defects are classified as cyanotic and acyanotic based on the presence and absence of cyanosis. Cyanotic diseases are manifold including transposition of great arteries (TGA) and Fallot's Tetralogy (TOF) while common acyanotic heart diseases include atrial septal defect, ventricular septal defect, patent ductus arteriosus.[4] The aetiology of congenital heart defects is multifactorial and recurrence in families suggest a genetic basis.[5] Sporadic genetic variations, point mutations, deletions and duplications are some of the known aetiologies resulting in CHD.[6] Furthermore, nearly 5–8% of patients with chromosomal anomalies like Trisomies 21, 18 and 13 are found to have CHD.[7,8] Congenital heart defects are present in about 30% of patients with anomalies of other organ systems. Hence, presence of dysmorphology or anomalies of other organ systems in a patient with CHD gives a clue to an underlying genetic syndrome.[6]

Among the congenital heart defects, ventricular septal defects (VSD) are the commonest with the peri-membranous VSD comprising 80% of the total VSD[9]. Another quite prevalent defect is an isolated atrial septal defect (ASD).Having three major forms, the secundum ASD is the most frequent of all constituting 65 to 75% of the total atrial septal defects[10] Tetralogy of Fallot (TOF), an important cyanotic CHD, comprises four structural defects in heart; overriding of aorta, pulmonary stenosis, right

ventricular hypertrophy and a ventricular septal defect.[11]

Cyanotic congenital heart defects, often represented by the five T's include tetralogy of Fallot, total anomalous pulmonary venous return (TAPVR), Truncus arteriosus, Transposition of great arteries (TGA) and tricuspid valve anomalies especially tricuspid atresia. Of these, TOF and tricuspid atresia have reduced pulmonary blood flow.[12] Other defects like TAPVR, TGA and Truncus also cause systemic cyanosis but they have an increased pulmonary blood flow.[13]

CHD still remains a significant cause of neonatal mortality and morbidity in the world.[14] Due to paucity of data in our part of the country, no attention is being paid to this problem by the health authorities. The present study aims to identify different types of cardiac lesions in full term neonates along with their age of presentation in order to provide an insight into this matter of grave concern so that appropriate interventions could be planned to address this disorder of profound importance.

MATERIAL AND METHODS

The study was conducted at Neonatology unit of Department of Paediatrics, Ayub teaching hospital, Abbottabad from January 2015 to December 2016. Approval of the ethical committee was taken. All full-term neonates of either gender who presented in department of neonatology including those delivered in hospital or received from other sources (private settings, home deliveries), diagnosed as having congenital heart disease on echocardiography were included in the study. Preterm neonates of either gender were excluded from the study. Patient details were obtained from hospital charts and characteristics like age, gender, age of presentation, presence or absence of another anomalies and diagnosis were recorded on a structured proforma. The data was entered in SPSS version 20 and analysed.

RESULTS

A total of 89 full term neonates with proven congenital heart disease were included in the study. Of these, 57 (64 %) were males and 32 (36 %) were females (Table-1). Mean age of presentation was 6.34±7.058 days and range of 1–28 days. Mean weight of the patients was 2.85±0.384 kg. The commonest cardiac lesion was VSD found to be present in 34 (38.2%) patients. Of these, 26 (29.2%) had muscular VSD and 8 (9%) had a membranous VSD. Moderate VSD was present in 28 (31.5%) patients, small VSD in 4 (4.5%) patients and large VSD in 2 (2.2 %) patients. This was followed by complex congenital heart disease in 8 (9%) patients including 6 male and 2 female patients. ASD was noted in 7 (7.9%) patients including 2 males and 5 female patients. TGA was diagnosed in 7 (7.9%) patients. Of these, 5 were males and 2 were female patients. A total of 6 (6.7%) patients were diagnosed as having AVSD. Fallot's tetralogy and HLHS was diagnosed in 5 (5.6%) patients each. Truncus arteriosus and dextrocardia was diagnosed in 3 (3.4%) each. Tricuspid atresia and pulmonic stenosis was diagnosed in 2 (2.2%) each (Table-2). Only ASD and PDA were found to be more prevalent in females where 5 (71.42%) out of 7 and 3 (75%) out of 4 patients respectively were females. Rest of the defects were predominantly found in males (Table-3). A total of 28 (31.5%) patients had associated anomalies of other organ systems or syndrome while 61 (68.5 %) had isolated cardiac defect.

Table-1: Gender distribution of patients

Gender	No. of patients	Frequency
Male	57	64%
Female	32	36%
Total	89	100%

Table-2: Diagnosis of patients

Diagnosis	No. of patients	Frequency
Ventricular septal defect	34	38.2%
Complex congenital heart disease	8	9%
Atrial septal defect	7	7.9%
Transposition of great arteries	7	7.9%
Atrioventricular septal defect	6	6.7%
Fallot's tetralogy	5	5.6%
Hypoplastic left heart syndrome	5	5.6%
Patent ductus arteriosus	4	4.5%
Truncus arteriosus	3	3.4%
Dextrocardia	3	3.4%
Tricuspid atresia	2	2.2%
Pulmonary stenosis	2	2.2%
Others	3	3.4%
Total	89	100%

Table-3: Gender distribution of diseases

Diagnosis	Gender distribution		Total
	Male	Female	
Ventricular septal defect	24 (70.58%)	10 (29.41%)	34 (100%)
Complex congenital heart disease	6 (75%)	2 (25%)	8 (100%)
Atrial septal defect	2 (28.57%)	5 (71.42%)	7 (100%)
Transposition of great arteries	5 (71.42%)	2 (28.57)	7 (100%)
Atrioventricular septal defect	3 (50%)	3 (50%)	6 (100%)
Fallot's tetralogy	4 (80%)	1 (20%)	5 (100%)
Hypoplastic left heart syndrome	4 (80%)	1 (20%)	5 (100%)
Patent ductus arteriosus	1 (25%)	3 (75%)	4 (100%)
Truncus arteriosus	2 (66.6%)	1 (33.3%)	3 (100%)
Dextrocardia	2 (66.6%)	1 (33.3%)	3 (100%)
Tricuspid atresia	1 (50%)	1 (50%)	2 (100%)
Pulmonary stenosis	2 (100%)	0 (0%)	2 (100%)
Others	1 (33.3%)	2 (66.6%)	3 (100%)
Total	57 (64%)	32 (36%)	89 (100%)

DISCUSSION

A total of 89 patients were enrolled in this study. There was a preponderance of congenital heart disease in male neonates with a total of 57 (64%) males and 32 (36%) female patients. Similar results are reported in other studies as well. A study from Hyderabad also reported CHD to be more prevalent in males as compared to females 55.3% males vs 44.7% females.[15]

Among the congenital heart defects, only ASD (28.57% males vs 71.42% females) and PDA (25% males vs 75% females) were more frequently encountered in females as compared to males. Rest of the CHD were more common in males. Similar results are reported in a study from Taiwan where ASD and PDA were the predominant defects in females while TGA and TOF were more frequently encountered in males.[16] However, contrary to our study, AVSD and VSD were also more prevalent in females in that study.

Ventricular septal defect was the commonest congenital cardiac lesion being present in 34 (38.2%) patients. This is comparable to other studies in other parts of the world who also report VSD to be the commonest defect. A study in Hazara division in 2008 reported VSD as the most common lesion being present in 61.4% patients.[17] However, the frequency is quite high as compared to our study. A study from India reported VSD to be the most prevalent lesion with a frequency of 30.45% which is similar to our study.[18] The frequency of different cardiac defects in our study were found to be as follows: VSD 38.2%, ASD 7.9%, TGA 7.9%, CCHD 9%, AVSD 6.7%, PDA 4.5%, TOF 5.6%, HLHS 5.6%, PDA 4.5%, Truncus Arteriosus 3.4%, dextrocardia 3.4%, PS 2.2%, Tricuspid atresia 2.2%. Similar results are reported in a study from Czechoslovakia where frequency of different cardiac defects were found to be ventricular septal defect (VSD) (31.41%), atrial septal defect (ASD) (11.37%), aortic stenosis (AS) (7.64%), pulmonary stenosis (PS) (7.13%), coarctation of the aorta (CoA) (5.77%), and transposition of the great arteries (TGA) (5.43%), followed by persistent ductus arteriosus (PDA) (4.75%), atrioventricular septal defect (AVSD) and hypoplastic left heart syndrome (HLHS) (4.07% each), tetralogy of Fallot (TOF) (3.56%), and pulmonary atresia (PA) (2.38%).[19] In a study in Karachi, TOF was the commonest CHD being present in 24.4 % followed by VSD in 21.5% patients.[20] This was in contrast to our study. However, the frequencies of other defects in this study were quite similar to our results.

In our study, cardiac defects were associated with other congenital anomalies or syndrome in 28 (31.5%) patients whereas 61 (68.5%) patients had isolated cardiac defects. Similar results are reported in other studies as well. In a study on neonates undergoing interventions for congenital heart defects, 18.8% were found to have genetic syndromes/noncardiac malformations.[21]

CONCLUSION

Congenital heart disease is a problem of profound importance in our region. There is a diversity of cardiac lesions in our region that warrant early and prompt interventions so that the disease is recognized and treated at the earliest to reduce morbidity and mortality. However, there is a paucity of fully equipped paediatric cardiac centres in our province. A delay in diagnosis and non-availability of early interventions contribute significantly to mortality. There is a dire need of establishment of such centres to offer prompt treatments and reduce mortality associated with congenital heart disease.

AUTHOR CONTRIBUTION

SB: Principal author, designed the whole study, data collection and analysis. SYHG & SB: Literature review.

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