Pattern of congenital heart diseases in Western Rajasthan: an echocardiographic study

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Received: 10 February 2020
Revised: 25 February 2020
Accepted: 02 March 2020

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ABSTRACT

Background: Congenital heart disease (CHD) accounts for nearly one third of all major congenital anomalies. Globally the prevalence of CHD is 1.01 to 17.5 per 1000 live births. In India it is 1.3 to 26.4 per 1000 study population. CHD is an important cause of mortality and morbidity representing a global health burden. Early diagnosis and treatment may lead to improved prognosis in patients suffering from CHD. The aim of this study was to assess the pattern of CHD in Western Rajasthan, India by echocardiography.

Methods: This was a retrospective study carried out at Dr. S. N. Medical College and attached group of hospitals in Jodhpur, Rajasthan, India. The study period was from July 2014 to June 2017. Records of all patients undergoing transthoracic echocardiography from newborn to 25 years of age were analyzed for age, sex and CHD findings.

Results: In the study period, a total of 24,914 patients underwent echocardiography, of which 877 patients were identified as having CHD. Prevalence of CHD was 35.20 per 1000 study population. Amongst the total diagnosed CHD cases, 489 (55.76%) patients were male, with male to female ratio of 1.2:1. CHDs were diagnosed more commonly between 1 month and 1 year of age (41.28%). The commonest type of acyanotic CHD in the present study was ventricular septal defect (21.44%) and cyanotic CHD was tetralogy of Fallot (18.24%).

Conclusions: Prevalence of CHD in Western Rajasthan, India was 35.20 per 1000 study population. Profile of CHDs in the present study was similar to that in published literature.

Keywords: Congenital heart disease, Echocardiography, Prevalence, Western Rajasthan

INTRODUCTION

Congenital heart disease (CHD) is the most common congenital anomaly, representing a major global health problem. Twenty-eight percent of all major congenital anomalies consist of heart defects.

Understanding the distribution of CHDs in the population is key to assess the burden of these anomalies, including the factors influencing local case mix and severity, in order to anticipate health needs and provide effective and appropriately targeted services for the prevention and management of these conditions.

The most practical measurement of CHD occurrence is birth prevalence per 1,000 live births which is about 9 per 1000 live birth.5,6 Worldwide prevalence of CHD ranges from 1.01 to 18.1 per 1000 study population.5,6 The prevalence of CHD in India has been reported to be from 1.3 to 26.4 per 1000 study population.7,8 Since CHDs are important cause of mortality and morbidity, early diagnosis and treatment may lead to improved prognosis.

METHODS

This was a retrospective study carried out in departments of cardiology and pediatrics of Dr. S. N. Medical College and attached group of hospitals, Jodhpur, India. The
study period was of 3 years duration, from July 2014 to June 2017. Dr. S. N. Medical College Jodhpur is a tertiary care and teaching institute in Western Rajasthan, India. Population drained by this institute is approximately 15 million. The institute caters to patients belonging to all strata of society from Western Rajasthan. The source of information was medical and echocardiographic records of all patients who visited the outdoor and indoor patient departments of pediatrics and cardiology. Clinical examination and 2-D and color Doppler echocardiography were considered as definitive tool for diagnosis of CHD. Records of all patients undergoing transthoracic echocardiography from newborn to 25 years of age were analyzed for age, sex and CHD findings. Congenital heart disease was defined as "a gross structural abnormality of heart or intrathoracic great vessels that is actually or potentially of functional significance excluding the systemic great arteries and veins" as defined by Mitchell et al. Echocardiography was done as per standards laid down by the American Society of Echocardiography, using the M-mode, two-dimensional, color Doppler, pulse and continuous wave Doppler echocardiogram.

### Inclusion criteria
- Only first time visiting all comers diagnosed by clinical and transthoracic echocardiographic examination from newborn to 25 years of age were included in the study.

### Exclusion criteria
- Previously enrolled patients presenting on follow up visits.

### Data analysis
As this was a descriptive study, data was entered into a Microsoft office excel spreadsheet and analyzed. Ratios and percentages were used for evaluation.

### RESULTS
A total of 24,914 patients were examined and underwent detailed transthoracic echocardiographic examination during the study period of 3 years. CHD was diagnosed in 877 patients during this period, thus giving a prevalence of 35.20 per 1000 study population. Amongst the total diagnosed CHD cases, 489 (55.76%) patients were males and 388 (44.24%) were females with male to female ratio of 1.2:1.

#### Table 1: Age wise distribution of 877 cases of CHDs.

| Age of CHD                  | No. of cases | Percentage |
|-----------------------------|--------------|------------|
| Less than 1 month           | 176          | 20.06      |
| 1 month - 1 year            | 362          | 41.28      |
| 1 year - 5 year             | 161          | 18.36      |
| 5 year - 10 year            | 87           | 9.92       |
| 10 year - 15 year           | 54           | 6.16       |
| 15 year - 20 year           | 21           | 2.40       |
| 20 year - 25 year           | 16           | 1.82       |
| Total                       | 877          | 100        |

#### Table 2: Age wise and sex wise distribution of acyanotic CHDs.

| CHD                 | <1 month | 1 month - 1 year | 5 year - 10 year | 15 year - 20 year | 20 year - 25 year | Total | Male | Female | Total | Percentage of all CHD |
|---------------------|----------|------------------|------------------|-------------------|-------------------|-------|------|--------|-------|-----------------------|
| VSD                 | 30       | 85               | 30               | 19                | 14                | 6     | 4    | 102    | 86    | 188                   | 21.44 |
| ASD                 | 14       | 51               | 25               | 17                | 10                | 6     | 3    | 58     | 68    | 126                   | 14.37 |
| PDA                 | 25       | 40               | 27               | 8                 | 6                 | 2     | 1    | 47     | 62    | 109                   | 12.43 |
| BAV                 | 9        | 13               | 10               | 5                 | 4                 | 3     | 6    | 39     | 11    | 50                    | 5.70  |
| PS                  | 7        | 16               | 9                | 7                 | 3                 | 1     | 0    | 24     | 19    | 43                    | 4.90  |
| PFO                 | 7        | 16               | 3                | 1                 | 1                 | 1     | 0    | 14     | 15    | 29                    | 3.31  |
| AVCD                | 8        | 10               | 5                | 2                 | 1                 | 0     | 0    | 12     | 14    | 26                    | 2.97  |
| PAPVC               | 2        | 4                | 1                | 0                 | 0                 | 0     | 0    | 5      | 3     | 8                     | 0.91  |
| COA                 | 0        | 2                | 2                | 0                 | 0                 | 0     | 0    | 3      | 1     | 4                     | 0.46  |
| Cor triatriatm       | 0        | 2                | 0                | 0                 | 0                 | 0     | 0    | 2      | 0     | 2                     | 0.23  |
| Congenital MS        | 0        | 0                | 1                | 0                 | 0                 | 0     | 0    | 1      | 0     | 1                     | 0.11  |
| Total               | 102      | 239              | 113              | 59                | 40                | 19    | 14   | 307    | 279   | 586                   | 66.82 |

Abbreviations: ASD atrial septal defect; AVCD atrioventricular canal defect; BAV bicuspid aortic valve; COA coarctation of aorta; MS mitral stenosis; PAPVC partial anomalous pulmonary venous connection; PDA patent ductus arteriosus; PFO patent foramen ovale; PS pulmonary stenosis; VSD ventricular septal defect
Age wise distribution of the 877 cases of CHDs is shown in table 1. Of the total patients diagnosed in the present study, 20.06% were below 1 month, 41.28% were between 1 month to 1 year and 28.28% were 1-10 years of age.

Table 2 presents distribution of acyanotic CHDs. Ventricular septal defect (VSD) (21.44%) was commonest type of acyanotic CHD in the study, followed by atrial septal defect (ASD), patent ductus arteriosus (PDA), bicuspid aortic valve (BAV) and pulmonary stenosis (PS). Acyanotic CHDs were most commonly diagnosed between 1 month to 1 year of age (27.25%).

Table 3 presents distribution of cyanotic CHDs. Tetralogy of Fallot (TOF) (18.24%) was the commonest type of cyanotic CHD in the present study, followed by D- transposition of great arteries (D-TGA), double outlet right ventricle (DORV), tricuspid atresia (TA) and single ventricle (SV). Cyanotic CHDs were most commonly diagnosed between 1 month to 1 year of age (14.02%).

**DISCUSSION**

Over time, the reported total CHD prevalence at birth has increased substantially, from 1 per 1,000 live births in 1930 to 9 per 1,000 live births in recent years. With a worldwide annual birth rate around 150 million births, this corresponds to 1.35 million live births with CHD every year, representing a major public health issue. The increase in reported total CHD birth prevalence over time might be caused by changes in diagnostic methods and screening modalities such as echocardiography which helped in diagnosing very small defects rather than representing a true increase. Amongst the continents, highest reported total CHD birth prevalence was found in Asia (9.3 per 1,000 live births) and the lowest in Africa (1.9 per 1,000 live births).\(^4\)\(^,\)\(^13\) Table 4 presents prevalence of CHD in world in various studies. Globally high prevalence of CHD was found in hospital-based studies such as in Australia (17.5/1000) and Pakistan (10.0/1000).\(^12\)\(^,\)\(^13\)

Majority of prevalence studies in India were either school based or hospital based as shown in table 5. Kapoor et al.\(^8\) observed higher CHD prevalence of 26.4 per 1000 study population in India due to inclusion of mild, moderate and severe CHDs. Hospital based studies have higher CHD prevalence such as in Mysore (10.65/1000), Mumbai (13.28) and in Varanasi (19.14/1000).\(^18\)\(^-\)\(^20\) Higher CHD prevalence of 35.20 per 1000 study population was observed in the present study compared to previously done studies in India and other countries of world. The reason for higher prevalence in the present study was that prevalence of CHD was assessed in

### Table 3: Age wise and sex wise distribution of cyanotic CHDs.

| CHD                  | <1 month | 1 month-1 year | 1 year | 5 year | 10 year | 15 year | 20 year | 25 year | Total | Male | Female | Total | Percentage of all CHD |
|----------------------|---------|----------------|--------|--------|---------|---------|---------|---------|-------|------|--------|-------|----------------------|
| TOF                  | 21      | 73             | 34     | 19     | 10      | 2       | 1       | 95      | 65    | 160  | 18.24  |       |                      |
| D-TGA                | 17      | 12             | 3      | 3      | 0       | 0       | 0       | 28      | 7     | 35   | 3.99   |       |                      |
| DORV                 | 12      | 13             | 6      | 3      | 1       | 0       | 0       | 22      | 13    | 35   | 3.99   |       |                      |
| Tricuspid atresia    | 11      | 11             | 1      | 1      | 0       | 0       | 0       | 13      | 11    | 24   | 2.74   |       |                      |
| Single ventricle     | 4       | 2              | 3      | 0      | 0       | 0       | 0       | 5       | 4     | 9    | 1.03   |       |                      |
| Truncus arteriosus   | 3       | 3              | 0      | 1      | 0       | 0       | 0       | 4       | 3     | 7    | 0.79   |       |                      |
| Ebstein anomaly      | 0       | 2              | 1      | 1      | 2       | 0       | 0       | 3       | 3     | 6    | 0.68   |       |                      |
| L-TGA                | 0       | 2              | 0      | 0      | 1       | 0       | 1       | 3       | 1     | 4    | 0.46   |       |                      |
| TAPVC                | 0       | 4              | 0      | 0      | 0       | 0       | 0       | 4       | 0     | 4    | 0.46   |       |                      |
| Pulmonary atresia    | 3       | 1              | 0      | 0      | 0       | 0       | 0       | 2       | 2     | 4    | 0.46   |       |                      |
| HRHS                 | 2       | 0              | 0      | 0      | 0       | 0       | 0       | 2       | 0     | 2    | 0.23   |       |                      |
| HLHS                 | 1       | 0              | 0      | 0      | 0       | 0       | 0       | 1       | 0     | 1    | 0.11   |       |                      |
| Total                | 74      | 123            | 48     | 28     | 14      | 2       | 2       | 182     | 109   | 291  | 33.18  |       |                      |

Abbreviations: D-TGA D-transposition of great arteries; DORV double outlet right ventricle; HLHS hypoplastic left heart syndrome; HRHS hypoplastic right heart syndrome; L-TGA L-transposition of great arteries; TAPVC total anomalous pulmonary venous connection; TOF tetralogy of Fallot

Female preponderance in CHD was noted in ASD and PDA whereas male preponderance was noted in VSD, BAV, PS, TOF, D-TGA, DORV and total anomalous pulmonary venous connection (TAPVC).
patients who had undergone echocardiography as compared to other studies in which prevalence was assessed among patients attending the hospital outpatient and inpatient department and also inclusion of all small or mild CHDs, and inclusion of cases up to 25 years of age.

There was male preponderance with male to female ratio of 1.2:1 in the present study, as was also shown by Chadha et al, whereas female preponderance in CHDs was observed by Thakur et al. The age at detection of CHD varies due to normal hemodynamic changes such as time lag in fall of pulmonary vascular resistance and closure of PDA occurring after birth. In the present study maximum number of cases (41.28%) were diagnosed between 1 month to 1 year of age and only 10.38% cases were diagnosed after 10 years of age. These findings were also consistent with study by Bhat et al in which 43% cases were detected by infancy and Smitha et al, in which maximum CHDs were detected in first year of life when compared to later years of life.

Most common CHD observed in the present study was VSD accounting for 21.4% of cases. This was consistent with other studies done in India as shown in Table 6 except the study done by Thakur et al, in which ASD was the commonest CHD observed. TOF was the most common

Table 4: Prevalence of CHD in world studies.

| Author, Ref no. | Country | Study population | Method | Number studied | Prevalence / 1000 study population |
|----------------|---------|------------------|--------|----------------|-----------------------------------|
| Fixer et al,14 | USA     | Community (4-17 year) | Clinical + Echo + Cath + Surgery + Autopsy Prospective | 379,561 | 6.6 |
| Khalil et al,15 | Sudan   | School (5-15 years) | Clinical + Echo Prospective | 13,322 | 2.0 |
| Bassili et al,3 | Egypt   | School (5-15 years) | Clinical + Echo Prospective | 869,434 | 1.01 |
| Bolisetty et al,12 | Australia | Hospital (0-1 or more years) | Clinical + Echo Reretrospective | 6156 | 17.5 |
| Lindinger et al,16 | Germany | Community (0-1 year) | Clinical + Echo + Cath + Surgery + Autopsy Prospective | 670,000 | 10.8 |
| Masood et al,13 | Pakistan | Hospital (0-13 year) | Clinical + Echo Descriptive | 9614 | 10.0 |
| Liu et al,17 | China | Community (0-6 months) | Clinical + Echo Prospective | 90,796 | 16.4 |
| Ujuabi et al,6 | Nigeria | School (5-14 years) | Clinical + Echo | 1712 | 18.1 |

Abbreviations: Cath catheterization; Echo echocardiography

Table 5: Prevalence of CHD in Indian studies.

| Author, Ref No. | City | Study population | Method | Number studied | Prevalence / 1000 study population |
|----------------|------|------------------|--------|----------------|-----------------------------------|
| Shrestha et al,21 | Delhi | School (5-16 years) | Clinical + Echo + Cath + Surgery Collaborative | 34,198 | 3.2 |
| Vashishtha et al,22 | Agra | School (5-15 years) | Clinical + Echo Prospective | 8449 | 5.2 |
| Thakur et al,23 | Shimla | School (5-16 years) | Clinical + Echo Prospective | 15,080 | 2.25 |
| Chadha et al,24 | Delhi | Community (0-15 years) | Clinical + Echo Prospective | 11,883 | 4.2 |
| Smitha et al,18 | Mysore | Hospital (0 to 10 or more years) | Clinical + Echo Retrospective | 74,589 | 10.65 |
| Kapoor et al,8 | Kanpur | Hospital (0-15 years) | Clinical + Echo Retrospective | 10,641 | 26.4 |
| Misra et al,7 | Gorakhpur | School (4-18 years) | Clinical Prospective | 118,212 | 1.3 |
| Bhat et al,25 | Dehradoon | Hospital (0-18 years) | Clinical + Echo Prospective | 36,541 | 8.54 |
| Sawant et al,19 | Mumbai | Hospital (Live birth) | Clinical + Echo Prospective | 2636 | 13.28 |
| Bhawardaj et al,20 | Varanasi | Hospital (0-68 years) | Clinical + Echo Prospective | 34,517 | 19.14 |
| Present study | Jodhpur | Hospital (0-25 years) | Clinical + Echo Retrospective | 24,914 | 35.20 |

Abbreviations: Cath catheterization; Echo echocardiography
cyanotic CHD observed in 18.2% cases in the present study, correlating well with other Indian studies.18-22

Table 6: Profile of individual CHD in Indian studies.

| Author, Ref no. | Profile of individual CHD (% of all CHD) |
|-----------------|-----------------------------------------|
|                 | VSD  | ASD  | PDA  | PS   | AVCD | CoA  | TOF  | D-TGA | DORV | Ebstein Anomaly | TA | PA | TAPVC |
| Shrestha et al,21 | 30   | 23   | 11   | 10   | -    | -    | 4    | -     | -    | -              | -  | -  | -     |
| Vashishtha et al,22| 40.9 | 11.4 | 4.5  | 9.1  | -    | -    | 13.6 | -     | -    | -              | -  | -  | -     |
| Thakur et al,23   | 32.2 | 38.2 | -    | -    | -    | -    | -    | -     | -    | -              | -  | -  | -     |
| Chadha et al,24   | 46   | 18   | 14   | 4    | -    | -    | 10   | -     | -    | -              | -  | -  | -     |
| Smitha et al,19   | 40.5 | 19.1 | 9.5  | -    | -    | -    | 13.4 | -     | -    | -              | -  | -  | -     |
| Kapoor et al,5    | 21.3 | 18.9 | 14.6 | 3.2  | 10.3 | -    | 4.6  | 1.1   | 0.4  | 1.1           | -  | -  | 0.4   |
| Misra et al,7     | 40.8 | 18.3 | 2.1  | 8.5  | -    | -    | -    | 0.7   | 1.4  | -              | -  | -  | -     |
| Bhat et al,25     | 30.5 | 17.6 | 9.6  | 6.4  | 1.6  | 2.6  | 5.5  | 5.1   | 0.6  | 1.3           | 1.9| 3.2| 1.3   |
| Sawant et al,19   | 42.9 | 25.7 | 5.7  | 2.9  | -    | 2.9  | 8.6  | -     | -    | 2.9           | -  | -  | -     |
| Bhardwaj et al,20 | 33.3 | 19.1 | 4.1  | 1.5  | 2.4  | 0.6  | 16.8 | 2.9   | 1.5  | 0.6           | 0.8| -  | 0.9   |
| Present study     | 21.4 | 14.4 | 12.4 | 4.9  | 2.97 | 0.5  | 18.2 | 3.99  | 3.99 | 0.68          | 2.7| 0.5| 0.46  |

Abbreviations: ASD atrial septal defect; AVCD atrioventricular canal defect; CoA coarctation of aorta; DORV double outlet right ventricle; D-TGA D-transposition of great arteries; PA pulmonary atresia; PDA patent ductus arteriosus; PS pulmonary stenosis; TA tricuspid atresia; TAPVC total anomalous pulmonary venous connection; TOF tetralogy of Fallot; VSD ventricular septal defect

The present study was peculiar as the prevalence was highest as it was assessed in the patient population who underwent echocardiography as compared to other Indian studies in which prevalence was seen among patients attending inpatient and outpatient department. Authors have also studied patients up to 25 years of age which is higher as compared to most other Indian studies.

Limitation of the present study is that being a hospital-based study it does not reflect true community prevalence.

CONCLUSION

Prevalence of CHD in Western Rajasthan, India was 35.20 per 1000 study population. Profile of various CHDs in the present study was largely similar to the preexisting studies. In this era of most accurate diagnostic modalities, any clinical suspicion of CHD should be confirmed by echocardiography to hasten the diagnosis, timely management and prevention of complications.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Baroopal A, Mathur R, Sanghvi S, Soni JP. Pattern of congenital heart diseases in Western Rajasthan: an echocardiographic study. Int J Res Med Sci 2020;8:1385-90.