#### **Research Article**

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### Prevalence and pattern of congenital heart diseases in Karimnagar, Andhra Pradesh, India: diagnosed clinically and by trans-thoracic-twodimensional echocardiography

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#### ABSTRACT

**Background:** To find the prevalence and pattern of congenital heart diseases (CHD) at a Semi-Urban teaching hospital in Karimnagar, Andhra Pradesh, India.

**Methods:** A thorough history, clinical examination and Trans-Thoracic-Two-Dimensional Echocardiography (TTE) was done for all the live birth, children up to 18years of age and patients between 18 to 25 years, who were referred or presented to the Department of Medicine, Chalmeda Anand Rao Institute of Medical Sciences, Bommakal, Karimnagar (AP), over a period of 5 years from July 2008 through June 2013. Those suspected to having a CHD or referred in our department, were further evaluated with: Clinically, Twelve-Lead-Surface Electrocardiography, Chest Radiography and the diagnosis was confirmed by TTE. Trans-Thoracic-Two-Dimensional Echocardiography, M-Mode, Color flow doppler and Spectral doppler echocardiography was done in all patients in the various views.

**Results:** Total 13,554 patients were examined and underwent TTE. Out of 13,554 patients 116 were identified as having congenital heart diseases, thus giving a prevalence of 8.55 per 1,000 live births. Isolated Ventricular septal defect (28.44%), isolated atrial septal defect (18.10%), Patent ductus arteriosus (10.34%), isolated congenital pulmonary stenosis (6.03%) and tetralogy of Fallot's (6.03%), were the commonest defects observed and confirmed by TTE. TOF was the main cyanotic CHD (6.03%), with the prevalence of 0.51% per 1,000 live births. VSD, ASD and PDA were more prevalent in males. TOF and Complete A.V. Canal defect was prevalent in females. All small size muscular and perimembranous VSD was closed spontaneously. Spontaneous closure rate of 75.00% in Muscular VSD and 52.17% in perimembranous VSD was observed. Spontaneous closure rate of Ostium secundum type ASD was 53.33%.

**Conclusions:** The prevalence of CHD at a tertiary teaching hospital (CAIMS, Bommakal, Karimnagar, AP, India), is 8.55 per 1,000 live births. VSD, ASD, PDA are the most common acyanotic and TOF was the commonest cyanotic congenital heart defects respectively. Non-Invasive Cardiac diagnostic technique (like TTE) plays major in the diagnosis of CHD. When clinical evidences lead to suspicion of congenital heart defect, an echocardiography should be performed immediately.

Keywords: Congenital heart diseases, Prevalence and pattern of CHD in Karimnagar

#### **INTRODUCTION**

Congenital heart disease is one of the most common congenital defects and along with neural tube defects accounts for two-thirds of all congenital malformations.<sup>1</sup> The prevalence of CHD is not uniform in our country as various studies have reported it ranging from 1.3 to 50.89 per 1000 live births.<sup>5,6</sup> Also several studies from abroad report a changing pattern and incidence of CHD in various geographical locations.<sup>7,8</sup> Early recognition of such diseases has great implications. Despite advanced diagnostic facilities and improved medical care, CHD is considered one of the leading causes of neonatal mortality.<sup>2</sup> According to a status report on CHD in India, 10% of the present infant mortality may be accounted for by CHD.<sup>3</sup> CHD may present at different ages from birth to adolescence.<sup>2</sup> Many cases are asymptomatic and discovered incidentally during routine health check-up.<sup>4</sup> Other presentations can range from cyanosis, clubbing of fingers, fatigue to full blown congestive cardiac failure.<sup>2,4</sup> CHD has not been studied thoroughly in India, as in western countries. There are only few studies of prevalence and pattern of CHD in India especially in urban, semi-urban and remote villages. The present study reports prevalence and pattern at a semi-urban teaching hospital in Karimnagar, Andhra Pradesh, India. Accurate assessment of prevalence of CHD in a population is critical in understanding the social and economic burdens placed on the patients and their families, demands placed on the health care system and health planning.

#### **METHODS**

This was a prospective, longitudinal study conducted by the Department of Medicine at Chalmeda Anand Rao Institute of Medical Sciences, a semi-urban tertiary care teaching hospital - Bommakal - Karimnagar - India, over a period of five year from July 2008 to June 2013. All live birth at the hospital, children up to 18 years and patient between 18 to 25 years, who referred or presented to our department for further evaluation in outpatient department (OPD) and IPD (in patient department), over a period of five years from July 2008 through June 2013, were included in the present study. A thorough history and clinical examination was carried out, and CHD was suspected in the presence of a cardiac murmur, presence of cyanosis, feeding difficulties, squatting position, cyanosis associated with feeding difficulty, clubbing, differential cyanosis, failure to thrive and features of congestive cardiac failure.9 Those with history, symptoms or signs of heart disease were further evaluated with twelve lead surface electrocardiography (ECG), chest radiography and conformation of the diagnosis was done by Trans-Thoracic-two-Dimensional echocardiography, M-Mode, Spectral Doppler and color flow Doppler Echocardiography. TTE was done in various views such

as: subcostal, apical four chamber, Apical two chamber, Apical long axis, Parasternal long axis, Parasternal short axis (at various level of left ventricle like: basal, mid cavity or at level of papillary level and apical part ), parasternal high short axis (at aortic valve, pulmonary valve level) and suprasternal view. Congenital heart disease was defined as the structural abnormalities of heart or intrathoracic great vessels, present since birth that is actually or potentially of functional significances regardless of the age of detection, as defined by Mitchell et al.<sup>9</sup> Only patients with first time diagnosed were included and those presenting on follow up visits were excluded. Neonates less than 2 week of the age with a diagnosis of PDA were also excluded. The data was entered in to a Microsoft office excel spread sheet and analysed.

#### RESULTS

During the study period of 5 years total 13554 patients were examined and underwent TTE. Congenital heart defects were identified in 116 patients. Congenital heart defects were identifies in 116 patients (72 males and 44 females), thus giving raise a prevalence of 8.55 per 1000 live births. Maximum number of cases were diagnosed between 0-1 year age group (n=44, 37.93%) and less observed in other age group (Table 5). In our study CHD was more common in male (62.06%) as compare to female (37.93%). The various types of congenital heart diseases that are diagnosed and their pattern in our study, according to age group and sex are shown in table number 1. Isolated Ventricular septal defect followed by, Isolated Atrial septal defect, patent ductus arteriosus are the most common acyanotic and TOF was the commonest cyanotic congenital heart defects respectively. In Isolated VSD; perimembranous (69.69%) was the commonest, followed by Inlet (12.12%), muscular (9.09%) and supra-cristal type (6.06%) were observed (table 6). VSD associated with other cardiac heart defects like; TOF, AV canal defect, TGA, etc., are not included in the Isolated VSD, which were included with respective cardiac heart defects. The commonest Isolated ASD was Ostium secundum (71.42%), followed by Ostium primum (19.04%), Sinoseptal (9.52%), was observed (Table 7). ASD associated with other cardiac heart defects like; TAPVC, AV canal defect, Tricuspid Valve Atresia ,TGA ,etc., were not included in the Isolated ASD, which were included with respective cardiac heart defects. Tetralogy of Fallot's (6.03%), followed by transposition of the great arteries (3.44%), pulmonary atresia (2.58%) and Tricuspid atresia (0.86%), were the commonest cyanotic congenital heart defects (Table 1). Of the 116 patient diagnosed, only 13 (11.20%), underwent definitive treatment (surgical /Interventional cardiac) at the authors institute during study period.

Age Group	< 1 mo	nth		onth year	1 to yea		6 to yea	o 12 r	12 yea	to 18 ır	18 t yea	to 25 r	Total	(%)
CHD	М	F	М	F	М	F	Μ	F	Μ	F	М	F		
VSD	4	2	5	2	6	2	3	1	2	2	2	2	33	28.44
ASD	2	1	2	1	1	2	1	1	3	2	2	3	21	18.10
PDA	1	1	2	1	2	0	0	1	1	1	1	1	12	10.34
P.S.	0	1	1	0	2	0	0	2	1	0	0	0	07	6.03
TOF	0	1	2	0	0	2	0	0	1	1	0	0	07	6.03
TGA	1	1	0	0	0	0	1	0	0	0	1	0	04	3.44
A.V. canal defect	1	1	0	0	0	1	0	0	0	1	0	0	04	3.44
Bicuspid A.V.	0	0	0	1	0	0	0	1	1	0	0	0	03	2.58
Pulmonary Atresia	0	1	1	0	0	1	0	0	0	0	0	0	03	2.58
Complex	0	0	1	0	1	0	0	1	0	0	0	0	03	2.58
Dextrocardia	0	0	1	0	0	0	0	0	0	0	1	1	03	2.58
Ebstein's anomaly of T.V.	0	0	1	0	0	0	0	0	0	0	1	0	02	1.72
СоА	0	0	1	0	0	0	0	1	0	0	0	0	02	1.72
Enlargement of the coronary sinuses without left to right shunt: PLSVC	0	0	1	0	0	0	0	0	0	0	1	0	02	1.72
TAPVC	1	0	0	0	0	0	0	0	1	0	0	0	02	1.72
Single ventricle	0	0	0	0	0	0	0	0	0	0	1	0	01	0.86
QAV	0	0	0	0	0	0	0	0	0	0	1	0	01	0.86
T.V. atresia	0	0	0	0	1	0	0	0	0	0	0	0	01	0.86
Truncus arteriosus	0	0	0	0	1	0	0	0	0	0	0	0	01	0.86
Primary endocardial fibroelastosis	1	0	0	0	0	0	0	0	0	0	0	0	01	0.86
Aneurysm of the sinuses of valsalva	0	0	0	0	0	0	1	0	0	0	0	0	01	0.86
MVP	0	0	1	0	0	0	0	0	0	0	0	0	01	0.86
Cor-triatriatum- dexter	0	0	0	0	0	0	1	0	0	0	0	0	01	0.86
Total	11	09	19	05	14	08	07	08	10	07	11	07	116	

## Table 1: Pattern of congenital heat diseases (CHD) in the present study.(Total numbers 116 diagnosed out of 13,554 patients).

Table 2: Indian studies on prevalence of congenital heart diseases.

Author [reference, number]	Study population	Method	Number studied	CHD/1000 of study population
Shrestha et al. 1980 <sup>18</sup>	Community (5-16 y)	Clinical	34,198	3.2
Gupta et al. $1992^{24}$	Community (6-16 y)	Clinical	10,264	0.8
Vashishtha et al. 1993 <sup>25</sup>	Community (5-15y)	Clinical + Echo	8,449	5.2
Khalil et al. 1994 <sup>19</sup>	Hospital (Live birth)	Clinical + Echo	10,964	3.9
Thakur et al. 1995 <sup>26</sup>	Community (5-16y)	Clinical + Echo	40,950	2.25
Chadha et al. 2001 <sup>27</sup>	Community (<15y)	Clinical + Echo	11,883	4.2
Smitha R et al. 2006 <sup>2</sup>	Hospital (0 - 10 y)	Retrospective clinical & echo.	74,589	10.65
Kapoor et al. 2008 <sup>20</sup>	Hospital (0-15y)	Retrospective clinical & echo.	10,641	26.4
Misra et al .2009 <sup>05</sup>	School (5-15y)	Prospective clinical	118,212	1.3
Sawant et al 2012 <sup>29</sup>	Hospital (Live birth)	Prospective clinical & echo	2636	13.28
Bhat et al 2012 <sup>06</sup>	Hospital (0-18y)	Prospective clinical & echo	36541	8.54
Present study, 2013	Hospital (0-25y)	Prospective clinical &echo	13,554	8.55

Author [reference, number]	Study population	Method	Number studied	CHD/1000 of study population
Wallacee et al. 1953 <sup>11</sup>	Community	Clinical	162755	0.2
Gentry et al. 1955 <sup>30</sup>	Community	Clinical	124744	2.2
Fyler et al. 1980 <sup>31</sup>	Community (≤1 year)	Clinical & Echo	1083083	2.1
Ferencz et al. 1993 <sup>07</sup>	Community (Newborn)	Clinical + Echo	906646	4.8
Botto et al. 2001 <sup>32</sup>	Community (Newborn, stillborn – 1 y)	Clinical + Echo	937195	9.0
Tegnander et al. 2006 <sup>33</sup>	Community (GA 18 wk – 2 y)	Fetal Echo	30149	14.6
Wu et al. 2010 <sup>12</sup>	Community (Newborn registry data)	Retrospective, clinical & echo.	238143	13.1

#### Table 3: Selected large epidemiological studies on prevalence of CHD.

#### Table 4: Pattern of CHD in literatures.

Author	Study	Pattern (Individual CHD expressed as % of total cases)								
[Reference number]	population	VSD	ASD	PDA	PS	CoA	TOF	TGA	PA	TA
Shrestha et al. <sup>18</sup>	5-16yea	30	23	11			4			
Vashishtha et al. <sup>25</sup>	5-15year	41	11	4			14			
Thakur et al. <sup>26</sup>	5-16year	32	38							
Sharma et al. <sup>34</sup>	<12year	53	13	13		8	3.2	2.2	6	
Kapoor et al. <sup>20</sup>	<15year	21	19	14	3		5	1		
Misra et al. <sup>05</sup>	5-15year	40	18	2	8					
Smitha R et al. <sup>28</sup>	0 - ≥10year	40	19	9.5			13	0.6		
Amro K <sup>35</sup>	<14year	43.4	13.6	8.3	6.2	3.4	9.5	5.5		3.6
Abbag F et al. <sup>23</sup>	Children	32.5	10.4	15.8	10.1	3.3	4.5	1.5		1.5
Present Study	0 - 25 year	28.44	18.10	10.34	6.89	1.72	6.03	4.31	2.58	0.86

## Table 5: Percentage of CHD according to Age<br/>group in the present study.

0 to 1 year	43	37.06%
1 to 6 year	22	18.96%
6 to 12 year	16	13.79%
12 - 18 year	17	14.65%
18 - 25 year	18	15.5%

# Table 6: Various types of isolated ventricularseptal defects are observed and diagnosed in the<br/>present study.

Perimembranous	23	69.69%
Inlet type	04	12.12%
Muscular	04	12.12%
Supra cristal	02	6.06%

(Total numbers of Isolated VSD are 33 out of 116)

## Table 7: Types of isolated atrial septal defectsin the present study.

Ostium Primum	04	19.04%
Ostium Secundum	15	71.42%
Sino-Septal	02	9.52%

(Total numbers of Isolated ASD are 21 out of 116)

#### DISCUSSION

Congenital heart disease remains the commonest cause of death from structural birth defect. It is generally accepted that 8 per, 1000 live birth result in CHD.<sup>10</sup> In our study prevalence was 8.55 per 1,000 live births and it is slightly higher side. How ever, reported prevalence of CHD varies widely among large epidemiological studies. Hoffman reported low frequency of CHD at about 4-5 per 1,000 live births.<sup>13</sup> Recent studies suggest an increase in the prevalence of CHD with reported frequency of 12 -14 per 1,000 live births or even higher.<sup>14</sup> Studies using echocardiography may include cardiac lesions like small muscular VSD and ASD with little clinical importance and high rate of spontaneous closure and consequently, over estimate the prevalence. This is evident by a steep rise in prevalence of CHD reported by studies in echocardiography era.<sup>15</sup> with some of the highest prevalence rate reported by studies using echocardiography for screening.<sup>14</sup> Some of the large epidemiological studies are summarized in Table number 3. According to a recent systemic review, highest prevalence of CHD reported from Asia (9.3 per 1,000 live births) while the least is reported from Africa (1.9 per 1,000 live births). Survival of premature infants has improved over the last century, attributing to an increase in total CHD and especially PDA.<sup>13</sup> Furthermore: because late marriage, increasing numbers of women are delaying the childbearing, maternal age has increased causing a higher prevalence of congenital abnormalities.<sup>16</sup> In addition with advances in healthcare worldwide, the patient population with adult CHD is steadily increasing, with their offspring at increased risk of having CHD.<sup>17</sup> In contrast to large epidemiological studies from developed countries, studies from India.<sup>5-18</sup> have reported a wide variation in the prevalence of CHD from 2.25 to 26.3 per 1,000 live births (Table 2).

Prevalence also depends on the study population, the population based studies being the best. The few recent studies available from India have taken into consideration only particular groups i.e., new born, school children. The study by Khalil et al.<sup>19</sup> includes 10964 hospital live births and observed the incidence of 3.9/1000 live births. Misra et al.<sup>5</sup> reported a prevalence of 1.3 /1000 school children 5 to 15 year of age. The former may miss out on a large number of small VSD, TOF or ductus dependent lesions, which present a little later than at birth. It also fails to focus the prevalence of CHD. The latter study automatically excludes all children with severe lesion who would be school dropouts. Kapoor et al.<sup>20</sup> reported a

prevalence of 26.3/1000 patients aged 0 to 15 year at a tertiary corporate hospital. Hence they do not present a true picture. The authors observation of prevalence of 8.55/1000 live births, should be more representative as compare to earlier studies. Bhat N.K. et al.<sup>6</sup> have reported a prevalence of 8.54/children in Uttrakhand. Smitha et al.<sup>21</sup> have reported a prevalence of 10.65/children at three hospital of Mysore. Since a large number of births in India still take place at home. Especially in villages, remote areas, semi-urban, and urban areas and most of deliveries are conducted by an unqualified doctor, so the hospital statistics are unlikely to be truly reflective.<sup>3</sup> The actual prevalence in community may be somewhat lower as there will be many children are who did not need to attend the authors' institution during the study period. This highlights the pitfall in finding the prevalence of CHD by certain group based surveys and the need and importance of the community based surveys. Such studies are nonexistent in India.<sup>3</sup> Some of the prevalence studies available from India and other countries are summarized in table number 2, 3 and 4.

The age at detection of CHD varies due to the normal hemodynamic alterations occurring after birth like fall in pulmonary vascular resistance, physiological and anatomical closure of PDA. Many congenital heart diseases, especially minor defects tend to be asymptomatic and hence missed unless specifically sought. Most authors agree that half of all cases of CHD are detected by one month of age, three fourth by three month, remaining by the age of 3-4 year but also diagnosed after the age 18 year.<sup>19,21,22</sup> In the present study only 37.06% CHD cases were detected by the age of one year and remaining shown in table number 5. In the present study we found that up to 15.51% were detected after the age of 18 year, so this delay in the diagnosis can be explained due to lack of awareness, health facilities and pediatric care program's in India.<sup>3</sup>

Estimates of the frequency of specific lesions vary, depending on whether the data are drawn from infants or older children and whether the diagnosis is based on clinical, echocardiography, catheterization, surgical or post-mortem studies.<sup>9</sup> The pattern from various countries is remarkably similar. Table number 4 summaries the pattern of CHD observed in some of the comparative age group studies available in literature.

VSD is the most common malformation, occurring in 25-30% of all patients with congenital heart disease.<sup>22</sup> In our study VSD was the commonest congenital heart defect which was accounting for 28.44% of CHD cases and correlates well with the reported range of 21-53% in the literature (Table 1). ASD was the second most common CHD in our study, comprising 18.10%.This correlates well with the frequency of 10 - 23% reported in various Indian studies, but it is higher than 6-8% reported from western countries.<sup>22</sup> TOF was the most common cyanotic CHD comprising 6.03%, correlating well with other studies.<sup>18,20,23</sup> An impotent observation from present study is the increasing number of children with unrecognized or uncorrected CHD, growing into adolescents and adults. In the preset study 15.51% patients were diagnosed after the age of 18 and by that time most of patients were developed pulmonary hypertension. Only 11 of 142 children's

had undergone definitive treatment in the study by Misra et al.<sup>5</sup> Of the 116 patient diagnosed with CHD, only 13 (11.20%), underwent definitive treatment (surgical /Interventional cardiac) at authors institute during study period and they came for follow up. However the number of children who after diagnosis underwent definitive treatment at other centers is not available. Prevention of all cases of CHD is impossible as the cause of most congenital heat diseases is unknown.<sup>22</sup> The best approach, therefore, is early identification and management of the problem. This may be achieved by increasing awareness and early evaluation of suspected cases. Many affected children do achieve cure or long-term palliation. Therefore knowledge of prevalence of CHD is important and establishing advanced diagnostic and treatment facilities within region.

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

#### CONCLUSION

The prevalence of CHD at a semi-urban tertiary teaching hospital in, Bommakal - Karimnagar, Andhra Pradesh is 8.55/1000 live births. In the present study diagnosis was delayed in 15.5%. Isolated VSD (28.44%) and TOF (6.03%) are the most common acyanotic and cyanotic congenital heart defects respectively. In absence of known cause, early diagnosis and treatment appears to be the best approach to minimize the morbidity and mortality attributed to CHD.

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#### Abbreviations

CHD- Congenital heart diseases, VSD - Ventricular septal defect, ASD - Atrial septal defect, PDA - Patent ductus arteriosus, P. S.- Congenital pulmonary stenosis, TOF- Tetralogy of Fallot, TGA - Transposition of great arteries, MVP - Mitral valve prolapse, Bicuspid A.V.-Bicuspid aortic valve, QAV - Quadricuspid aortic valve, A.V. canal defect - Atrioventricular canal defect (Endocardial cushion defect), T.V. atresia - Tricuspid valve atresia, Ebstein's anomaly of T.V. - Ebstein's anomaly of tricuspid valve, TAPVC - Total anomalous pulmonary venous connection, PLSVC - Persistent left superior vena cava and CoA - Coarctation of aorta. Conflict of interest: None declared

*Ethical approval: The study was approved by the institutional ethics committee* 

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