

**A Hospital-Based Assessment of the Clinical Profile of Congenital Heart Disease in Children and its Correlation with Echocardiography****Sushil Kumar Pathak<sup>1</sup>, Jaymala Mishra<sup>2</sup>, Saroj Kumar<sup>3</sup>, Binod Kumar Singh<sup>4</sup>**<sup>1</sup>Assistant professor, Department of Pediatrics, Nalanda Medical College and Hospital, Patna, Bihar, India<sup>2</sup>Assistant Professor, Department of Cardiology, Katihar Medical College and Hospital, Katihar, Bihar, India<sup>3</sup>Assistant Professor, Department of Pediatrics, Nalanda Medical College and Hospital, Patna, Bihar, India<sup>4</sup>Professor and HOD, Department of Pediatrics, Nalanda Medical College and Hospital, Patna, Bihar, India

Received: 10-02-2023 Revised: 20-03-2023 / Accepted: 25-04-2023

Corresponding author: Dr. Jaymala Mishra

Conflict of interest: Nil

**Abstract****Aim:** The aim of the present study was to assess the clinical profile of congenital heart disease in children and establishing correlation of clinical diagnosis with echocardiography diagnosis.**Material & methods:** This was a prospective cross-sectional study conducted in the Department of Pediatrics, over a period of 3 years. All children between 1 month to 14 years admitted for suspected CHD were included in the study. Clinical diagnosis was made based on detailed history and physical examination supported by chest X-ray and ECG. Final diagnosis was confirmed by echocardiography and clinico-ECHO correlation was deduced.**Results:** During this study period of 3 years, total number of admissions was 11250 and 200 CHD cases were admitted as per the inclusion criteria. The hospital incidence of CHD found to be 1.77%. The 52% were male and 48% were female. Patients from rural area constituted 80% and urban area 20%. Maximum children belonged to age group of 1 month to 1 year with 72%. Among all CHD, most common symptom was breathlessness (80%). Among ACHD, most common presentation was breathlessness (73.34%), cough (62.5%), forehead sweating (45.83%), feeding difficulty (37.50%) and among CCHD were breathlessness (90%). Overall murmur (85%) was the commonest clinical sign among CHD. Leading clinical sign among ACHD were murmur (91.66%), chest retraction (70.83%), growth retardation (50%) and among CCHD were desaturation (100%), murmur (75%), growth retardation (55%). CCF observed in 35% in ACHD and 32.5% in CCHD. Most common complication in CHD was growth retardation in both categories of ACHD (50%) and CCHD (62.5%) respectively. Clinico-echo correlation was correlated in 34%, partially correlated in 36%, not correlated in 30% cases. Clinico-echo correlation with aid of CXR and ECG correlated in 40%, partially correlated 32%, not correlated 28% cases.**Conclusion:** So, echocardiography remains the gold standard for diagnosis of CHD and guiding the treatment. Clinical diagnosis alone may not detect associated CHD.**Keywords:** CHD, Clinical diagnosis of CHD, Echocardiography.

This is an Open Access article that uses a funding model which does not charge readers or their institutions for access and distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/4.0>) and the Budapest Open Access Initiative (<http://www.budapestopenaccessinitiative.org/read>), which permit unrestricted use, distribution, and reproduction in any medium, provided original work is properly credited.

**Introduction**

Congenital heart disease (CHD) is defined as an abnormality in 'cardio circulatory' structure or function that is present since birth. [1] CHDs remains the leading cause of death in children with malformation. [2] Incidence of CHDs being 8 per 1000 live births and is the most common severe congenital abnormality. It is the most common congenital birth defect affecting 28% of all major congenital anomalies. [3] The incidence of CHD in the normal population is approximately 0.5-0.8% of

live-born children, with a higher percentage in those aborted spontaneously or stillborn.

CHDs are categorized as acyanotic congenital heart disease (ACHD) and cyanotic congenital heart disease (CCHD) according to the pathophysiology and the affected heart structure. [4] The acyanotic lesion consists of septal cardiac defects such as atrial septal defect (ASD), ventricular septal defect (VSD), and atrioventricular canal defects (AVSD). It also includes left ventricular outflow obstructive

lesions such as aortic stenosis (AS) and coarctation of the aorta (COA). CHD includes tetralogy of Fallot (TOF), transposition of great arteries (TGA), total anomalous pulmonary venous returns (TAPVC), hypoplastic left heart syndrome (HLHS), truncus arteriosus, and tricuspid atresia.<sup>4</sup> Although CHD affects both males and females equally, some lesions such as COA, AS, transposition of great vessels, and TOF show a male preponderance whereas females report more ASD. [5] Clinical spectrum of CHD is versatile and varies with the type of CHD ranging from poor sucking, cyanosis, respiratory distress, failure to thrive to frank heart failure. Physical diagnosis consists of synthesis of information from FIVE sources: physical appearance, arterial pulse, JVP, precordial examination and auscultation. Asymptomatic presentation is common and discovered accidentally during routine check-up visits and requiring subsequent ECHO evaluation.<sup>6</sup> With newer diagnostic modalities including echocardiography, it is now considered as near confirmatory in diagnosing most of the congenital heart lesions. X-ray chest and ECG are complimentary to echocardiography. It needs to be ascertained as to what would be the diagnostic reliability of clinical diagnosis versus Echocardiography diagnosis.

Hence the aim and objective of the study was to know the clinical profile including modes of presentations of CHD in children and also to find the correlation between physical diagnosis (clinical examination/chest x-ray/ electrocardiogram (ECG) with echocardiography diagnosis.

### Material & Methods

A prospective observational, cross-sectional hospital-based study was conducted in the Department of Paediatrics, Nalanda Medical College and Hospital, Patna, Bihar, India over period of 3 years. Criteria used to suspect heart disease was followed as per NADAS criteria formulated in 1959 by Alexander Nadas.<sup>7</sup> The study was conducted after institutional ethical committee approval. All children from 1 months-14 years age groups full filling the inclusion criteria were enrolled in the study. During this study period of 3 years, total number of admissions was 11250 and 200 CHD cases were admitted as per the inclusion criteria. The hospital incidence of CHD found to be 1.77%.

### Inclusion Criteria

- All children between 1 month to 14 years of age suspected of CHD admitted to paediatric ward NMCH, Patna fulfilling NADA's criteria

(1959) for heart disease and also those having following things.

- Patient showing desaturation, syndromic baby which is most likely to be associated with CHD and presence of only murmur not fitting to NADA's criteria were included in the study.

### Exclusion Criteria

- Any other causes of heart disease like-acquired heart diseases pericarditis, myocarditis, rheumatic heart disease, previously diagnosed patients of CHD, post-operative case of CHD, abnormal BP and pulse due to other vasculitis, unstable patients who died before confirmation of diagnosis, echocardiography showing normal study and family not willing for the study were excluded from the study.

### Methodology

A detailed history and thorough clinical examination were performed on the study subjects as per pre-structured proforma revealing a provisional clinical impression. They were further investigated with ECG and chest X-ray and a final clinical diagnosis was assigned. Investigations like CBC, CRP, ESR and other accessory investigations as per requirement of individual case like blood culture and sensitivity, CT Scan brain etc were also done. All these patients were subjected to Echocardiography in the Department of Cardiology, Nalanda Medical College and Hospital. ECHO was done by paediatric cardiologist using Phillips machine by S4-2 phased array probe of frequency 2-4 MHz. Different modes used in doing transthoracic echo were M Mode, 2D Echo, colour doppler, pulse wave doppler, continuous wave doppler. The views were apical four chamber, parasternal long axis, parasternal short axis, subxiphoid/ subcostal, suprasternal view. Echo was done in a segmental approach after which final diagnosis was done. Finally, the results were tabulated, analysed and the correlation between clinical diagnosis and echocardiography diagnosis were obtained.

### Statistical Analysis

Data collected was entered into the excel sheet for completeness. All the statistical operations were performed through SPSS version 23. Descriptive statistics like percentage was used for the characteristics. Tests used to calculate various associations were unpaired t test (for normal data), chi-square test (for dichotomous variable).  $P < 0.05$  was considered as statistically significant.

### Results

**Table 1: Demographic profile of CHDs**

Variable	CHD, (n=200)	Percentage (%)
<b>Incidence</b>	200/11250	1.77
<b>Sex</b>		
Male	104	52
Female	96	48
<b>Area</b>		
Urban	40	20
Rural	160	80
<b>Age</b>		
1 month-1 year	144	72
1-5 years	32	16
>5 years	24	12

During this study period of 3 years, total number of admissions was 11250 and 200 CHD cases were admitted as per the inclusion criteria. The hospital incidence of CHD found to be 1.77%. The 52% were male and 48% were female. Patients from rural area constituted 80% and urban area 20%. Maximum children belonged to age group of 1month-1year with 72%.

**Table 2: Clinical symptoms and signs distribution in CHD**

Variables	ACHD	ACHD (%)	CCHD	CCHD (%)	Total (%)
Breathlessness	88	73.34	72	90	80
Cough	75	62.5	45	56.25	60
Chest pain	12	10	0	0	3
Fever	60	50	60	50	60
Irritability	60	50	30	37.5	45
Forehead sweating	55	45.83	25	31.25	45
suck rest suck cycle	35	29.16	15	30	25
Feeding difficulty	45	37.50	25	31.25	35
Palpitations	7	5.83	5	6.25	3
cyanotic spell	0	0	20	25	10
Failure to thrive	60	50	44	55	52
Swelling of limbs	5	4.16	5	6.25	5
Seizure	0	0	1	1.25	0.5
Headache	0	0	2	2.5	1
Desaturation	12	10	60	75	36
BP UL-LL difference	2	1.66	0	0	1
Growth retardation	60	50	44	55	52
Cyanosis	0	0	56	70	28
Clubbing	1	0.83	23	28.75	12
CCF	42	35	26	32.5	34
Extra cardiac anomaly	18	15	10	12.5	14
Chest retraction	85	70.83	51	63.75	68
Precordial bulge	48	40	22	27.5	35
Hyper dynamic precordium	24	20	16	20	20
Thrill	20	16.66	10	12.5	15
Parasternal leave	7	5.83	5	6.25	6
Abnormal S2	9	7.5	11	13.75	10
Murmur	110	91.66	60	75	85
Neurological deficit	0	0	2	2.5	1

Among all CHD, most common symptom was breathlessness (80%). Among ACHD, most common presentation was breathlessness (73.34%), cough (62.5%), forehead sweating (45.83%), feeding difficulty (37.50%) and among CCHD were breathlessness (90%). Overall murmur (85%) was the commonest clinical sign among CHD.

Leading clinical sign among ACHD were murmur (91.66%), chest retraction (70.83%), growth retardation (50%) and among CCHD were desaturation (100%), murmur (75%), growth retardation (55%). CCF observed in 35% in ACHD and 32.5% in CCHD.

**Table 3: Complications associated with CHD**

Complications	ACHD	ACHD (%)	CCHD	CCHD (%)
Pneumonia	60	50	24	30
CCF	42	35	22	27.5
Brain abscess	0	0	3	3.75
Thromboembolism	0	0	1	1.25
Cyanotic spell	0	0	22	27.5
Growth retardation	60	50	50	62.5
Infective endocarditis	1	0.83	0	0

Most common complication in CHD was growth retardation in both categories of ACHD (50%) and CCHD (62.5%) respectively.

**Table 4: ECG findings in CHD**

ECG changes	ACHD	%	CCHD (N)	%	Total (N)	%
Normal	28	23.34	8	10	36	18
LAD/LVH	48	40	4	5	56	28
RAD/RVH	12	10	30	37.5	42	23
CVH/BVH	4	3.33	0	0	4	2
RSR'+RVH/RAD	3	2.5	3	3.75	6	3
RSR'+LVH/LAD	4	3.33	0	0	4	2
RAD/BVH	2	1.66	2	2.5	4	2
LAD/BVH	4	3.33	4	5	8	4
Sinus tachycardia	2	1.66	6	7.5	8	4
Himalayan p wave	0	0	1	1.25	1	0.50
Deep q in lead I and Avl	1	0.83	0	0	1	0.50
Not done	15	12.5	14	17.5	29	16
Total	120	100	80	100	200	100

After a provisional clinical impression, Chest X-ray and ECG were done.

**Table 5: Echocardiography diagnosis (ACHD)**

ECHO (ACHD)	N	Percentage (%)
VSD	24	20
VSD+PAH	12	10
VSD+ASD+PAH	6	5
VSD+ASD+dextrocardia	1	0.83
VSD+ASD	4	3.33
VSD+PDA	1	0.83
PDA	12	10
PDA+PAH	1	0.83
ASD	10	5.3
ASD+PAH	2	1
PS	4	3.33
AS	2	1.66
COA	1	0.83
PDA+BAV+AS	1	0.83
VSD+PDA+PAH	1	0.83
AVSD	3	2.5
AVSD+PS	1	0.83
VSD+PS	4	2.50
PDA+PS	1	0.83
ASD+PS	4	2.50
VSD+COA+PAH	2	1.66
Peripheral PS	2	1.66
ASD+PDA	3	2.50
VSD+ASD+PDA+PAH	1	0.83
Alcapa	1	0.83

**Table 6: Echocardiography diagnosis (CCHD)**

ECHO (CCHD)	N	Percentage (%)
<b>TOF</b>	8	10
Falot's pentalogy	1	1.25
TOF+PDA	1	1.25
AVSD+TOF	1	1.25
D-TGA+VSD	3	3.75
D-TGA+VSD+PAH	2	2.5
D-TGA+VSD+PS	2	2.5
TAPVC+PAH (supracardiac, cardiac, mixed)	2	2.5
D-TGA+VSD+ASD+PDA	1	1.25
D-TGA+ASD+PDA	1	1.25
D-TGA+ASD+VSD	1	1.25
COMMON ATRIUM	1	1.25
DORV+VSD+PAH	1	1.25
DORV+VSD+pulmonary atresia	1	1.25
DORV+AVSD+pulmonary atresia	1	1.25
AVSD+pulmonary atresia	1	1.25
Ebstein's anomaly	1	1.25
SV+ASD+PDA	1	1.25
SV+ASD+PS	1	1.25
Truncus arteriosus	1	1.25

**Table 7: Clinic-echo correlation**

Variables	Clínico (only)-ECHO correlation, n (%)	Clinical + CXR+ ECG with ECHO correlation, n (%)
Correlated	68 (34)	80 (40)
Partially correlated	72 (36)	64 (32)
Not correlated	60 (30)	56 (28)
Total	200 (100)	200 (100)

Clinico-echo correlation was correlated in 34%, partially correlated in 36%, not correlated in 30% cases. Clinico-echo correlation with aid of CXR and ECG correlated in 40%, partially correlated 32%, not correlated 28% cases.

### Discussion

Congenital heart disease (CHD) is the most frequently occurring congenital disorder responsible for 28% (1/3rd) of all congenital birth defects. [8] Its estimated incidence is up to 6-8 in 1000 live births and 45% of deaths caused by congenital defects include cardiac malformations. [9,10] Clinical spectrum of CHD is versatile and varies with the type of CHD ranging from poor sucking, cyanosis, respiratory distress, failure to thrive to frank heart failure. Asymptomatic presentation is common and discovered accidentally during routine check-up visits and requiring subsequent ECHO evaluation. [11]

During this study period of 3 years, total number of admissions was 11250 and 200 CHD cases were admitted as per the inclusion criteria. The hospital incidence of CHD found to be 1.77%. The 52% were male and 48% were female. Patients from rural area constituted 80% and urban area 20%. There was sex preponderance of VSD, COA, AS, PS, TOF, TGA, Falot's pentalogy observed to be

more common in male whereas ASD, PDA, TAPVC, Truncus arteriosus, Ebstein's anomaly more common in female. Extra cardiac anomalies (ECA) were found in 13.3% of cases which is comparable to study by Pandey et al who reported 8.5%. [12] Maximum children belonged to age group of 1month-1year with 72%. Among all CHD, most common symptom was breathlessness (80%). Among ACHD, most common presentation was breathlessness (73.34%), cough (62.5%), forehead sweating (45.83%), feeding difficulty (37.50%) and among CCHD were breathlessness (90%). Overall murmur (85%) was the commonest clinical sign among CHD. Leading clinical sign among ACHD were murmur (91.66%), chest retraction (70.83%), growth retardation (50%) and among CCHD were desaturation (100%), murmur (75%), growth retardation (55%). Almost similar observations have been reported by Kuntal, Ashish and Sharmin as well with few discrepancies. [13-15]

CCF observed in 35% in ACHD and 32.5% in CCHD. Most common complication in CHD was growth retardation in both categories of ACHD (50%) and CCHD (62.5%) respectively. Clinico-echo correlation was correlated in 34%, partially correlated in 36%, not correlated in 30% cases. Clinico-echo correlation with aid of CXR and ECG correlated in 40%, partially correlated 32%, not

correlated 28% cases. Most cases of TGA and DORV presented in CCF were in early infancy and were associated with multiple lesions as evidenced by standard literature. [16,17] Anisworth et al found 48% of CHD in infancy without heart murmur and Shima et al found 40% CHD without murmur and cyanosis. [18-20] The difference between clinical, final clinical and echocardiography findings is due to similar presentation of different types of CHD like TOF, Fallot's physiology have clinically similar findings and complex CHD like DORV, TGA, Single Ventricle have similar presentation. So, the actual structural anomaly was picked up after echocardiography.

Our study has comparable individual correlation of CHD with study by Pestana among all common lesions except VSD and COA. [21] This can be explained by the fact that COA was missed due to missing out on detecting pulse and BP difference in 4 limbs. Low clinico-echo correlation of VSD can be explained by over diagnosis of VSD clinically due to missing of other varieties of CHD in presence of VSD murmur. Combination of fully and partially correlated CHD comes out to be 72% which is comparable to few other studies like Tandon et al (80-85%), Klewer (81%). [22,23] But most of these studies have been done by experienced paediatric cardiologist. The reported defect in accuracy of clinical diagnosis probably reflects the training skills and experiences of the paediatricians which needs urgent upgradation for better patient management. Though clinical diagnosis is reasonably accurate but important misclassification and inaccuracies can occur.

### Conclusion

Total 1/3rd of our echo diagnosis was of multiple defects and complex heart diseases. Echocardiography remains the gold standard and is mandatory in all suspected cases of CHD to derive at specific diagnosis, to know the severity of lesion and timing for surgical or catheter intervention and follow up. Presence or absence of murmur does not assure either the presence or absence of significant CHD. Absence of cyanosis does not rule out CCHD so presence of desaturation in pulse oximetry should be addressed. Pulse oximetry is also important beyond neonatal age. Correlation was found to be least in complex heart disease with multiple defects. Inaccuracy of clinical diagnosis is because of lack of systematic step wise examination of cardiovascular system. Timely intervention, referral and close monitoring is crucial for optimal outcome of CHD patients. Training workshops and upgradation of clinical skill is crucial for better pediatric cardiac care.

### References

1. Bernstein D. Congenital heart disease. In: Behrman, Kleigman, Jenson, eds. Nelson Textbook of Pediatrics. 17th ed. Philadelphia: Saunders; 2004:1499-1502.
2. Webb GD, Smallhorn JF, Therrein J. Congenital Heart Disease. In: Zipes, Libby, Bonow, Braunwald, editors. Braunwald's Heart Disease. 7 th ed. Philadelphia: Saunders; 2005: 1489-1547.
3. Dolk H, Loane M, Garne E, a European Surveillance of Congenital Anomalies (EUROCAT) Working Group. Congenital heart defects in Europe: prevalence and perinatal mortality, 2000 to 2005. *Circulation*. 2011 Mar 1;123(8):841-9.
4. Tulloh RM, Medrano-Lopez C, Checchia PA, Stapper C, Sumitomo N, Gorenflo M, Bae EJ, Juanico A, Gil-Jaurena JM, Wu MH, Farha T. CHD and respiratory syncytial virus: global expert exchange recommendations. *Cardiology in the Young*. 2017 Oct;27(8):1504-21.
5. Di Filippo S: [Infective endocarditis prophylaxis in congenital heart disease]. *Presse Med*. 2017, 46:606-11.
6. Wren C, Richmond S, Donaldson L. Presentation of congenital heart disease in infancy: implications for routine examination. *Archives of Disease in Childhood-Fetal and Neonatal Edition*. 1999 Jan 1;80(1):F49-53.
7. Alexandar S. Nadas. Approach to diagnosis of congenital heart disease without recourse to special tests. *Circulation*. 1959;602-5.
8. Dolk H. Congenital Heart Defects in Europe 2000-2005. EUROCAT; 2009.
9. Hoffman JI. The global burden of congenital heart disease. *Cardiovascular journal of Africa*. 2013 May 1;24(4):141-5.
10. Hoffman JI. Incidence of congenital heart disease: I. Postnatal incidence. *Pediatric cardiology*. 1995 May; 16:103-13.
11. Wren C, Richmond S, Donaldson L. Presentation of congenital heart disease in infancy: implications for routine examination. *Archives of Disease in Childhood-Fetal and Neonatal Edition*. 1999 Jan 1;80(1):F49-53.
12. Pandey N, Pooniya V. Clinico-epidemiologic profile of children with congenital heart disease in a tertiary care hospital in North India. *J Adv Med Dent Scie Res*. 2019;7(8):92-5.
13. Roy K, Shahed H, Roy K, Sarah QS, Chowdhury NS. Clinical Presentation and Complications of Different Congenital Heart Disease in Children. *Am J Pediatr*. 2020;6 (4): 481-7.
14. Varma A, Sharma V, Damke S, Meshram RJ, Kher A, Vagha J. Clinical presentation of cyanotic congenital heart diseases in the

- pediatric population. *J Datta Meghe Inst Med Sci Univ.* 2020; 15:7-11.
15. Sharmin LS, Haque MA, Bari MI, Ali MA. Pattern and clinical profile of congenital heart disease in a teaching hospital. *TAJ J Teach Assoc.* 2008; 21:58- 62.
  16. Bernstein D. Congenital heart disease. *Nelson textbook of paediatrics.* Kliegman RM, Geme ST, Blum N J, Tasker R, Shah S, Wilson K M. (editors): *Nelson Textbook of Pediatrics.* 21st ed. Saunders Elsevier Philadelphia. 2020; 187 8-81.
  17. PARK's Pediatric cardiology for practitioners, Elsevier Saunders, 6th edition. 2014.
  18. Anisworth S, Wyllie JP, Wren C. Prevalence and clinical significance of cardiac murmurs in neonates. *Arch Dis Child Fetal Neonatal Ed.* 1999;80:F43-5.
  19. Shima Y, Takechi N, Ogawa S. Clinical characteristic of congenital heart disease diagnosed during neonatal period. *J Nippon Med Sch.* 2001; 68:510-15.
  20. Hoque MM, Begum JA, Jahan R, Chowdhury MA, Hussain M. Importance of Cardiac Murmur in Diagnosing Congenital Heart Disease in Neonatal Period. *Bangladesh J Child Health.* 210;32(1):17-20.
  21. Pestana C, Weidman WH, Swan HJC. Accuracy of preoperative diagnosis in CHD. *Am Heart J.* 1966;72(4):446-50.
  22. Tandon R. Bedside evaluation of congenital heart disease. *Indian Pediatrics* 1972; 9(6): 30 1-305.
  23. Klewer SE, Samson RA, Donnerstein RL, Lax D, Zamora R, Goldberg SJ. Comparison of accuracy of diagnosis of congenital heart disease by history and physical examination versus echocardiography. *Am J Cardiol.* 2002; 89:1329-31.