

Original Research Article

Clinical profile of congenital heart disease in children with special reference to echo correlation

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ABSTRACT

Background: Congenital heart disease (CHD) is the most frequently occurring congenital disorder responsible for 1/3rd of all congenital birth defects. Diagnostic accuracy of CHD is crucial for timely intervention and optimal patient outcome. So, the study has been done to look for the clinical profile and establishing correlation of clinical diagnosis with echocardiography diagnosis.

Methods: This is a prospective cross-sectional study conducted in the department of pediatrics MKCG MCH, Berhampur over a period of 2 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: The 188 patients with CHD were included in this study. The most common age of presentation was 1 month to 1 year with 71.2%, male to female ratio was 1.06:1. Acyanotic CHD (ACHD) was found to be 70.29%, while 29.7% had congenital cyanotic heart disease (CCHD). Clinically specific diagnosis was attributed to 67.1% of cases and no specific diagnosis could be made in 32.9%. Isolated CHD was found to be 66.4% and Complex CHD with multiple defects was found to be 33.6%. Clinic echo correlation was accurate in 39.4% cases, partially correlated in 32% and no correlation in 28.6% cases.

Conclusions: 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

INTRODUCTION

Congenital heart disease (CHD) is the most frequently occurring congenital disorder responsible for 28% (1/3rd) of all congenital birth defects.¹ Its estimated incidence is up to 6-8 in 1000 live births and 45% of deaths caused by congenital defects include cardiac malformations.^{2,3} 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.⁴

However, role of chest x-ray and ECG in complementing the diagnosis of CHD should not be underestimated.⁵ There have been many previous studies in Odisha showing prevalence, clinical features, diagnosis of CHD but very few studies show clinico-echo correlation in CHD patients. Our hospital is prime tertiary care teaching hospital not only for southern Odisha but also for neighbouring state like Andhra Pradesh. So, we wanted to know what is the diagnostic reliability of clinical

diagnosis versus Echocardiography diagnosis in the our area.

Aim and objective of our 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.

METHODS

This study was conducted in the department of paediatrics, MKCG medical college and hospital, Berhampur over a period of 2 years (2019-2021). This was a prospective observational, cross-sectional hospital-based study. Criteria used to suspect heart disease was followed as per NADAS criteria formulated in 1959 by Alexander Nadas.⁶

Inclusion criteria

All children between 1 month to 14 years of age suspected of CHD admitted to paediatric ward MKCG, Berhampur 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.

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. 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, MKCG. 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

Total number of cases taken as per inclusion criteria=191. After echocardiography, 3 cases were excluded as echocardiography was normal. Finally, cases included for analysis was 188. 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

During this study period of 2 years, total number of admissions were 10,080 and 188 CHD cases were admitted as per the inclusion criteria. The hospital incidence of CHD found to be 1.86%. The 51.5% were male and 48.5% were female. Patients from rural area constituted 78.7% and urban area 21.3%. Maximum children belonged to age group of 1month-1year with 71.2% (Table 1).

Table 1: Demographic profile of CHDs.

Variable	CHD, (n=188)	Percentage (%)
Incidence	188/10,080	1.86
Sex		
Male	97	51.5
Female	91	48.4
Area		
Urban	40	21.3
Rural	148	78.7
Age		
1 month-1 year	134	71.2
1-5 years	30	15.9
>5 years	24	12.7

Among all CHD, most common symptom was breathlessness (78.3%). Among ACHD, most common presentation was breathlessness (72.7%), cough (62.1%), fever (50.8%), FTT (50.7%), forehead sweating (45.4%), feeding difficulty (37.12%) and among CCHD were breathlessness (83.9%), fever (67.3%), FTT (60.7%). Overall murmur (86.2%) was the commonest clinical sign among CHD. Leading clinical sign among ACHD were murmur (88.6%), chest retraction (70.5%), growth retardation (50.7%) and among CCHD were desaturation

(100%), murmur (80.3%), cyanosis (91%), growth retardation (60.7%). CCF observed in 35.6% in ACHD and 26.7% in CCHD (Table 2).

Risk factors associated with CHD constituted consanguinity 20.3%, syndromic association 9.04%, family history of CHD in 5.8%, mother age>35 years in 4.7%, maternal diabetes in 2.12%, maternal hypertension in 1.6%, intrauterine infections 2.1% (2 CMV+2 toxoplasma). Syndromic association observed in 9.04% cases of CHD.

Most common complication in CHD is growth retardation in both categories of ACHD (50.7%) and CCHD (60.7%) respectively. Other common complications (Table 3).

Clinically specific diagnosis could be given for 67.1% cases, in rest 33% we were unable to give specific diagnosis After a provisional clinical impression, Chest X-ray and ECG was done. The 84% of CHD cases had

abnormal CXR findings and overall common findings were cardiomegaly+ pulmonary plethora 21.8%, cardiomegaly 14.9%, cardiomegaly with opacity 12.2%. Among ACHD cardiomegaly+ pulmonary plethora 21.2% being most common and in CCHD, common abnormal findings were booting shaped heart +oligemia 25%, cardiomegaly+ plethora 23.2%, cardiomegaly+ oligemia 10.7%, egg on side+ cardiomegaly+ plethora 9%. ECG abnormality found in 79.8% cases. 15% cases ECG couldn't be done due to COVID pandemic and other reasons (Table 4).

Details of echocardiographic findings are shown in (Table 5 and 6).

Clinico-echo correlation was correlated in 31.9%, partially correlated in 34.04%, not correlated in 34.04% cases. Clinico-echo correlation with aid of CXR and ECG correlated in 32%, partially correlated 34%, not correlated 34% cases (Table 7). Clinico-Echo correlation of major CHD is depicted in (Table 8).

Table 2: Clinical symptoms and signs distribution in CHD.

Variables	ACHD	ACHD (%)	CCHD	CCHD (%)	Total (%)
Breathlessness	96	72.7	47	83.9	78.3
Cough	82	62.1	34	60.7	61.70
Chest pain	6	5.1	0	0	2.5
Fever	59	50.8	33	67.3	59
Irritability	59	50.8	28	57.1	46.2
Forehead sweating	60	45.4	25	44.6	45.2
suck rest suck cycle	34	31.7	18	35.2	26
Feeding difficulty	49	37.12	16	28.6	34.6
Palpitations	6	5.1	6	1.2	3
cyanotic spell	0	0	18	32.1	9.6
Failure to thrive	67	50.7	34	60.7	53.7
Swelling of limbs	5	4.3	3	6.1	5.2
Seizure	0	0	1	1.7	0.5
Headache	0	0	2	3.5	1
Desaturation	15	11.3	56	100	37.7
BP UL-LL difference	1	0.7	0	0	0.53
Growth retardation	67	50.7	34	60.7	53.7
Cyanosis	0	0	51	91	27.1
Clubbing	1	0.7	20	35.7	11.1
CCF	47	35.6	15	26.7	33
Extracardiac anomaly	22	16.6	3	5.3	13.3
Chest retraction	93	70.5	33	58.9	67
Precordial bulge	55	40.1	10	17.8	34.5
Hyperdynamic precordium	29	22.2	7	12.5	19.14
Thrill	24	17.9	3	0.5	14.36
Parasternal leave	7	7.4	5	9	6.4
Abnormal S2	10	7.5	7	12.5	9
Murmur	117	88.6	45	80.3	86.2
Neurological deficit	0	0	2	3.5	1

Table 3: Complications associated with CHD.

Complications	ACHD	ACHD (%)	CCHD	CCHD (%)
Pneumonia	66	50	17	30.3

Continued.

Complications	ACHD	ACHD (%)	CCHD	CCHD (%)
CCF	47	35.6	15	26.7
Brain abscess	0	0	2	3.5
Thromboembolism	0	0	1	1.8
Cyanotic spell	0	0	18	32.1
Growth retardation	67	50.7	34	60.7
Infective endocarditis	1	0.8	0	0

P=0.013, which is less than 0.05 shows significance.

Table 4: ECG findings in CHD.

ECG changes	ACHD (N)	Percentage (%)	CCHD (N)	Percentage (%)	N	Total (%)
Normal	30	22.7	2	3.6	32	17
LAD/LVH	50	37.9	4	7.1	54	28.7
RAD/RVH	16	12.1	26	46.4	42	22.34
CVH/BVH	3	2.2	0	0	3	1.6
RSR'+RVH/RAD	4	3.0	2	3.6	6	3.2
RSR'+LVH/LAD	3	2.3	0	0	3	1.6
RAD/BVH	2	1.5	1	1.8	3	1.6
LAD/BVH	5	3.8	2	3.6	7	3.7
Sinus tachycardia	2	1.5	5	8.9	7	3.7
Himalayan p wave	0	0	1	1.8	1	0.53
Deep q in lead I and Avl	1	0.7	0	0	1	0.53
Not done	17	12.9	12	21.4	29	15.4
Total	132	100	56	100	188	100

P value is less than 0.05 shows significance.

Table 5: Echocardiography diagnosis (ACHD).

ECHO (ACHD)	N	Percentage (%)
VSD	38	20.2
VSD+PAH	15	8
VSD+ASD+PAH	7	4
VSD+ASD+dextrocardia	1	0.5
VSD+ASD	6	3
VSD+PDA	2	1
PDA	15	8
PDA+PAH	1	0.5
ASD	10	5.3
ASD+PAH	2	1
PS	5	2.6
AS	2	1
COA	1	0.5
PDA+BAV+AS	1	0.53
VSD+PDA+PAH	1	0.53
AVSD	3	1.6
AVSD+PS	1	0.53
VSD+PS	4	2.12
PDA+PS	1	0.5
ASD+PS	4	2.12
VSD+COA+PAH	3	1.6
Peripheral PS	3	1.6
ASD+PDA	4	2.12
VSD+ASD+PDA+PAH	1	0.53
Alcapa	1	0.53
Total	132	70

P value is less than 0.05 shows significance.

Table 6: Echocardiography diagnosis (CCHD).

ECHO (CCHD)	N	Percentage (%)
TOF	20	10.6
Fallot's pentalogy	3	1.6
TOF+PDA	2	1
AVSD+TOF	1	0.5
D-TGA+VSD	4	2
D-TGA+VSD+PAH	2	1
D-TGA+VSD+PS	2	1
TAPVC+PAH (supracardiac, cardiac, mixed)	4	2.1
D-TGA+VSD+ASD+PDA	2	1
D-TGA+ASD+PDA	1	0.5
D-TGA+ASD+VSD	1	0.5
COMMON ATRIUM	1	0.5
DORV+VSD+PAH	2	1
DORV+VSD+pulmonary atresia	3	1.5
DORV+AVSD+pulmonary atresia	2	1
AVSD+pulmonary atresia	1	0.5
Ebstein's anomaly	1	0.5
SV+ASD+PDA	1	0.5
SV+ASD+PS	1	0.5
Truncus arteriosus	2	1
Total	56	29.7

P=0.013, which is less than 0.05 shows significance.

Table 7: clinic-echo correlation.

Variables	Clinico (only)-ECHO correlation, n (%)	Clinical + CXR+ ECG with ECHO correlation, n (%)
Correlated	60 (32)	74 (39.4)
Partially correlated	64 (34)	60 (32)
Not correlated	64 (34)	54 (28.6)
Total	188 (100)	188 (100)

Table 8: Clinico (clinical+CXr+ECG)-echo correlation of major individual CHD (%).

CHD	Correlated	Partially correlated	Not correlated
VSD	42.7	37.8	19.5
ASD	37.5	25	37.5
PDA	52.3	14.2	33.5
AS	0	0	100
PS	60	40	0
AVSD	50	0	50
VSD+ASD	0	100	0
VSD+ASD+dextrocardia	100	0	0
ASD+PAH	100	0	0
VSD+PDA	0	100	0
VSD+PAH	100	0	0
TOF	61	17.3	21.7
TGA	37.5	12.5	50

P value 0.04 shows significance.

DISCUSSION

The incidence found was 1.86% comparable with studies by Kondapalli et al (1.56%), Gupta et al (2.3%).^{5,7} Most common age group of presentation of CHD was 1 month-1 year 71.2% followed by under 5 and above 5 in 15.9%

and 12.7% respectively similar to other studies.^{8,9} There was a male preponderance of 97 (51.5%) over female 91 (48.4%). Male: female ratio was 1.06:1. Hospital based studies from India have reported a male preponderance in children with CHD to be 52.4-66.7%.^{10,11-14} There was sex preponderance of VSD, COA, AS, PS, TOF, TGA,

Fallot's pentalogy observed to be more common in male whereas ASD, PDA, TAPVC, Truncus arteriosus, Ebstein's anomaly more common in female. Extracardiac anomalies (ECA) were found in 13.3% of cases which is comparable to study by Pandey et al who reported 8.5%.¹⁵

Among ACHD, most common presentation was breathlessness followed by feeding difficulty, forehead sweating, fever, failure to thrive and among CCHD, breathlessness, bluish discolouration of body, failure to thrive. Though cyanotic spell constituted only 32.1% cases of CCHD but were specific to CCHD as it was not found in ACHD. Almost similar observations have been reported by Kuntal, Ashish and Sharmin as well with few discrepancies.¹⁶⁻¹⁸ Clinical signs revealed leading findings in ACHD group were murmur (88.6%), chest retraction (70.5%), growth retardation (50.7%), precordial bulge (40.1%), CCF (35.6%) and in CCHD group four leading clinical signs were desaturation (100%), murmur (80.3%), cyanosis (91%), growth retardation (60.7%), clubbing (35.7%) comparable to other studies.^{15,19} No cyanosis was seen in ACHD but clubbing seen in 1 case of VSD with infective endocarditis. Cardiac findings revealed murmur (86.2%) with or without thrill were the most frequently observed feature. The 13.8% CHD had no murmur which reveals that presence or absence of a murmur does not assure either the presence or absence of significant CHD. Anisworth et al found 48% of CHD in infancy without heart murmur and Shima et al found 40% CHD without murmur and cyanosis.²⁰⁻²² There was an overestimation of holosystolic murmur as discrimination of exact type of murmur whether early or late or pan systolic murmur or diastolic murmur could not be done in many cases of CHD. It was noted that in 5 cases of ACHD diagnosed clinically, only desaturation was seen but cyanosis was not noticed which eventually diagnosed as CCHD in echocardiography. In such cases, clinical presentation of desaturation was attributed to severe pneumonia but CCHD of increased pulmonary blood flow was not considered. Five cases of CCHD had desaturation without any cyanosis which ultimately diagnosed to be 2 TAPVC, 1 DORV+VSD, 1 Single ventricle+ ASD+ PDA and CCHD associated with anemia where cyanosis was not detectable.

Most common complication was pneumonia, growth retardation. CCF was found in 31.9% in ACHD and 26.7% in CCHD with an overall 29.3% in contrast to a study showing 23.7% in CHD.²³ TOF rarely present with CCF, but this study showed 2 cases of TOF with CCF due to severe anemia. Most cases of TGA and DORV presented in CCF were in early infancy and were associated with multiple lesions as evidenced by standard literature.^{24,25}

Clinically specific diagnosis could be given for 67.1% cases, in rest 33% we were unable to give specific diagnosis After a provisional clinical impression, chest x

ray and ECG was done. The 84% of CHD cases had abnormal CXR findings and overall common findings were cardiomegaly+ pulmonary plethora 21.8%, cardiomegaly 14.9%, cardiomegaly with opacity 12.2%. Among ACHD cardiomegaly+ pulmonary plethora 21.2% being most common and in CCHD, common abnormal findings were booting shaped heart +oligemia 25%, cardiomegaly+ plethora 23.2%, cardiomegaly+ oligemia 10.7%, egg on side+ cardiomegaly+ plethora 9%. ECG abnormality found in 79.8% cases. The 15% cases ECG couldn't be done due to COVID pandemic and other reasons.

Out of 188 CHD cases, clinically without the aid of CXR and ECG, ACHD constituted 70% and CCHD constituted 30%. VSD was the most common clinically diagnosed CHD (45.13%) followed by PDA (12.7%), ASD (1.6%), TOF (3.2%), Fallot's physiology (16.5%), transposition physiology (9%). 68.6% ACHD and 31.3% CCHD were clinically detected supported by CXR and ECG, of which most common CHD were VSD (43%), TOF (12.2%), PDA (11.1%), TGA physiology (6.4%), TGA+VSD (4.4%). The 77.5% of the cases diagnosed clinically were within the 8 common diagnosis of VSD, ASD, PDA, COA, AS, PS, TOF, TGA.^{26,27} Rest were having combined defect or complex CHD. After CXR and ECG, final clinical diagnosis was altered. Among ACHD, 85 VSD changed into 82 VSD cases, 24 PDA into 21 PDA, 3 ASD into 8 ASD, 2 PS into 5 pulmonary stenosis, VSD+ dextrocardia into VSD+ ASD+ dextrocardia, 3 cases of ACHD Lt to Rt shunt into 1 case of ACHD with shunt lesion, 4 ACHD with obstructive lesion into 2 ACHD with obstructive lesion and 5 cases of no categorisation changed to only 2 cases. Apart from that few new diagnoses like 2 VSD+ASD, 2 AVSD could be done. Similarly, among CCHD, 6 cases of TOF changed to 23 TOF, 31 Fallot's physiology to 13, 17 Transposition physiology to 12, 3 cases of no categorisation into 2 cases and 8 TGA could be diagnosed only with the help of CXR and ECG. 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.

Echocardiography revealed 70.2% ACHD and 29.7% CCHD. VSD (28.2%) was the most common CHD, TOF (10.6%), PDA (8.5%), VSD+ ASD (7.5%), TGA+VSD (3%), TAPVC (2%) etc.^{15,28} Though VSD most commonly observed but we found only 28.2%. This can be explained by the fact that apart from isolated VSD, we also found VSD associated with other cardiac defects like VSD+ASD, VSD+PDA, VSD+COA, VSD+PS. A higher frequency of complex CHD with multiple defects 63 (33.6%) detected by echocardiography in comparison to all other studies. This could be attributed to improvement in skill and analytical judgement of pediatric cardiologists in interpreting echocardiography.

Clinical correlation with ECHO without the aid of CXR and ECG was found in 31.9%, partial correlation was in 34.04% which was less in compared to Mitchell et al who found accurate correlation only clinically in 44% cases without CXR and ECG.²⁹ Clinico (CXR+ECG)-echo correlation was accurate in 39.4% of CHD, partial correlation in 31.9% cases and no correlation in 32% cases. The high percentage of partial and no correlation is due to inability to appreciate abnormal pulse, abnormal S2, missing four limb blood pressure measurement, categorisation of different types of murmur. So, we missed diagnosis like PAH, COA, PS, AS and complex heart diseases.

Though VSD was most commonly seen clinically but clinico-echo correlation was maximum for VSD+PAH (100%), VSD+ASD+ dextrocardia (100%), ASD+PAH (100%) which were diagnosed clinically during the end of our study due to upgradation of our skill in due course of conducting the study. Another fact was 61% correct diagnosis was done for TOF, 60% pulmonary stenosis, 52.3% PDA, 50% ECD which comes above VSD 43.2%. This reemphasises that typical findings in clinical CXR and ECG is an important parameter to make a correct diagnosis. Our study has comparable individual correlation of CHD with study by Pestana among all common lesions except VSD and COA.³⁰ 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 overdiagnosis 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 71.7% which is comparable to few other studies like Tandon et al (80-85%), Klewer (81%).^{31,32} 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.

The increasing dependence of newer cardiologist on non-invasive techniques is imperative. The accuracy of clinical diagnosis may vary with examiner discrepancy by training, acumen and experience or by lack of typical findings in a particular case. However, before any operative intervention is undertaken these initial diagnoses must be confirmed by imaging techniques.

CONCLUSION

Our study showed clinico-echo correlation in 32%, with inclusion of CXR and ECG it was 39.4%. So, the role of CXR and ECG cannot be underestimated. 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.

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