

Spectrum of Cyanotic Heart Defects and Their Modes of Presentation at Cardiology Clinic in a Tertiary Care Hospital.

Akash Gupta¹, Shaad Abqari², Tabassum Shahab³, MU Rabbani⁴, S Manazir Ali⁵, Uzma Firdaus²

¹Resident, Department of Pediatrics, J. N. Medical College, AMU Aligarh

²Assistant Professor, Department of Pediatrics, J. N. Medical College, AMU Aligarh.

³Professor, Department of Pediatrics, J. N. Medical College, AMU Aligarh

⁴Professor, Centre of Cardiology, J. N. Medical College, AMU Aligarh.

Received: September 2016

Accepted: October 2016

Copyright: © the author(s), publisher. It is an open-access article distributed under the terms of the Creative Commons Attribution Non-Commercial License, which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

Background: Cyanotic Congenital heart defect (CHD) is the high risk group which requires prompt medical attention. Immediate management can drastically alter the natural history otherwise most of the children will succumb to their defect very early in infancy. It is thus important to have reliable information of the profile of various cyanotic CHDs as well as their mode of presentation for the early detection. **Methods:** The study was carried out in Department of Pediatrics and Center of Cardiology, Jawaharlal Nehru Medical College, Aligarh. All patients referred with complaints or clinical examination suggestive of congenital heart defects were further evaluated with echocardiography. On echocardiography patients having congenital heart defects were included as cases which were further divided into cyanotic and acyanotic heart defects, preterms having PDA and PFO and those with acquired heart defects were excluded. The profile and mode of presentation of various cyanotic CHDs was further described in detail. **Results:** Acyanotic heart defects were 290(72.50%) of the total heart defects, while the contribution of cyanotic heart defects was 110 (27.50%). Out of all CHDs, VSD was the most common lesion with contribution of 152 (38.00%) cases, while among the cyanotic heart defect, Tetralogy of Fallot (TOF) was the most common lesion (18% of total cases). Spectrum of various cyanotic lesions were TOF 65.45%, Single Ventricle 12.72%, TAPVC (Total Anomalous Pulmonary Venous Connection) 8.18%, TGA(Transposition of Great Arteries) 7.27%, Tricuspid Atresia 3.63%, Ebsteins malformation 1.81% and Truncus Arteriosus 0.90%. **Conclusion:** Profile of various cyanotic CHDs was similar to the previous studies, however spectrum of various CHDs was quite wide, a large number of cases were missed in infancy and presented late.

Keywords: Cyanotic Congenital heart disease, profile, TOF, TAPVC, Single Ventricle.

INTRODUCTION

Congenital heart defect is the commonest among birth defects in our country. In India, lack of screening tools, poor availability of echocardiography, lack of awareness and poor socio-economic status of people contribute towards poor detection of CHDs and it is not rare to encounter a cyanotic heart defect for the first time when it presents with one or other complications. Knowledge about profile and age of presentation of cyanotic defects will help in identifying children who are at utmost need of medical and surgical care and to plan the strategies for early identification of this high risk group.

Name & Address of Corresponding Author

Dr. Shaad Abqari
Assistant Professor,
Dept of Paediatrics,
J. N. Medical College,
AMU Aligarh.

MATERIALS AND METHODS

All children suspected of congenital heart disease presenting to Pediatric OPD/IPD/Nursery, on the basis of history and clinical examination were included. A suspected case was defined as any child with spo₂ <93% at room air/or visible cyanosis, any child with h/o frequent chest infections /unexplained failure to thrive/feeding difficulty/effort intolerance, unexplained CHF, murmur, abnormal ECG, abnormal heart sounds, abnormal Blood Pressure, differential peripheral pulses and abnormal chest X-Ray. Parents of children who had not given consent of participating in the study and cases of acquired heart disease (RHD etc) were excluded. All children were then screened through ECG and Chest X-ray and the diagnosis was confirmed by Echocardiography.

A total of 1154 subjects were screened by echocardiography during February 2014 to September 2015. Normal echocardiography were

present in 754, total numbers of CHDs detected were 400. Acyanotic heart defects were 290(72.50%) of the total heart defects, while the contribution of cyanotic heart defects was 110 (27.50%).

RESULTS

Cyanotic heart defects constituted 27.50% of total CHDs. Spectrum of various cyanotic lesions were as follows: TOF 65.45%, Single Ventricle 12.72%,

TAPVC 8.18%, TGA 7.27%, Tricuspid Atresia 3.63%, Ebstein malformation 1.81% and Truncus Arteriosus 0.90%. Tetralogy of fallot having most favourable natural history among all the cyanotic heart defects was detected beyond first decade in 4 cases, two of which presented with brain abscesses. Rest of cyanotic heart defects were common in infancy with patients with TAPVC presented very early in newborn period, however 2 cases of supracardiac TAPVC presented beyond 5yrs of age.

Table 1: Spectrum of cyanotic heart disease according to age of presentation.

S.no.	Diagnosis	<1month	1month-12month	>12month-5years	>5years-11years	>11years	Total
1.	Tetralogy of fallot (TOF)	6(5.50%)	18(16.36%)	30(27.27%)	14(12.72%)	4(3.63%)	72(65.45%)
2.	Single ventricle	6(5.50%)	7(6.36%)	1(0.90%)	0	0	14(12.72%)
3.	Total anomalous pulmonary venous connection (TAPVC)	3(2.72%)	4(3.63%)	0	2(1.81%)	0	9(8.18%)
4.	Transposition of great vessels (TGA)	1(0.90%)	2(1.81%)	2(1.81%)	3(2.72%)	0	8(7.27%)
5.	Tricuspid atresia (TA)	1(0.90%)	3(2.72%)	0	0	0	4(3.63%)
6.	Ebsteins malformation	0	2(1.81%)	0	0	0	2(1.81%)
7.	Truncus arteriosus	0	1(0.90%)	0	0	0	1(0.90%)
	Total	17(15.45%)	37(33.63%)	33(30.00%)	19(17.27%)	4(3.63%)	110(100%)

Tetralogy of Fallot (TOF)

Tetralogy of Fallot was detected in 72 cases which contributed to 18% of total CHD. TOF was the second most common lesion among all CHD and most common lesion among cyanotic heart defects.

Out of total 72 cases of TOF, 60(83.33%) cases had classical TOF while 10 (13.88%) cases were of TOF with Pulmonary Atresia and 2(2.78%) cases were of TOF with Absent Pulmonary valve. There were 54 males and 18 females.

Table 2: Clinical spectrum of TOF

Age	Murmur screening on	Cyanosis	Cyanotic spell	Cyanosis with exertional dyspnoea	Total
<1mon	3(4.17%)	3(4.17%)	0	0	6(8.33%)
1mon-12mth	2(2.78%)	10(13.89%)	6(8.33%)	0	18(25.00%)
>12mon-5yrs	0	5(6.94%)	10(13.89%)	15(20.83%)	30(41.67%)
>5yrs-11yrs	0	1(1.38%)	3(4.17%)	10(13.89%)	14(19.44%)
>11yrs	0	0	0	4(5.55%)	4(5.55%)
Total	5(6.94%)	19(26.38%)	19(26.38%)	29(40.27%)	72(100%)

Most of the cases of classical TOF had cyanosis as its clinical presentation. In neonatal period, most of the cases of classical TOF were detected on screening of murmur on auscultation. In infancy, most common presentation was cyanosis and patients also presented with cyanotic spells. As age increased, patient presented with cyanosis, exertional dyspnoea and spell in the form of squatting episodes.

Total Anomalous Pulmonary Venous Connection (TAPVC)

TAPVC was another less frequent cyanotic lesion. Out of the 110 cases of cyanotic CHD, 9 cases were of TAPVC with contribution of around 2.25% of all CHD. Out of the 9 cases, 5 (55.56%) cases were of Supracardiac TAPVC, 2 (22.22%) cases of Infracardiac TAPVC and 2 (22.22%) cases were of Cardiac TAPVC, however no mixed TAPVC was

detected which is the commonest as per available literature.

Most of the TAPVC presented in neonatal and early infancy period with complaints of fast breathing, CHF and mild degree of cyanosis. All the 9 cases were sent for surgical correction and are on medical follow up.

Single Ventricle

Out of the 400 cases of CHD, 14 cases were of single ventricle with contribution of around 3.5%. Out of 14 cases 10(71.42%) cases were males while 4 (28.58%) cases were females. Pulmonary stenosis was present in 8 out of 14 cases

Most of the cases of single ventricle had presentation since birth, cyanosis as most common presentation. 10 patients presented with cyanosis only while 4 cases without pulmonary stenosis were having

cyanosis along with features of increased pulmonary blood flow.

Transposition of Great Vessels

Out of 110 cases of CHD, TGA was present in 8 cases (7%). 6 (75%) cases were males while 2 (25%) cases were females. All cases of TGA were associated with VSD and PS. Most of the patient had presentation at birth with cyanosis.

Tricuspid Atresia

Four cases were of Tricuspid Atresia and equally disturbed among males and females. All 4 cases were associated with VSD and PS. Cyanosis was common presenting complaint and 2 cases in infancy period had hypoxic spell. Out of 4 cases, 2 cases had undergone stage 1 repair and are on regular follow up.

Ebsteins Malformation

2 cases were of Ebsteins malformation. All cases were associated with ASD. Presentation was mainly in infancy with cyanosis as common complaint. 2 cases are on regular medical follow up. Cyanosis has improved over the period of time.

Truncus Arteriosus

One case was of truncus arteriosus with presenting at 5 month of age with cyanosis and CHF. Patient had undergone surgical correction and is on medical follow up.

DISCUSSION

Most of the studies had shown male preponderance in having CHDs. Similarly our study also found M:F ratio 1.90 similar to the studies of Hussain^[1] and Kumar^[2] which was 2.08:1 and 1.78:1 respectively. In contrast, Khalid^[3] and Khan^[4] didn't find much of gender disparity. Gender discrepancy is quite striking, might be because of early reporting of male progeny by the parents.

Hussain^[1] and Khan^[4] found that infants constituted 64.10% and 71.00% respectively of all CHDs.

As far as worldwide studies concerned, most of the studies had shown TOF to be 7% to 12% of all CHDs. However study done by Sharmin^[5] which was prospective study had 18% cases of TOF, which was similar to our study. Another study by Smitha et al^[6] had showed TOF to be 13.80% of total CHDs, While study done by Kapoor^[7] had contribution of TOF as low as 4.6% which was similar to studies of Bhat^[8] and Jatav et al^[9] 5.45% and 6.03%.

TAPVC constituted 2.25% of all CHDs. Study done by R Kapoor^[7] and Jatav et al^[9] had less contribution of TAPVC, which was 0.4% and 1.72% respectively.

Study done in India showed marked variation in occurrence of TGA as low as 0.6% in study done by Smitha et al^[6] to as high as 6% in a study by Kumar.^[2]

Single ventricle constituted 3.5% cases of all CHDs. Other studies found very low prevalence of single ventricle. Study done by Sharmin^[5], Hussain^[1],

Kapoor^[7] and Jatav et al^[9] showed following prevalence of single ventricle to be 0.9%, 0.6%, 0.4% and 0.86% respectively.

The spectrum of various cyanotic heart defects is similar to other studies but what is concerning is the late presentation of these defects. Two of the TOF patients presented with brain abscesses and cyanosis and clubbing was completely missed, similarly one of the patient with supracardiac TAPVC with history of failure to thrive and characteristic Chest X-ray was treated on lines of Pulmonary Koch. We have predominance of children with single ventricle or Transposition with pulmonary stenosis which have better natural history as compared to those without PS, again pointing towards delayed detection of these defects. Majority (>90%) of TGAs and TAPVCs will succumb during infancy if left untreated so it become utmost important to detect these defects as early as possible before irreversible Pulmonary hypertension sets in. There is an urgent need for robust screening programmes along with dedicated Pediatric Cardiology Unit for the early detection and management of these defects which have an excellent natural history post surgery and the child can have a near normal life.

CONCLUSION

Profile of various cyanotic CHDs was similar to the previous studies, however spectrum of various CHDs was quite wide, a large number of cases were missed in infancy and presented late.

REFERENCES

1. Hussain M, Tahura S, Sayeed M. A, Rahman M. M, Rahman Mahbubur M, Kar S. K. Past and Present Pattern of Congenital Heart Disease at DSH: A situation Analysis Bangladesh. *J Child Health* 2010; 34 (2): 51-55.
2. Kumar BD, Reddy KR, Elizabeth B. "Study of Incidence of Congenital Heart Diseases in Children of Age Group 1 Month to 12 Yrs". *Journal of Evolution of Medical and Dental Sciences* 2015; 4(07): 1151-1159.
3. Khalid Y, Ghina M, Fadi B, Fadi C, May K, Joseph R, Makhoul G, Hala T. 2006. Consanguineous marriage and congenital heart defects: A case-control study in the neonatal period. *Am J Med Genet Part A* 140A:1524-1530.
4. Inayatullah Khan, Amir Muhammad, Taj Muhammad Pattern of Congenital Heart Disease Gomal Journal of Medical Sciences July-December 2011, Vol. 9(2):174-177.
5. L Shamima Sharmin , M Azizul Haque , M Iqbal Bari , M Ayub Ali Pattern and Clinical Profile of Congenital Heart Disease in A Teaching Hospital TAJ 2008; 21(2): 58-62
6. Smitha R, Karat SC, Narayanappa D, Krishnamurthy B, Prasanth SN, Ramachandra B et al. Prevalence of congenital heart diseases in Mysore. *Indian J Hum genet.* 2006;12(1):11-16.
7. Kapoor R, Gupta S. Prevalence of congenital heart disease, Kanpur, India. *Indian Pediatr.* 2008;45:309-11.
8. Bhat NK, Dhar M, Kumar R, Patel A, Rawat A, Kalra BP. Prevalence and pattern of congenital heart disease in Uttakhand, India. *Indian J Pediatr.* 2013;80(4):281-5.

- Jatav RK, Kumbhare MB, Srinivas M, Rao DR, Kumar P.G., Reddy P.R. et al Int J Res Med Sci. 2014;2(1):186-192.

How to cite this article: Gupta A, Abqari S, Shahab T, Rabbani MU, Ali SM, Firdaus U. Spectrum of Cyanotic Heart Defects and Their Modes of Presentation at Cardiology Clinic in a Tertiary Care Hospital. Ann. Int. Med. Den. Res. 2016; 2(6):PE05-PE08.

Source of Support: Nil, **Conflict of Interest:** None declared