

Down Syndrome with Congenital Heart Diseases : Referral to Echo Lab for Screening and Diagnosis

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Background : Down syndrome (Trisomy 21) is associated with Congenital Heart Disease (CHD) in 40-50% cases. In Bangladesh, parents of Down syndrome cases are visiting to pediatrician for multiple associated comorbidities (Developmental problems, learning problems, hypothyroidism, recurrent infections etc) along with CHD. Though screening of newborns with Down syndrome for congenital heart diseases is recommended in international guidelines, it is not well recognized among the parents in Bangladesh despite of postnatal counseling in hospital deliveries or during visiting pediatrician. Most of the parents do delayed evaluation by pediatric cardiologists. Though clinical findings along with ECG & Chest xray are tools for diagnosis of congenital heart diseases, Echocardiography is still considered the best noninvasive diagnostic procedure. This study was aimed for detecting congenital heart diseases among down syndrome cases, the age at which cases are referred to echocardiography lab for screening & not the least awareness among parents about association of CHD with Down syndrome.

Materials and methods : In Chattogram Maa Shishu-O-General Hospital (CMSOGH) a retrospective observational study on cases of down syndrome visiting Echo lab over 1 year (Jan 2020 to Dec 2020). A total of 25 cases of down syndrome were selected based on Karyotyping report.

Results : Among 25 cases, 19(76%) of the patients diagnosed for CHD and still a portion of parents (40%) remains unaware of cardiac problems & its association with down syndrome. A number of 9 cases (36%) were detected at the age below 6 months.

Conclusion : A major portion of Down syndrome cases are associated with CHD whereas clinically asymptomatic cases may escape diagnosis in hospital settings. Counseling the parents about its association with CHD & significance of early screening should be a routine practice among physicians.

Key words: Congenital Heart Disease (CHD); Down syndrome; Echocardiography; Screening.

INTRODUCTION

Down syndrome (Trisomy 21) is associated with cognitive impairment and characteristic facial and other dysmorphic features. Affected individuals are more prone to congenital heart defects (Atrioventricular septal defects, ventricular septal defects, isolated secundum atrial septal defects, PDA, Tetralogy of Fallot), gastrointestinal anomalies, leukemia, Alzheimer disease, immune dysfunction, hypothyroidism, diabetes mellitus and problems with hearing and vision. The incidence of down syndrome in live birth is approximately 1 in 750, the incidence at conception is more than twice the rate.¹ The associated co morbidities, most of which evident in the first year of life & drive the parents to quest treatment & counseling. The most frequently observed problems are developmental & learning

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disabilities, followed by congenital heart diseases, endocrine problems, hematological cancers, gastrointestinal anomaly & others. The association between down syndrome and Congenital Heart Disease (CHD) is well known and widely recognized that CHD contributes significantly to the morbidity of children with Down syndrome. The reported incidence of CHD in Down syndrome patients is between 40-60%.² Most frequently observed cardiac anomaly is AV canal defect, 30% of the defect occurs in children with down Syndrome.³ Screening strategy for congenital heart diseases is consisted of physical examinations, ECG, CXR & Echocardiography. Echocardiography was the gold standard for calculating sensitivity, specificity, positive and negative predictive values for major CHD, defined as any heart defect that would typically require intervention during early childhood.⁴ Screening for CHD by clinical examination, chest radiography, electrocardiography or a combination of all of these techniques may fail in the neonatal period. This insensitivity might be due because symptoms are not present and clinical and radiological signs of a left to right shunt may not be developed in the immediate postnatal period owing to high pulmonary vascular resistance at this time.⁵ In CMSOGH, parents of all newborn with down syndrome are counseled about the co morbidities after birth. Some of the parents are advised to exclude other possible health problems while visiting child development center or outpatient department. Early diagnosis of CHD & appropriate measures can further improve the health while struggling with other comorbid conditions among down syndrome cases.

To detect congenital heart disease among Down syndrome cases referred to Echo lab, find out the presenting age for diagnosis of congenital heart diseases among down syndrome and to determine the awareness among parents of respective cases about association & early screening for congenital heart disease.

MATERIALS AND METHODS

A retrospective study was conducted over a period of 1 year (Jan 2019 to Dec, 2019) in Chattogram Maa Shishu O General Hospital (CMSOGH). A total of 25 cases were enrolled with Karyotype proven down syndrome. Age group from newborn up to 12yrs was selected as purposive convenient sampling. All the cases were referred from inpatient Department of Neonatology & Pediatrics and also from Child Development Center (CDC) & outpatient Department to Echo Lab. All of the cases visited echo lab for the first time to evaluate for cardiac anomaly without prior echocardiographic assessment. Echocardiography machine, Vivid S5 by GE healthcare was operated for reporting color doppler echocardiography report.

RESULTS

Among 25 cases of Down syndrome (Karyotype confirmed cases for Trisomy 21) 10 cases were female & 15 were male. Major portion, 19 cases (76%) found to be diagnosed for CHD. Early screening & referral, that is age below 6 months was

documented 9 (36%) in number & above 2 yrs were 4 cases. Most of the cases, 16 (64%) were referred to echo lab for evaluation while attending the child in neurodevelopmental center at age of 6months to 2 yrs. The pattern of congenital heart disease was variable, from simple defects like Atrial Septal Defect/ASD(5) Ventricular Septal Defect/VSD(5), Patent Ductus PDA(2)to complex congenital heart diseases like Atrioventricular Canal Defect/AVCD(1), Pulmonary atresia(1), & Hypoplastic Left Heart Syndrome/HLHS(1). This study revealed only a single case of AVCD among 19 diagnosed cases. Both ASD (20%) & VSD (20%) revealed as common CHD among presented cases of Down syndrome. Three cases (12%) detected as complex CHD among 19 CHD cases. Normal cardiac anatomy with function was revealed in 6(24%) cases & and all were above 1 yr of age. A case of large PDA and two cases of ASD were led to delay diagnosis for late screening or referral. Parental awareness & education about association of CHD with Down syndrome was detected among 60% of parents. All the parents of newborn with Down syndrome were counseled about association of CHD by the physicians.

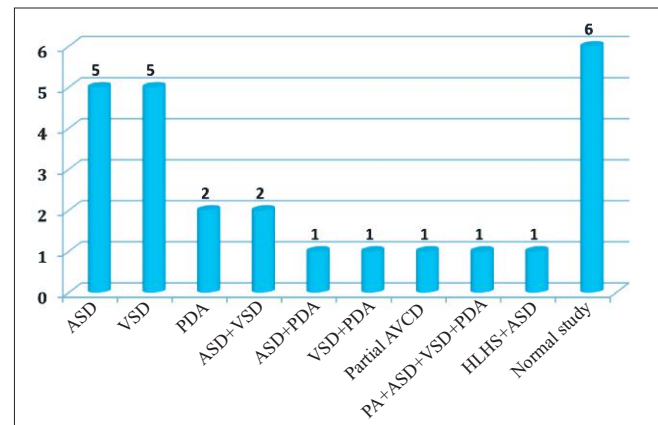


Figure 1 Echocardiography impression for Congenital heart diseases among Down's syndrome (n=25)

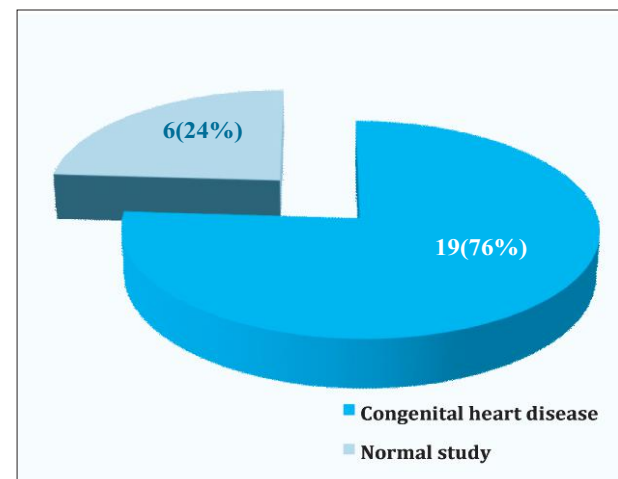


Figure 2 Proportion of CHD among cases of Down Syndrome visiting Echo Lab (n=25)

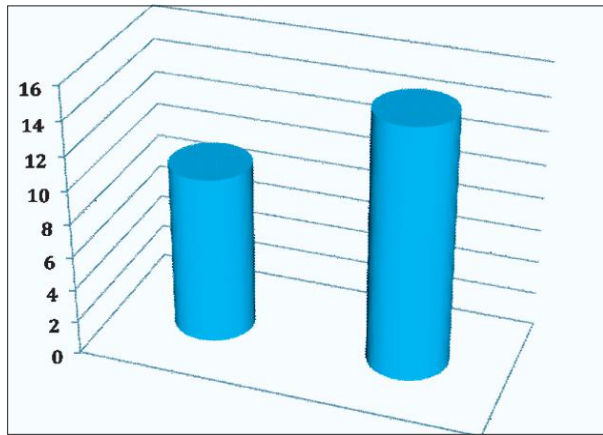


Figure 3 Number of male and female among Down syndrome cases (n=25)

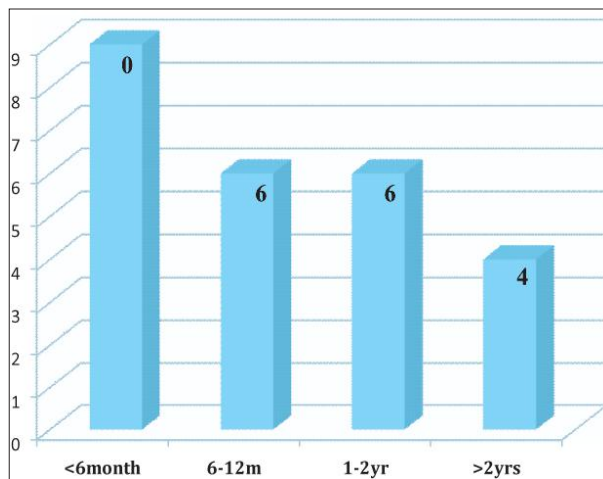


Figure 4 Age at the time of assessment in Echo Lab

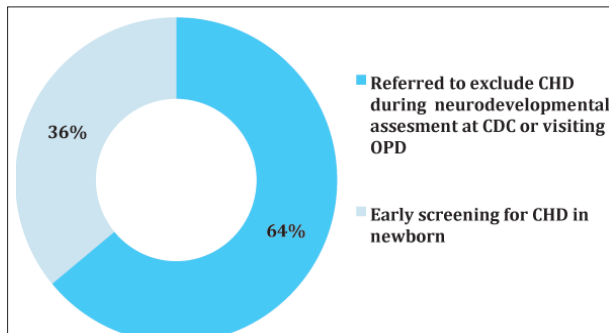


Figure 5 Reason for referral to at Echo lab

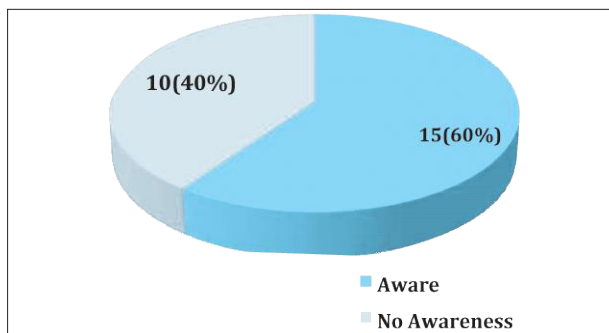


Figure 6 Awareness among parents about early screening for CHD

DISCUSSION

Incidence of down syndrome (Trisomy 21) cases has increased significantly in recent years.⁶ Frequency in general population is 1%. Cardiac malformation is the main cause of mortality in the first 2 yrs of life.⁷ All international guidelines recommend early screening for CHD in Down Syndrome.⁸ On the contrary screening for congenital heart diseases is not routinely conducted for every newborn with down syndrome in Bangladesh. In some institutions assessment is done by clinical examination & referred to an echo lab only after suspicion for congenital heart diseases. This study on screening and diagnosis of Congenital Heart Disease (CHD) in Down syndrome was conducted in Chattogram Maa Shishu O General Hospital (CMSOGH) over 1 yr. A number of 25 cases of Karyotyping confirmed Down syndrome (Trisomy 21) was selected visiting the echo lab for diagnosis of CHD. And the result revealed that significant number that is 19 (76%) cases had CHD. Jesus de Rubens Figueroa et al stated in their study that 160(58%) had CHD among 275 selected cases.⁷ They also stated in their study that 14 cases (8%) had AVCD where this study confirmed only a single case (4%) with AVCD. Most frequently encountered cardiac anomalies in this study, were ASD(20%), VSD(20%) and PDA(8%). Soujanya Bogarapu et al in a previous study on Down syndrome (From Jan 1, 2000 to June 30, 2012) revealed that among 408 eligible infants, 240 had major CHD, of whom 228 had positive screen. Sensitivity & specificity of the screening for CHD were 95% & 41% and positive & negative predictive values were 69% and 85% respectively.⁴ A two year prospective study on early screening by T R J Tubman et al found that among 81 babies, 69 were referred by clinicians and 12 by genetics service for screening. Fifty three were boys and 28 girls. Thirty four babies had CHD & the proportion for male female was similar.⁵ But in this study it is revealed that early screening conducted for 9 cases (36%) only and 3 cases of silent CHD were missing in early infancy due lack of screening & referral. Though the most commonly described defect for down syndrome is complete AVCD, this study revealed only a single case among 19 diagnosed cases.³ Most of the cases (64%) were referred by child neurodevelopmental center of the hospital although all the parents of newborn with down syndrome were counseled about possibility of having CHD. A significant portion of parents (40%) were unaware about early screening & diagnosis of CHD. Moreover in our setting the parents are advised for karyotyping with other investigations results in delay to visit for echocardiographic assessment.

CONCLUSION

Through this study we can conclude that 76% of the down syndrome cases had CHD. Only the clinically apparent cases for Congenital Heart Diseases (CHD) are call for early evaluation. In this regard some cases of atrial septal defects or other clinically silent cases maybe missed in early infancy. So evaluation of all newborns with Down syndrome for CHD even though clinically silent is highly recommended with due counseling the parents.

DISCLOSERS

All the authors declared no competing interests.

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