Congenital Heart Diseases in Patients with Down Syndrome in Gadarif Pediatrics Teaching Hospital, Eastern Sudan

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Received: December 02, 2019; Published: December 17, 2019

Abstract

Background: Congenital heart diseases in Down’s syndrome (DS) result in major implications for the patients, their parents and community. The objective of this study was to determine the frequency and distribution of CHD in children with DS in Gadarif state eastern Sudan.

Material and Methods: This was a cross sectional hospital based study conducted at Gadarif pediatrics teaching hospital from 1st June to 31st December 2018, to study congenital heart defects in Down syndrome. Diagnosis was based on clinical features confirmed by echocardiogram. Age, sex, clinical manifestations, mother’s age, type of heart defect was recorded. Structured questionnaires were used to collect data from the patients and their parents. 30 Down’s syndrome patients with confirmed congenital heart defects were studied.

Results: Comprehensively 27 (86.6%) patients had single cardiac defect, while 4 patients (13.3%) had more than one defect, the most common single diagnosed was VSD accounting for 11 (37%) followed by ASD in 7 (23%) patients. To F was the defect 2 (7%) found only in females, AVSD showed a higher frequency distribution in the female 6/7 (86%).

Conclusion: In the present study the frequency and distribution of CHD patterns are consistent with some international studies, but contradicted with other Sudanese study. VSD and ASD was the most common cardiac lesion. It appears that the frequency of CHD in Down's syndrome varies in different geographical regions even in similar ones.

Keywords: Congenital Heart Disease; Down Syndrome; Gadarif State; Eastern Sudan

Abbreviations

CHD: Congenital Heart Defects; VSD: Ventricular Septal Defect; ASD: Atrial Septal Defect; AVSD: Atrioventricular Septal Defect; PS: Pulmonary Stenosis; ToF: Tetralogy of Fallot; PDA: Patent Ductus Arteriosus; SOB: Shortness of Breath

Citation: Mohammed Ahmed A Ahmed, Samia O Massad, et al. “Congenital Heart Diseases in Patients with Down Syndrome in Gadarif Pediatrics Teaching Hospital, Eastern Sudan”. EC Paediatrics 9.1 (2020): 01-06.
Introduction

Down syndrome (DS) or trisomy 21 is a chromosomal disorder frequently associated with a varied combination of morphological and structural birth defects [1]. Down syndrome is the most frequent chromosome abnormality among newborns [2]. The diagnosis based on the patient's phenotype and later confirmed by the karyotype. Approximately 95% of the carriers of Down syndrome have primary trisomy; the remaining have the translocation (5%) or mosaic (2 to 3%) type [3] and the recurrence risk in the general population is 1% [1]. The clinical manifestations of Down syndrome are numerous and can present in any body system, include mental retardation, hypotonia, short stature, flat nasal bridge, and orthopedic abnormalities which are present in virtually all Down syndrome (DS) individuals, 1% for leukemia [4] and 12% for different digestive system defects [5] and 40% - 60% for congenital heart disease. Congenital heart disease is the most common cause of death among patients with DS and the affected children have an increased risk of mortality [6]. Cardiac defects are the principal cause of mortality in the first two years of life [7,8]. Recently life expectancy of patients with Down syndrome with congenital heart disease significantly increases due to early diagnosis and managements including surgical intervention [9].

Understanding of anatomic features of congenital heart disease in Down syndrome and their complications that causes morbidity and mortality is important for preventive purposes and to improve the quality of patient's life [10,11]. Studies of the frequency of CHD in DS patients are limited in Sudan; there have been no studies of CHD among children with DS in Gadarif state, eastern Sudan.

Aim of the Study

Therefore the aim of this study was to determine the distribution and frequency of patterns of CHD among children with DS in Gadarif pediatrics teaching hospital, Gadarif state, eastern Sudan.

Materials and Methods

This was a cross sectional hospital based study conducted at Gadarif pediatrics teaching hospital from 1st June to 31st December 2018. To study congenital heart defects in Down syndrome. Diagnosis was based on clinical and echocardiogram. Age, sex, clinical manifestations, mother’s age, type of heart defect was recorded. Structured questionnaires were used to collect data from the patients.

Statistical analysis

Data were analyzed using the Statistical Package for the Social Sciences, version 20 (SPSS Inc., Chicago, ILL, USA).

Results

A total of 30 Down syndrome patients diagnosed with CHD were collected in the study period. There were 19 females (63.3%) and 11 males (36.7%), the mean ages of the patients were 4.4 years (the ages ≤ 16 years. 2 Children with down syndrome were born to young mothers (7%), 25 and 29 years of age. Mean maternal age at delivery was 36 years. Most mothers were multiparas 26 (86.7%) and 4 (13.3) were Primiparity. 2 patients had Family history of cardiac diseases.

24 (80%) patients had single cardiac defect, but 6 patients (20%) had more than one defect. The most common single diagnosed was VSD accounting for 11 (37%) (Figure 1).

To F was found only the in the 2 females (7%), AVSD showed a higher frequency distributed in females 6/7(86%) (Figure 2).

All cases had heart murmur of different type according to the cardiac lesions, 7(23%) had heart failure, 13(86%) had fever, 12 (40%) had respiratory distress and 6 patients (20%) had retarded growth.

12 cases had respiratory tract infection in a form of pneumonia, acute bronchiolitis and one had neonatal sepsis. The main causes of death are heart failure, sever pneumonia and neonatal sepsis.
Discussion

This study included 30 patients; females were predominated (63.3%) over males (36.7%), similar to previous studies [12,13]. The children in the present study were aged between 15 day of age - 16 years, and the majority of them were over 5 years of age, this may be due to early neonatal death of DS at home of complex congenital heart defects or sever bronchopneumonia before reaching hospital. Rodríguez and Reyes [14] reported that cardiac malformation is the principal cause of mortality in the first two years of life. Maternal age is a known risk factor for Down syndrome, in this study the mean maternal age was 36.2 years, similar to study carried out by Kim., et al. [15] in Jamaica who reported that the mean age was 36 years, in Morocco 39 years [16] and 32 years in Brazil [17].

The frequency and pattern of CHD in DS varies between different populations [18,19] even in similar geographical region. In the present study the most common single defects were VSD, ASD and AVSD accounting for 37%, 23% and 20%, of all congenital heart defects,

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respectively. This observation is contrary to other Sudanese study conducted at Sudan Heart Centre, Khartoum which reported that AVSD was most common form [20] but our finding is similar to some studies, in Singapore reported that VSD is the most common subtype of CHD followed by PDA, ASD and AVSD with the frequency of 39.2%, 34.5%, 23.4% and 15.6% respectively [15]. Also VSD was the most common in Aseer (35.3%) [21] and Riyadh studies (43%) [22]. Vida., et al. [18] found that VSD was the most common and AVSD the least common CHD among 349 Guatemalan infants and de Rubens Figueroa., et al. [23] reported that VSD, ASD and PDA were the most common defects in Mexican children with DS. Marino., et al. [24] mentioned that VSD, ASD and PDA embryologically and anatomically are quite different from that of AVSD.

In this study AVSD was observed more frequent in females, this was in line with the study carried out by Pierpont., et al. [25] who reported higher frequency of AVSD in female. On the other hand ToF (7%) distributed just among the females in current study. This in contrast to the result of Pierpont., et al. [25] that found ToF was more frequent in males. The present study revealed that frequency of ToF anomaly was 6.6% agrees with that finding in India (6.6%) [26] and slightly higher than that in Caucasian and Saudi Arabian children with the frequency of 4% and 5.3% respectively [18,27]. These differences might be explained by a potential different susceptibility of gender to different CHD pathogenic pathways for example, AVSD is correlated with extracellular matrix anomalies and ToF with ectomesenchymal tissue migration anomalies [28].

Although there is some similarity of the distribution of patterns of CHD in certain different geographical regions, until now there is no clear explanation for differences in the frequency and distribution of CHD in global region even in similar geographic area, Ferencz., et al. [29] mention that Genetic factors, specific embryological mechanisms and cell characteristics can determine the type of cardiac malformation. Geographic factors and ethnicity may also influence the formation of these abnormalities [23,30].

Conclusion

In the present study frequency and pattern of the CHD are consistent with some international studies that VSD and ASD are the most common type but contradict with other Sudanese study. This frequency is varies in different geographical regions even in similar one. The most common isolated defect was VSD (37%) followed by ASD 23% and AVSD 20%. The main causes of death are heart failure, severe pneumonia and neonatal sepsis.

Recommendations

DS patients should undergo cardiac evaluation earlier in life for the early detection of heart defects and treatment of congenital heart disease to improve the quality of their life.

This study is based on clinical criteria to diagnose DS so genetic analysis in further studies is recommended to detect the genetic factors of these patients. Furthermore, environmental factors should be assessed for better understanding of the differences in the frequency and distribution of CHD in Various geographical regions.

Conflict of Interests
The authors declare that, they do not have any conflict of interest regarding this study.

Acknowledgements
The authors are very thankful for the children included in this study and for their parents.

Author’s Contribution
The Mohammed Ahmed A Ahmed and Samia O Massad authors consider as first author, equally contributed to this work

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Volume 9 Issue 1 January 2020
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