Global Scientific JOURNALS

GSJ: Volume 8, Issue 10, October 2020, Online: ISSN 2320-9186
www.globalscientificjournal.com

Spectrum of Congenital Heart Diseases in Children with Down Syndrome in Ejdybia, Libya.

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Abstract

Down Syndrome:- Is by far the most common and best known chromosomal disorder in humans.
Aim of Study:- to define the frequency and patterns of Congenital heart disease among children with Down syndrome in Ejdabyia North eastern of Libya.
Patients & Methods:- this is prospective descriptive study include all phenotypic down syndrome children referred to Echo department at AL-Mugref Teaching Hospital, Ejdybia North eastern of Libya. from 1\4\12019 to 31\8\2020 total 50 case aged between 2 day to 8 years and verbal consent was taken from guardiants of all participants using GE Vivid S5 Echo Machine. Data was analyzed using Excel spread sheets.
Results:- out of 813 cases 50 (6.1%) case were down syndrome included in our study. 29 (58%) of them were female and 21 (42%) were male with Female : Male ratio 1.3: 1.
46 (92%) of cases have congenital heart disease. where 40 (87%) have single cardiac defect & the most common defect was AV Canal 27 (54%) case & other form include ASD 6 (12%), VSD 3 (6%), PDA 3(6%) and TOF 1 (2%). the combined lesions were present in 6 (13%) cases include 3(6%) (TOF+ AV Canal) and 3 (6%) (VSD+ ASD+ PDA). **Conculsion:-** Highly prevalence of CHDs among Down syndrome patients with Female dominance. AV Canal was the most common Congenital Heart lesions. So, important to early referral and screening for CHDs in DS. **Key Ward:-** Down Syndrome, CHDs.

Abbrevations:- DS:- Down Syndrome, **CHDs:-** Congenital Heart Diseases, **AVCanal or AVC:-** Atrioventricular canal, **ASD:-** Atrioseptal defect, **VSD:-** Ventricular septal defect, **PDA:-** Patent ductal arteriosus, **TOF:-** Tetralogy of fallot.

An introduction

Down Syndrome:- Is by far the most common and best known chromosomal disorder in humans and the most common cause of intellectual disability. It is primarily caused by trisomy of chromosomal 21 which gives rise to multiple systematic complications as part of the syndrome. However, not all defects occurs in all patient and there is a wide range of phenotypic variation.[(1)]

It occurs in around 1in ever 700 pregnancies , the research suggests that there is a higher chance if the mother is older than 35yrs of age. It is characterized by intellectual disability, dysmorphic facial features and other distinctive phenotypic traits.[(2)]

The association between Down syndrome (DS) and congenital heart disease (CHD) was first recognised by Garrod in 1894 and it is the most common cause of death in people with Down syndrome in the first 2 year of life. Therefore, echocardiography is recommended for early detection of CHDs which may help to prevent many complications.[(3)]

Aim of Study:- to define the frequency and patterns of Congenital heart disease among children with Down syndrome in Ejdabyia North eastern of Libya.

Patients and Methods:-

Study Design & Sitting:- this is prospective descriptive study include all down syndrome children referred to Echo department at AL-Mugref Teaching Hospital, Ejdybia North eastern of Libya.

Study Population:- All potential cases underwent full clinical assessment including phenotypic features that suggested Down Syndrome are included in our study(n= 50 cases aged between 2days to 8years) with verbal consent were taken from the guardiants of all the participants. Echocardiography examination was performed using a GE Vivid S5 ultrasound Machine.

Study Period:- 17 month from 1\ April\ 2019 to 31\8\2020 for data collection and rest for data analysis & writing.

Data analysis:- Data were exported from collected database to Excel spread sheets in preparation for data analyses.

Results:- out of 813 cases 50 (6.1%) case were down syndrome included in our study. where 29 (58%) of them were female and 21 (42%) were male. 46 (92%) of cases have congenital heart disease.

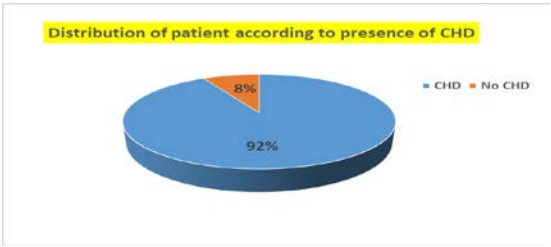

Figure 1: Distribution of Patient according to presence of CHD

where 40 (87%) have single cardiac defect, the most common defect was AV Canal 27 (54%) case & other form include ASD 6 (12%), VSD 3 (6%), PDA 3(6%) and TOF 1 (2%). the combined lesions were present in 6 (13%) cases include (TOF+ AV Canal) 3 (6%) and (VSD+ ASD+ PDA) 3 (6%).

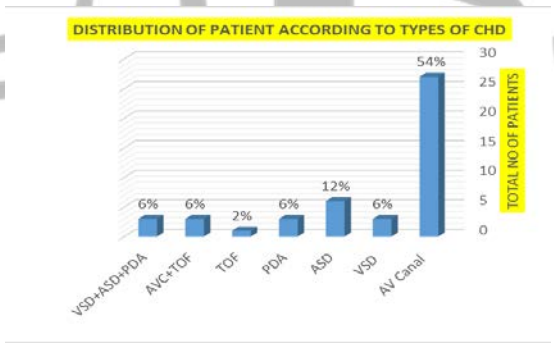

Figure 2: Distribution of patient according to pattern of CHD

Where the female predominance seen in cases of AV Canal, PDA, VSD,& (AVCanal+ TOF) & TOF.

Male dominance seen in (VSD+ ASD+ PDA).

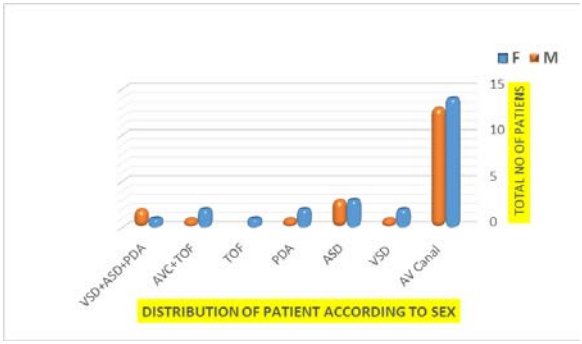

Figure 3: Distribution of patient according to Sex.

Discussions

7.1: Distribution of patient according to presence of congenital heart disease.

Congenital heart disease are common in children with Down syndrome where our study showed that:

-among 813 cases , 50 (6.1%) of them were down syndrome with prevalence of congenital Heart disease among them was 92% and this is going with (Ali SK. 2009), (Shrestha, M., & Shrestha, U. 2013), (Mustafa Asani , Ibrahim Aliyu et al. 2013), (Wilson E Sadoh, Fidelis E Eki-udoko, et al. 2018).

- The female preponderance with **F:M** ratio was 1.3: 1

where 29 (58%) of them were female and 21 (42%) were male and this is going with (Chika O Duru, Olukemi O Ige, Frances S Okpokowuruk, et al. 2020), (El-Shazali O, Ahmed H, El-Shazali H. 2015), (Ahmed Muntha, Tamirat Moges. 2019), (Beatriz E. Bermudez, Sandra L. Medeiros, et al. 2015), (Mourato FA, Villachan LRR, et al. 2014), (Elmagrpy Z, Rayani A, et al. 2011).

7. 2: Distribution of patient according to pattern of congenital heart disease.

It appears that the distribution and frequency of CHD in DS varies in different geographical regions.

In this study, 40 (87%) of patient have single cardiac lesion and the most common a cyanotic cardiac defect were AV Canal (54%) followed by ASD (12%) and these are going with (Wilson E Sadoh, Fidelis E Eki-udoko, et al. 2018), (De Rubens Figueroa J, Del Pozzo Magaña B, et al. 2003), (Jaiyesimi O, Baichoo V. 2007), (Mohamed M. Morsy, Osama O. Algrigri, et al. 2016), (John Akintunde Okeniyi , Uvie Ufuoma Onakpoya, Ibitoye Samuel, et al. 2017), (Ali SK. 2009). (Mokhtar M. M, Abdel-Fattah. M. 2001), (Park MK. 2014).

Where, the commonest cyanotic cardiac defect was TOF(2%) and this is going with

(John Akintunde Okeniyi , Uvie Ufuoma Onakpoya, Ibitoye Samuel, et al. 2017),

(Asim A, Agarwal S and Panigrahi. 2016), (Chika O Duru, Olukemi O Ige, Frances S Okpokowuruk, et al. 2020), (Inayatullah Khan, Taj Muhammad. 2012).

There are wide difference in combined lesion Types among Down Syndrome patient's studies, in our study 6(13%) of cases have (AVC+ TOF & VSD+ ASD+ PDA) and no other study going with our study result in this point.

Female preponderance in:-

1- AV Canal and this is going with (Ahmed Muntha, Tamirat Moges.2019), (Tereza Cristina Pinheiro, Diogenes FA, et al. 2017), (Elmagrpy Z, Rayani A, et al. 2011), (Muhammad Yaqoob, Jaida Manzoor, et al. 2019), (Almawazini AM, Ali A Sharkawy, et al. 2017).

2- VSD and this is going with (Almawazini AM, Ali A Sharkawy, et al. 2017).

3- PDA and this is going with (Elmagrpy Z, Rayani A, et al. 2011), (Ahmed Muntha, Tamirat Moges. 2019), (Muhammad Yaqoob, Jaida Manzoor, et al. 2019), (Almawazini AM, Ali A Sharkawy, et al. 2017).

4-In (AV Canal+ TOF) and TOF, no study correlate with my result.

Male preponderance in

1-(VSD+ ASD+ PDA) and this is going with (Wilson E Sadoh, Fidelis E Eki-udoko, et al. 2018).

We certainly are strong proponents of providing high-quality health care for people with Down syndrome. We are certainly not in favor of short-changing a person with Down syndrome. However, the first rule of medicine is "Do no harm." We want to provide optimal care for people with Down syndrome. It is important that we continue to study what that means. Sometimes that means providing the same care as people without DS and sometimes that treatment should be modified based on disease patterns in people with DS.[25]

Limitations

1st - The figures reported herein are not population based and of only one center.

2nd -The cytogenetic studies were not performed and the diagnosis was mainly based on clinical grounds. As a result we could not comment on the frequency of CHD in different chromosomal alterations of DS.

3rd - The frequency was not compared with international rates.

Conclusions

1- Prevalence of Down Syndrome in my study was 6.1%.
2-Prevalence of congenital heart disease among Down Syndrome was 92% with Male to Female ratio was 1: 1.3.
3-AV Canal was the most common CHDs among Down Syndrome patient's.
4-Female preponderance among patient with AV Canal, VSD, PDA, TOF & (AVC + TOF) and Male dominance in (VSD+ ASD+ PDA).

Recommendations:- **Congenital heart disease is highly prevalence in Down syndrome patients so,**

1- All children with Down syndrome should have a cardiac evaluation at birth.
We stress on the importance of early referral and screening for CHDs in DS.

2-The Effort has to be made to do Karyotyping in all suspected cases.
3- National registry for Down syndrome and Congenital Heart Disease would provide more accurate data to study contributing factors for CHD in DS.

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