Congenital heart diseases among patients with Down syndrome consulting cardiology unite in Alsader Teaching Hospital in Basrah

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ABSTRACT

Aim of the study: This study is aimed **to assess the types and the frequency of congenital heart disease among patients with Down syndrome (DS).**

Patients & methods: **Sixty-five patients with DS collected from echocardiographic department in Basrah cardiology center at Alsader Teaching Hospital and a private clinic were evaluated by echocardiography from February 2009 to August 2013.**

Results: **Their ages range from 6 months to 25 year. Of 65 patients with DS diagnosed clinically, 89% have CHD and 11% have normal heart. Most of the cases were isolated congenital heart disease (CHD).Ventricular septal defect (VSD), pulmonic stenosis (PS) and atrial septal defect (ASD) were the commonest isolated CHD respectively while tetralogy of Fallot (TOF) and single ventricle was the commonest complex CHD in DS.**

Conclusion: **This study had concluded that CHD are common in DS and most of the cases are amenable for surgical corrections.**

Keywords: **Down Syndrome, Congenital heart diseases, Echo.**

أمراض القلب الخلقية بين المرضى الذين يعانون من متلازمة داون والذين يراجعون وحدة القلب في مستشفى الصدر التعليمي في البصرة

طريقة العمل: **تم فحص ٦٥ مريض مصاب بمتلازمة داون في شعبة الايكو في مستشفى الصدر التعليمي للفترة من بداية شهر شباط ٢٠٠٩ ولغاية شهر اب ٢٠١٣.**

النتائج: **٨٩% كانوا يعانون من تشوهات خلقية في القلب و١١% كانت قلوبهم سليمة من أي عاهة خلقية .معظم الحالات كانت تشوهات بسيطة وليست معقدة ،واكثرها شيوعا كانت الفتحة الولادية بين البطينين وتضيق الصمام الرئوي والفتحة الولادية بين الاذينين. ومعظم الحالات يمكن علاجها عن طريق التداخلات الجراحية والقسطارية**

الاستنتاجات: وعليه فان نسبة كبيرة من المرضى يعيشون لفترات متقدمة من العمر مما يتطلب من المؤسسات الصحية والاجتماعية توفير الخدمات الملائمة لهذه الشريحة المهمة من المجتمع.

الكلمات المفتاحية: **متلازمة داون، امراض القلب الخلقية، فحص الايكو.**

INTRODUCTION

Down syndrome (DS) is one of the important problem in our society, it is the most common chromosomal disorder,[1,2] with variable clinical presentations including short stature, typical odd face, mental retardation, endocrine abnormalities, gastrointestinal diseases, cardiovascular defects, hematological abnormalities and skeletal abnormalities.[1,3] DS considered as commonest cause of moderate mental retardation, however, most of the patients with DS are trainable patients. [4] The incidence of DS varies from 1:650 to 1:1000 live births all over the world; the recorded incidence was variable from area to area. [5] The congenital heart diseases (CHD) is the most common presentation account for 40 to 60%,[7] while leukemia and gastrointestinal defects are present in 1% and 12% respectively.[5] In Sallie B Freeman et.al. Study they conclude that 4% to 10% of CHD are associated with DS. [4] Cardiac defects in DS are the major cause of mortality in the first two

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years of life.[9] The characteristic cardiac abnormalities seen in DS came from abnormal development of endocardial cushions and results in variety of defects involving the valves and interatrial and interventricular septum. [10] These heart defects cause significant increase in morbidity and mortality of children with DS,[9] who may develop congestive heart failure, pulmonary hypertension, recurrent chest infections and failure to thrive. [11] Independent and intersecting approaches to identify the genes for CHD, that associated with DS, have included mapping genes known to be involved in cardiac development (none of which localized to chromosome 21) and studying rare individuals with CHD and partial duplications of chromosomes 21.[12] In this descriptive study 65 patients with DS were studied by Doppler echocardiography to evaluate the frequency and the type of CHD among patients with DS, in order to plan how we can help such patients.

PATIENTS AND METHODS

A descriptive study was carried out on patients with DS visited the echocardiography unite at Alsader Teaching Hospital and Aleihab private clinic, their age range from 6 moths to 25 year. This study was done from February 2009 to august 2013. DS was diagnosed clinically with their typical clinical features. Whenever it was feasible, karyotyping was done for the patients. A careful history and physical examination were done by the author himself, physical findings looking for typical mangloid face, depressed nasal bridge, low set ears, macroglossia, slanted eyes with epicanthal fold, broad hand with single transverse palmer crease. Doppler echocardiography was performed by the author himself using GE Vivid 3 and Vivid 5.

All collected data were analyzed by SPSS version 15 statistical software.

RESULTS

This descriptive study included 65 patients with DS, that diagnosed clinically and referred for echocardiographic examination at Aleihab private clinic and Alsader Teaching Hospital, 30 patients were male and 35 were female. 20 (31%) of those patients their mother`s age were more than 40 year at time of delivery and 45 (69%) their mother`s age at time of delivery were less than 45 year. Thirty-eight (58%) have consanguity parents and 27(42%) have no consanguity parents. chromsomal analyses was done for only 32 (49%) of them and the others no chromosomal study was done. Of those 32 patients only 22 (69%) had trisomy 21 and the 10 (31%) had no trisomy 21

Table 1. The characteristics of the patients

Gender	• Male: 30(46%) • Female: 35(54%)
Maternal age at time of delivery	• ≤30 year, 15 (23%) • 30-45 Year, 30(46%) • ≥40 year; 20(31%)
Chromosomal analyses	• Done in 32(49%) • Not done in 33(51%)
Trisomy 21	• 22(69%) had positive trisomy 21. • 10(31%) had negative trisomy 21.
Consanguinity parents	• 38(58%) had positive consanguinity parents. • 27(42%) had negative consanguinity parents.

(Table-2), shows the cause of referral to the echocardiographic department, this table shows that 10(15.4%) of patients was referred for routine checking, 12(18.5%) patients were referred for assessment for the cause for recurrent chest infections, 24(37%) patient referred for explanation of central cyanosis and 19(29.1%) patient were referred for explanation of a murmur.

Table 2. The cause of referral for echocardiography

Cause of referral	No. of th patients	%
Routine checking	10	15.4
Recurrent chest infections	12	18.5
Central cyanosis	24	37
Explanation of a murmur	19	29.1
Total	65	100

Table 3. The age distribution of the patients.

Age group	No. of the patients	%
6 months - 1 year	10	15.4
1-2 years	6	9
3-4 years	13	20
4-6 years	8	12.3
6-10 years	17	26.2
10-15	6	9
15-20	3	4.6
> 20 year	2	3.1
Total	65	

Fifty-eight (89%) patients with DS had congenital heart disease and 7(11%) had normal heart, 50 patients out of 58 had isolated congenital heart defect and only 8 patients had complex combined lesions. (Table 4 and 5) describe the distribution of the congenital heart diseases among patients with DS.

Table 4. The percentages of cardiac defects among the patients with DS and CHD

The cardiac defect	No. of patients	%
VSD	15	26.3
ASD secondum	5	8.6
ASD primum	1	1.7
CAVC	10	17
PDA	4	6.9
PS	12	20.7
MVP with MR	2	3.4
TOF	3	5.2
Single ventricle	2	3.4
DORV	1	1.7
VSD and PS	1	1.7
PDA and VSD	1	1.7
PPH	1	1.7
Total	58	1.7

DISCUSSION

Most of the individuals with DS are trainable and they can benefit from social programmers that aimed for stimulation, development and education,[15] and as far as the life expectancy for children with DS are approximately 50 years, [16] therefore, it is important to study this problem and give an idea about the size and the types of CHD among those patients The real incidence of DS during pregnancy is more than twice of the actual incidence among the lived birth,[13,14] so it is one of the important problems all over the world. There were 65 patients with DS visited outpatient and private clinic for echocardiography. In this study 69% of the patients their mother age at time of the delivery was less than 40 year and only 31% their mother age at time of delivery was more than 40 year, this finding is not compatible with well-known fact that the incidence of DS is higher among older women, [17] this can be explained by older women did not ask for medical advice or because of increase birth rate among younger women. This fact also explains why the incidence of DS is variable depending on the maternal age, and this indicates that screening for DS should be done regardless the mother age. In this descriptive study chromosomal analyses was done for 49% of the patients, and of those only 69% had trisomy 21, which is the commonest chromosomal abnormality that cause DS,[18] this can be explained by the fact that other genetic abnormality that cause DS is also present among our patients like mosiasem or translocation of the chrosome 21.so we needs more advance genetic analyses to detect those abnormality. Relative parents were present in 58% of the patients this indicate that DS can be suspected in both those with relative or irrelative parents. (Table-2), showed the cause of referral for cardiac assessment; cyanoses was the most obvious problem for the mothers to ask for medical advice whiles the routine checking is the least, so mothers and doctors should be educated for awareness of possible CHD among

patient with DS (Table-3), shows the age distribution of the studied sample, of patients only 31% their age is more than 20 year this is because most of our patients do not reach such ages or they have no CHD or treated properly by surgical correction and they escape from medical follow up. Eighty four percent of the patients with DS had simple congenital heart disease, others had complex cardiac lesions, and this goes with the same findings in Libya (65%),[21] Mexico (74%),[22] Turkey (78%)[23] and Guatemala[24] (80%). VSD was the most common isolated cardiac defect, found in 26.3% of patents with DS and CHD. This finding goes with same findings in Nepal,[25] India[26], China[27], KSA[28], While in Italy[29], USA[6], Sudan[30] and Libya[21] VSD was the second most common isolated cardiac defect in DS. The second most common isolated abnormality was PS Followed by CAVC. By Sallie *et.al.* in USA study,[6] Palacidi, et al., in Italy [29] and Ali KS et.al. in Sudan[30] CAVC was the first most common abnormality, while no isolated PS was seen in previous studies. ASD secondum was the first common isolated lesion in Ebnagrapy, et al. study in Libya[21] and in Rubens et.al study in Mexico, [22] while in our study ASD was the third common abnormality followed by PDA. The (table-5) showed comparison of our findings with other previous same studies.

Table 5. Comparison of CHDs in DS among different countries.

Cardio pathy	VSD %	CAVC %	ASD %	PDA %	TOF %	COA %	Others
Present study	26.3	17	8.6	6.9	5.2	0	36
Nepal study[25]	22.5	15	10	10	7.5		35
India[26]	26	3	12	5	16	0	39
China[27]	44	15	13	12	13	0	2
KSA[28]	33	23	21	14	5	0	4
Guatemala[24]	28	10	13	29	0	0	22
Italy[29]	25	55	5	5	6	0	4
USA[6]	35	45	8	7	4	1	1
Sudan[30]	23	48	5	7	6	0	11
Mexico[22]	30	9	38	21	1	0	1
Libya[21]	12	19	24	4	2	1	39

We found most of the cases are amenable for surgical or interventional treatment that are available in Iraq so offering of cardiac care for that patient is helpful to increase their life expectancy. This fact is important if we know that those patients had friendly behavior in the society, some of them even can reproduce and most of them are trainable persons.

CONCLUSION AND RECOMMENDATIONS

DS is one of the important problems in our community. Most of them had treatable congenital heart diseases. High percentage of DS is present among young mothers. It is not necessary for patient with DS to have trisomy 21 karyotyping. Cyanoses is the most common clinical problem that makes the parents seek medical advice, this was noticed during crying of the baby in most of the cases, and in most of the cases VSD was large enough to cause early pulmonary hypertension. A wider study with bigger sample size is recommended to have more obvious picture about the size of this problem in Basrah city. Development of educational programs for people and another for medical personals to identify, recognize and easy diagnoses of DS. Putting plans for helping these patients because most of them had treatable CHD. Putting a plan for prevention of DS and by this we decrease the incidence of

CHD, and mental retardation that cause heavy burden on our community.

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