Congenital Heart Disease in Pediatric with Down's Syndrome

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Abstract: The aim of this study is to determine the type and distribution of congenital heart defects (CHDs) in pediatric patients affected by Down syndrome (DS), and compared with previously published studies. Retrospective study of (2108) pediatric patients referred from cardiac clinics to echocardiography department at King Abdulaziz University Hospital, Jeddah, KSA. Data was collected from January 2011 to October 2013. This study was done after obtaining ethical approval from committee of Biomedical Ethics, Faculty of Medicine, King Abdulaziz University. The results indicated that a total of (2108) patients reviewed, 1668 (79%) had abnormal cardiac findings [1546 (93%) Non DS patients (854 (55%) had CHDs and 692 (45%) had other cardiac disease), 122 (7%) patients known case of DS (88 (72%) had CHDs and 34 (28%) had other cardiac disease)] and 440(21%) were had a normal findings. Among the 88DS patients who had CHDs, 42 (48%) had single cardiac abnormalities and 46 (52%) had multiple cardiac abnormalities. The most frequent congenital heart disease was ASD found in 52 (42.6%) patients, followed by PDA in 47(38.5%), VSD in 39(32%), and only 17 patients (13.9%) had AVSD. The concluded that DS is strongly related with CHDs by (72%) and this result is slightly higher to the result published in previous studies. The atrial septal defect (ASD) was the most common congenital cardiac lesion seen in DS patients.
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1. Introduction

Down syndrome (DS) or trisomy 21 is a chromosomal disorder which is often related with different morphological and structural defects. These defects include heart disease, orthopedic abnormalities and other systemic congenital disorders [1].

Among all cases of congenital heart defects (CHDs) 4-10% are related with DS, and 40-60% of DS patients have CHD [2]. Congenital cardiac diseases are considered as the largest cause of death in patients with DS over the first two years of life [3]; so that echocardiography recommended for early detection of congenital heart disease which may help to prevent many complications. Heart defects are usually single, but also they can be multiple. The common CHDs in DS patients are atrioventricular septal defect (AVSD), atrial septal defect (ASD), ventricular septal defect (VSD) and patent ductus arteriosus (PDA) [4].

Heart is hollow muscular organ which have pyramid shape, located within the pericardium in the mediastinum and directed downward, forward, and to the left. The heart walls are made up of a thick layer of cardiac muscle, the myocardium, epicardium and endocardium. In the heart the atrial portion has thin walls and separated by the atrial (interatrial) septum into the right and left atria. Also in the heart there is ventricular portion has thick walls and is separated by the ventricular (interventricular) septum into the right and left ventricles [5].

The heart control the circulation of the blood throughout the body which carries valuable nutrients and oxygen required for permanence of the tissues. Normal heart contracts rhythmically at about 70 to 90 beats per minute in a resting adult. The circulation of heart begins in the right atrium. Deoxygenated blood collects from the upper and lower portion of the body via the superior and inferior vena cava, respectively and enters the right atrium. Now the right atrium is full, deoxygenated blood flows toward right ventricle through the tricuspid valve. Then the blood will pass from right ventricle into the main pulmonary artery through the pulmonary valve. Pulmonary artery enters each of the lungs to re-oxygenate blood. Through the four pulmonary veins newly oxygenated blood return from the lungs to the left atrium. Then the blood passes from the left atrium through a mitral valve and into the left ventricle. The blood pumps from the left ventricle and into the aorta past the aortic valve. The oxygenated blood from this circulation is diffuse to the heart and the whole body through the arterial system [6].

Atrial septal defect (ASD) is a type of CHDs which occur in the septum separating the left and right atria. Defective of this septum cause the blood to flow from the left to the right side of the heart, or vice versa [7]. (See figure, 1) [8]

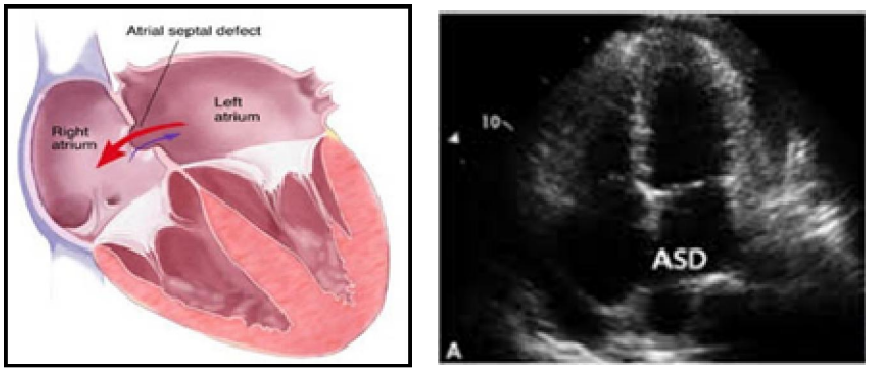

Figure (1): ASD

Patent ductus arteriosus (PDA) means the blood flowing abnormally between two main arteries the aorta and the pulmonary artery. This problem in the heart affects some babies shortly after birth. The two major arteries before birth of baby are connected together by a blood vessel called the ductus arteriosus. The ductus arteriosus vessel after birth, may closed and this change is normal in newborns or it may be remains open (patent). If it is stay opening this allows mixing the oxygenated blood from the aorta with deoxygenated blood from the pulmonary artery [9]. (See figure, 2) [10]

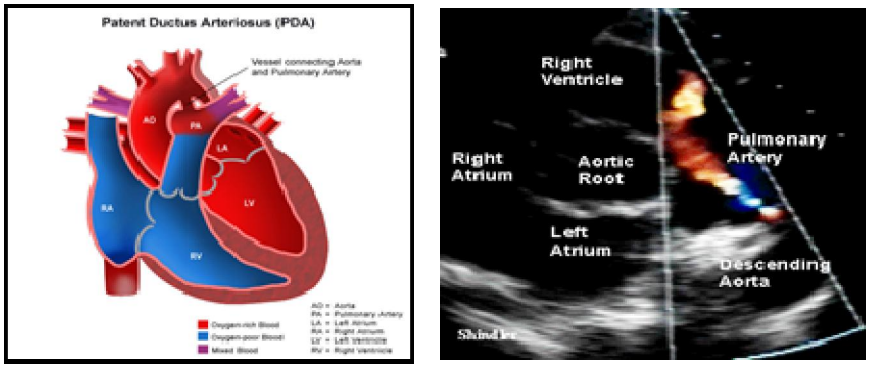

Figure (2) PDA

Atrioventricular septal defect (AVSD) is relatively the common family of CHDs. It is characterized by insufficiency of the atrioventricular septum of the heart [11], (see figure, 3) [12]

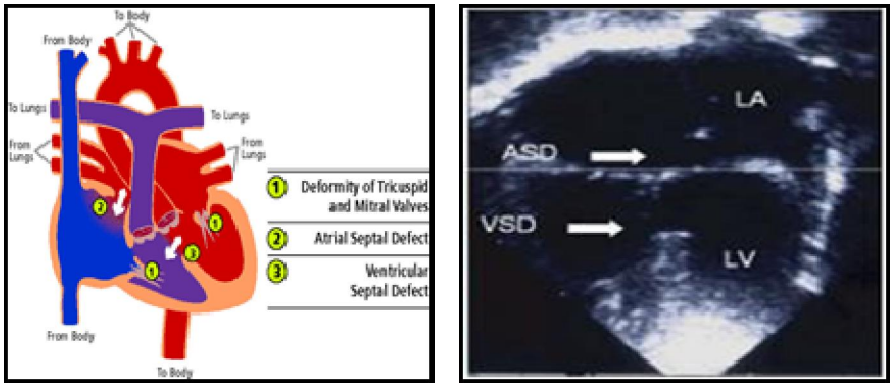

Figure (3) AVSD

Ventricular septal defect (VSD) is a form of CHDs present in the septum separating two ventricles of the heart. This disorder in the septum can appear as either an enlarged foramen oval or lack of tissue of the septum usually extending towards the atrioventricular valves [13], (see figure, 4) [14]

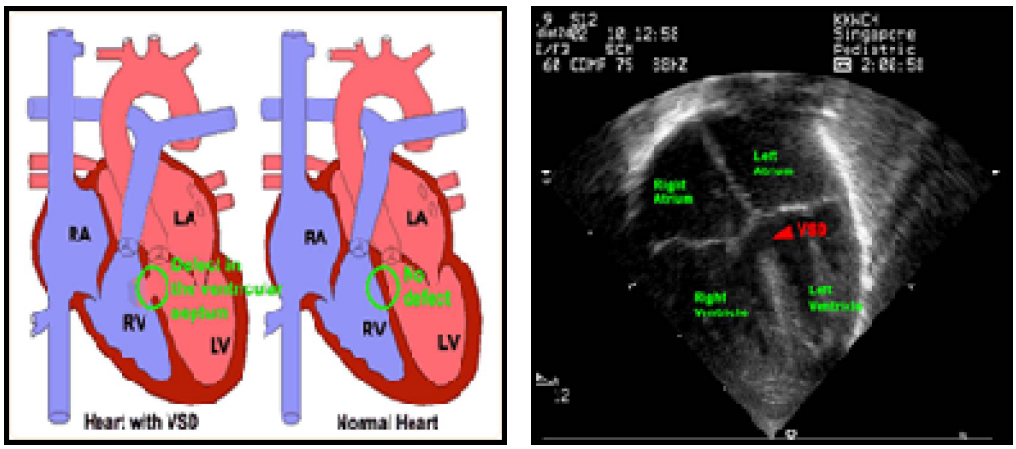

Figure (4) VSD

In 1866 John Langdon Down was characterized as a first DS. According to the National Down Syndrome Association, it is the most common chromosomal disorder, occurring relatively once in every 800 to 1000 births. Down syndrome or trisomy 21 which is often associated with a variety of birth defects including mental retardation, specific facial features, heart defects and other congenital malformations. Pregnancy with DS has a high risk according to the age of the mother. For example, a mother at age 20 has a chance about 1in 2000 of having a child with DS; at 49 years old, she has a chance about 1 in 12, [15], [16].

Echocardiogram or cardiac echo is a branch of ultrasound that uses sound waves to make a moving picture of the heart. There is no special preparation for this test. It is noninvasive and has no known risk or side effect. It is used routinely in the diagnosis and follow-up of patients with any suspected or known heart problems. It is provide useful information like location, size and shape of the heart, pumping capacity and extent of any damage tissue. These modalities of echo are clinically used: two-Dimensional echo (2-D echo), motion- mode echo (M-mode echo) and Doppler Echo. The 2D imaging is the fundamental of echo imaging which allows structures of the heart appear moving in real time in a cross-section view. The cross-sectional views are commonly used in echo are the parasternal long and short axis, and the apical view, subcostal and suprasternal views [17].

Many conditions affect the heart. Previous studies show the prevalence of DS related to congenital heart defects in pediatric patients by 40 to 60%. From our side we are interested to review congenital cardiac abnormalities mainly in DS, and to compare with the previous studies. The aim of this study is to determine the type and distribution of congenital heart defects (CHDs) in pediatric patients affected by DS, and compared with previously published studies.

2. Methods and Material

Study population:

The population of the study is (2108) pediatric patients presented with suspected cardiac problems to echocardiography department at King Abdulaziz University Hospital, Jeddah, KSA. Data was collected from January 2011 to October 2013.

This study has a retrospective nature, and the data was obtained from hospital information system (phoenix). The way followed to collect the data in this study was data collecting sheet. This study was done after obtaining ethical approval from committee of Biomedical Ethics, Faculty of Medicine, King Abdulaziz University.

Method:

Echocardiography examination was performed at the echocardiography department for the all patients enrolled in the study by using echocardiography machine (Philips). A standard echocardiogram used is known as a transthoracic echocardiogram (TTE). The optimal transducer used for transthoracic echo is phased array, which appears on the monitor as a sector image. The typical frequency range of the transducer for pediatrics echo is (3-5MHZ). The cross-sectional view commonly used is apical 4-Chamber View (AP4CH) here the transducer is placed on the chest wall (or thorax) at the apex of heart. (Figure, 5) [17]

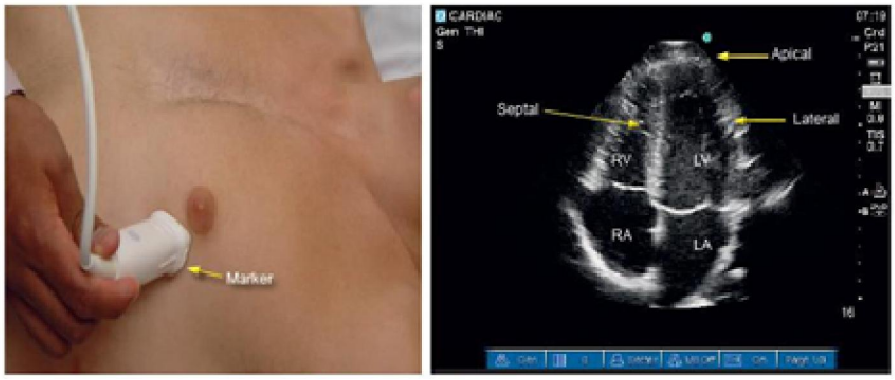

Figure (5) cross sectional view

Statistical analysis:

The data obtained statistically analyzed by descriptive statistic, using SPSS (version 16) and Microsoft Office Excel 2010.

3. Results

Between (January 2011 to October 2013) retrospective study of (2108) pediatric patients referred from cardiac clinics to echocardiography department at King Abdulaziz University Hospital, Jeddah, KSA.

The analysis was based on data from the 2108 patients. A total of 1152 (54.6%) male; 956 (45.4%) female mean age 5.5 years, 4.13 (Standard deviation) **(Tables, 1 and 2).**

In 122 patients with Down syndrome, 66 (54.1%) were male, and 56 (45.9%) were female as shown in **Table (3).**

The echocardiography examination of 2108 total patients included in this study shows 1668 (97%) had abnormal cardiac findings [1546 (93%) cases of Non DS and 122 (7%) known cases of DS patients], 440 (21%) patients had normal findings. **(Tables, 4 and 5)**

Among 1546 (93%) Non DS patients [854 (55%) had CHDs and 692 (45%) had other cardiac disease]. **Table (6)**

In 122 (7%) patients of DS [88(72%) of them had CHD and 34(28%) had other cardiac disease]. **Table (7)**

In1668 Non DS and DS patients who had abnormal cardiac findings [942 (56%) patients had CHDs and 726 (44%) had other cardiac disease]. **Table (8)**

Among all cases of CHDs who were 942 we found 88 (9%) patients with DS. **Table (9)**

Among the 88 DS patients whose had CHD, 42 (48%) had single cardiac abnormalities and 46 (52%) had multiple cardiac abnormalities. **Table (10)**

Among the 122 DS patients the most frequent heart defect was ASD found in 52 (42.6%) patients, followed by PDA in 47(38.5%), VSD in 39(32%), and only 17 patients (13.9%) had AVSD. **(Tables, 11, 12, 13 and 14)**

Table (1) The table shows distribution of gender groups among 2108 patients enrolled in the study.

Gender	Frequency	Percentage
Male	1152	54.6%
Female	956	45.4%
Total	2108	100%

Table (2) The table shows mean and standard deviation for age and gender.

Statistics		
	AGE	**GENDER**
Mean	5.5000	1.4590
Std. Deviation	4.12861	.50037

Table (3) The table shows distribution of gender groups among 122 of DS patients enrolled in the study.

Gender	Frequency	Percentage
Male	66	54.1%
Female	56	45.9%
Total	122	100%

Table (4) The table shows distribution among 2108 patients enrolled in the study.

	Frequency	Percentage
Abnormal	1668	79%
Normal	440	21%
Total	2108	100%

Table (5) The table shows distribution among 1668 patients had abnormal cardiac findings.

	Frequency	Percent
Non DS	1546	93%
DS	122	7%
Total	1668	100%

Table (6) The table shows distribution among 1546 of Non DS patients who have CHDs or other cardiac diseases.

	Frequency	Percentage
CHDs	854	55%
Other Cardiac Diseases	692	45%
Total	1546	100%

Table (7) The table shows distribution among 122 of DS patients who have CHDs or other cardiac diseases.

	Frequency	Percentage
CHD	88	72%
Other Cardiac Diseases	34	28%
Total	122	100%

Table (8) Distribution among 1668 patients had abnormal cardiac findings.

	Frequency	Percentage
NON DS + DS (CHD)	942	56%
NON DS + DS (OTHER DISEASES)	726	44%
Total	1668	100%

Table (9) Distribution of Non DS and DS patients who had CHD among the (942) cases.

	Frequency	Percent
Non DS (CHDs)	854	91%
DS (CHDs)	88	9%
Total	942	100

Table (10) Distribution among 88 DS patients who have single or multiple CHDs.

	CHD	Other Diseases
Single	42	48%
Multiple	46	52%
Total	88	100%

Table (11) Distribution among 122 of DS patients with ASD.

	Frequency	Percent
YES	52	42.6%
NO	70	57.4%
Total	122	100%

Table (12) Distribution among 122 of DS patients with VSD.

	Frequency	Percent
YES	39	32%
NO	83	68%
Total	122	100%

Table (13) Distribution among 122 of DS patients with AVSD.

	Frequency	Percent
YES	17	13.9%
NO	105	86.1%
Total	122	100%

Table (14) Distribution among 122 of DS patients with PDA.

	Frequency	Percent
YES	47	38.5%
NO	75	61.5%
Total	122	100%

4. Discussion

The purpose of this study is to determine the type and distribution of congenital heart defects (CHDs) in pediatric patients affected by Down syndrome (DS), and compare with previously published studies.

Earlier researches suggest that, among all cases of CHDs, 4%-10% are related with DS, and 40%-60% of DS patients have CHD,[1], [18]. In our results we found that 9% of cases are related with DS and this is consistent to the result reported in [1], [18], and also we found that 72% of DS patient have CHD, and this result is slightly higher to the results published in previous studies, [1], [18].

Previous research reported that the most common single congenital cardiac defect in pediatric DS patients was ASD, which was found in (24%) patients, 1other study have reported the most frequent isolated heart defect was ASD by (24%) cases,18.Some cross-sectional studies have even found the arterial duct patency (28.6 %) was the most common single defect, [3].

The current study findings said that ASD was the most common single congenital cardiac defect in 52 (42.6%) of DS patients. Our results revealed no significant difference with what has been reported in previous researches, [1], [18] although these findings are not consistent with previous research, [3].

In conclusion, the study found that the Down syndrome is strongly related to the congenital heart disease with the most common single congenital cardiac defect was atrial septal defect (ASD).

Summary and Recommendation

The goal of this study is determining the frequency and type of commonest CHD in pediatric patients affected by DS. In our results we found that (9%) of cases are related with DS and this is consistent to the result published in earlier study which was reported that 4%-10% of cases are related with DS. DS is strongly related with CHD by (72%) and this result is slightly higher to the result

published in previous studies which was said that 40%-60% of DS patients have CHD. From the results we found also in this study that the most common congenital cardiac lesion seen in DS patients was ASD that is found in 52 (42.6%) patients, followed by PDA in 47(38.5%), VSD in 39(32%), and only 17 patients (13.9%) had AVSD.

Recommendations:

The study recommends that if the study to be in prospective way in order to: add the mother's age to know it's relation to development of DS. Follow up of DS patients to know the common age and cause of death. Finally, increase population by selecting more different areas.

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