

## The Common Types of Congenital Heart Disease in Down Syndrome Patients at King Abdul-Aziz Medical City-Jeddah

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### ABSTRACT:

**Background:** Down syndrome (DS) is a type of mental retardation caused by extra genetic material on chromosome 21. Congenital heart defects (CHD) are the common abnormality in this syndrome. This study is to determine the frequency of congenital heart defects (CHD) in Down syndrome (DS) patients at King Abdul-Aziz Medical city-Jeddah (KAMC-JD) and to characterize the types of CHD.

**Methods:** A retrospective chart review study of 178 Down syndrome patients born in or referred to (KAMC-JD) between January 2000 and January 2014 was conducted. For all patients, the diagnosis of Down syndrome was confirmed by karyotyping. DS patients with no echocardiography results were excluded.

**Results:** a total of 178 DS patients were included in our study. CHD were detected in 110 patients (61.8%) while the echocardiography was normal in 68 patients (38.2), out of those 110 patients, 62 patients (56.4%) were male and 48 patients (43.6%) were female. Seventy-four patients (67.3%) had isolated CHD, while combined CHD were detected in thirty-six patients (32.7%). For both sexes, the most common detected lesions, either isolated or combined, were the atrial septal defect (ASD) in 71/110 (64.5%), ventricular septal defect (VSD) in 39/110 (35.5%), and patent ductus arteriosus (PDA) in 25/110 (22.7%). On the other hand, atrioventricular septal defect (AVSD) in 16/110 (14.5%), and tetralogy of Fallot (TOF) in 2/110 (1.8%) were the least common lesions.

**Conclusion:** The frequency of CHD in our study was higher than the international studies and most of the national studies. The most common CHD detected in our study (ASD) was different than other lesions in international and domestic studies.

**Keywords:** Trisomy 21, Down syndrome, congenital heart defects.

### INTRODUCTION

Down syndrome (DS), or trisomy 21, is "a type of mental retardation caused by extra genetic material on chromosome 21"[1]. It is considered as the most common chromosomal abnormality in live births individuals with incidence worldwide ranging from 1 in 1000 to 1 in 1100 live births [1]. The rate increases in correlation with increased maternal age. There are three types of DS. Nondisjunction is the most common type, and it accounts for 95% of all cases [2].

The two other kinds are Translocation and Mosaic. DS patients are known to have characteristic dysmorphic features including most parts of the body. Some of these features are epicanthic folds, abundant neck skin, broad hands and hypotonia [2]. There are also many congenital abnormalities that are associated with DS patients in different proportions [2]. Congenital heart defects (CHD) are one of the common congenital abnormalities that DS patients might have. It is estimated that about 40%-50% of DS patients have CHD, either isolated or combined lesions [3].

Many types of these CHDs might be seen in DS patients. Some of these common types of CHD that might be observed in DS patients are atrioventricular septal defects (AVSD), ventricular septal defects (VSD), patent ductus arteriosus(PDA), and atrial septal defect(ASD). Since the clinical manifestations, treatment, and prognosis differ depending on the type of CHD and the complicity of the disease, it is better to identify and characterize the types of CHD in each region or area to prepare the appropriate facilities and allocate the resources in dealing with these complications.

Though many studies had been conducted locally and internationally, they revealed different results from various regions and countries. For example, two local studies showed different results. The first study conducted in the western region and included 130 patients with 86.8% of patients had CHD. The result of this study revealed that most common type of CHD that patients had was PDA [4].

Another study conducted in Aseer Central Hospital showed a different result, in which VSD is the most common type in DS [5]. The result of the last study was consistent with the results from other studies in Middle East, Singapore, and China (6, 7). A study in Libya showed that ASD is the most common (8). AVSD was showed as the most common type of CHD in DS in many international studies [3, 9]. After explaining differences in results of many studies, this study is going to determine the common types of CHD in patients admitted to King Abdul-Aziz Medical City, Jeddah and compare the results with other local and international studies.

## **MATERIALS AND METHODS**

This study was conducted in King Abdul-Aziz Medical City (KAMC-JD). It was founded in 1982 in Kingdom of Saudi Arabia, western region-Jeddah. It is a tertiary center concerned with enhancing the individual and public health. It serves patients' needs in all main specialties through outpatient clinics and other departments. It is also associated with King Saud bin Abdul-Aziz University for health sciences which includes the college of medicine and a college of nursing. It also has many specialized centers. One of which is King Faisal cardiac center, which considered one of the main two cardiac centers serving people of the western region (over 5 million population).

In this study, the patients' MRN was used without using their names, since there is no need to uncover such data, and also that is for the sake of the confidentiality. It was approved by the international review board (IRB) committee in King Abdullah International Medical Research Center(KAIMRC). It is a retrospective cross-sectional study. Exclusion and inclusion criteria were applied; all males and females with Down syndrome patients who visited the clinic from January 2000 to January 2014. We excluded Down syndrome patients who did not have echocardiography results. For the sample size, non-probability consecutive sampling technique was used; we included all Down syndrome patients who met the inclusion and exclusion criteria. Also, the International Statistical Classification of Diseases and Related Health Problems (ICD) recording system was used to go back to the old files. And then the inclusion and exclusion criteria were applied to the collected data. So, the variables were used includes the age, the patient gender, and the echocardiography results. After that,

data has been analyzed by using Statistical Package for the Social Sciences (SPSS) program. Frequency and proportion were utilized for the categorical data mentioned above.

The expected outcomes of doing the analysis are summarized into two main points: first, determining the prevalence of congenital heart diseases in Down syndrome patients at King Abdul-Aziz Medical City in Jeddah. The second thing is identifying the most common type of congenital heart diseases in those patients. Finally, the results will be compared to other local and international studies.

## RESULTS

In this study, a total of 191 patients diagnosed with Down syndrome were born or referred to King Abdul-Aziz Medical City between January 2000 and January 2014. Thirteen patients were excluded due to unavailability of their echocardiography results. Of the remaining 178 patients, one hundred and ten patients (62.9%) had congenital heart defects while the echocardiography results were normal in 66 patients (37.1%). Out of those 112 patients, 64 patients (57.1%) were male and 48 patients (42.9%) were female.

Seventy-five patients (67.0%) had isolated CHD, while combined CHD were detected in 37 patients (33.0%). For both sexes, the most common lesion that was detected in both isolated and combined lesion was atrial septal defect (ASD) in 72/112 (64.3%), ventricular septal defect (VSD) in 39/112 (34.8%), patent ductus arteriosus (PDA) in 25/112 (22.3%), and atrioventricular septal defect (AVSD) in 16/112 (14.3%). Tetralogy of Fallot (TOF) in 2/112 (1.8%) and pulmonary stenosis (PS) in 2/112 (1.8%) were the least common as being clarified in Table 1. In male patients, ASD was most common CHD followed by VSD, PDA, AVSD, PS, and TOF. In female patients, ASD was also the most common detected CHD but the second most common lesion was PDA, then VSD, AVSD, and TOF.

TABLE 1.

Congenital Heart Defect	No of cases		
	Total (%)	Boys (%)	Girls (%)
Atrial septal defect	72(64.3)	39(60.9)	32(66.7)
Ventricular septal defect	39(34.8)	25(39.1)	14(29.2)
Patent Ductus Arteriosus	25(22.3)	8(12.5)	17(35.4)
Atrioventricular septal defect	16(14.3)	7(10.9)	9(18.6)
Tetralogy of Fallot	2(1.8)	1(1.6)	1(2.8)
Pulmonary stenosis	2(1.8)	2(3.13)	0
Total	112/178	64/112	48/112

Table 2 provides a summary of all types of CHD detected in our study. The most common isolated CHD in our study was ASD followed by VSD, AVSD, PDA, TOF and PS were mentioned according to their frequency. For the combined CHD, the most common abnormality was ASD with VSD and then followed by ASD with PDA. The combination of ASD with PS was the least common. Generally, Isolated ASD 37/112 (33.0%) was the most common lesion identified in our patients followed by isolated VSD 16/112 (14,3%) then ASD+VSD 14/112 (12.5%), AVSD 10/112 (8.9%) , isolated PDA 9/112 (8.0%), ASD+PDA 9/112 (8.0%), ASD+VSD+PDA 5/112(4.5%), ASD+AVSD 4/112 (3.6%), isolated TOF 2/112 (1.8%), ASD+VSD+AVSD 2/112 (1.8%), VSD+PDA 2/112 (1.8%),

ASD+PS 1/112 (0.9%) and isolated PS 1/112 (0.9%). Details of types of CHD for each male and female patients were summarized in Table 2.

**TABLE 2.** Details of types of CHD for each male and female patients

Congenital Heart Defect	No of patients		
	Total(%)	Male	Female
NORMAL	66(37.1)	36	30
ASD	37(20.8)	23	14
VSD	16(8.9)	13	3
AVSD	10(5.6)	5	5
TOF	2(1.1)	1	1
AVSD+ASD+VSD	2(1.1)	1	1
ASD+AVSD	4(2.2)	1	3
ASD+PDA	9(5.1)	4	5
ASD+VSD	14(7.9)	10	4
VSD+PDA	2(1.1)	1	1
ASD+VSD+PDA	5(2.8)	0	5
PDA	9(5.1)	3	6
PS	1(0.6)	1	0
ASD+PS	1(0.6)	1	0
Total	178	100	78

## DISCUSSION

Down syndrome is the most common chromosomal abnormality universally with estimated incidence of 1 in 1000-1100 [1]. Patients with DS have many congenital diseases. One of the most common congenital diseases that they might have is CHD. The worldwide prevalence of CHD in DS patients ranges from 40% to 50% [3]. It is also considered one of the main causes of death; a study reported that DS patient with CHD have significantly higher mortality rate than DS patient without CHD [10]. AVSD is accepted as the most common type of CHD in DS patients worldwide [3]. Our study showed slightly higher prevalence than the western countries. Also, ASD, unlike global data, was the most common lesion identified in our study.

Four studies were previously conducted in different regions of Kingdom of Saudi Arabia; Aseer, Riyadh, Jeddah, and Al-Madinah. There were variable results regarding the frequency of CHD in DS patients and the most common types of CHD detected in DS patients. There is a study conducted in Aseer city which was published in 2006 [5]. Ninety-eight DS patients were included in their study. It showed that 61.3% of their DS patients had CHD and VSD was the most common type of CHD followed by AVSD and ASD. In 2009, another study was published which was done in Riyadh city [12]. One hundred and ten DS patients were included in their study. Fifty-four (49%) patients had CHD. The most common CHD was VSD followed by ASD.

In Jeddah, there was one study that was published in 2012[4]. One hundred and thirty DS patients were included in their study. They reported that 86.8% of their patients had CHD and PDA was the most common lesion reported in their study. Our explanation for this high percentage is that they included in their study some cardiac diseases that are not considered as part of CHD, e.g. pericardial effusion, pulmonary hypertension, etc. In Al-Madinah region, there was a study published in 2015 [11]. They included 110 DS patients in their study. They reported that 40.9% of their DS patients had CHD and the most common lesion was AVSD. Furthermore, there is a point that needs to be clarified about their results. They mentioned their result according to the most common cardiac presentation in their patients not according to the most common lesion found. In other words, they stated that isolated AVSD was found in 10 patients while it was found as combined lesion in 7 patients, but in the final results, they calculated the isolated VSD alone which affected the real results.

Most of the studies that were done in Kingdom of Saudi Arabia showed different results from other worldwide studies; higher prevalence was found in comparison with the global prevalence and AVSD was not the most common lesion in the local studies. However, AVSD was more common in DS patients than in the healthy population. Our study had larger sample size compared to the other local studies.

One of the limitations of this study is that we included only one center in the study. Including more than one center might give more precise results. Another limitation is that there were 13 DS patients whom we could not find their echocardiography results which might affect the results too.

## CONCLUSION

We did a retrospective cross-sectional study to determine the frequency of CHD in DS patients at King Abdul-Aziz Medical City in Jeddah between January 2000, and January 2014. One hundred and seventy-eight patients were included in the study. Prevalence of CHD in our study was 62.9%. Sixty-six percent of the patients had isolated CHD. The most common lesion found in our patients is ASD.

Our results were different from other international studies; higher prevalence of CHD in DS patients was found in our study, and AVSD, which is the most common CHD in DS internationally, was the fourth in our study. We also had different results of other local studies. However, we had the largest sample size compared to the other local studies. The Limitations of this study should be stated; we only included one cardiac center, and there are few patients whom we could not have their echocardiography results. We recommend to have other studies including multiple centers of the kingdom of Saudi Arabia, and also to use these studies in conducting more research about the management of CHD in DS patients.

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