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Congenital Heart Diseases in Down Syndrome Children at Albala Area, Saudi Arabia

Almawazini AM[1*], Ali A Sharkawy[2], Osama Mohammed Eldadah[3], Yahya Ali Sumaily[1] and Tarig Yassin Alamery[1]

[1]*Department of Pediatrics and Neonatology, King Fahad Hospital at Albaha, Saudi Arabia*

[2]*King Fahad Hospital Albaha, Saudi Arabia*

[3]*King Abdul Aziz Medical City, National Guard Hospital Cardiac Center, Riyadh, Saudi Arabia*

[*]**Corresponding author:** Almawazini AM, Department of Pediatrics and Neonatology, King Fahad Hospital at Albaha, Saudi Arabia, Tel: 00966508294471; E-mail: amawazini@gmail.com

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Abstract

Background: The worldwide incidence of DS in literature varies from 1/600 to 800, and 1/554 in Saudi Arabia. The most common syndrome associated with CHD in children is DS, and 40 to 50% of children with Down syndrome were diagnosed to have CHD, with increased incidence with consanguinity.

Methodology: This is hospital based retrospective cross sectional study, conducted in King Fahad hospital Albaha, Saudi Arabia. All children with Down features delivered in our hospital or referred to us from other centers and aged from 0-12 years were included and screened by echocardiography.

Objectives: The aim of this study was to know the prevalence and the most common types of CHDs in Down syndrome patients at Albala area, Saudi Arabia, and compare it with other studies from Saudi Arabia and international studies.

Results: Total of 150 Down syndrome children were screened in this study, 25 (16.7%) patient revealed normal heart, while125 (83.3%) had CHDs. Female pts 71 (56.8%) and male 54 (43.2%). The consanguinity was confirmed in 81 (54%). The most common lesion was found AVSD 61 (48.8%), followed by PDA 23 (18.4%), VSD 16 (12.8%), PFO 11 (8.8%) then ASD 3 (2.4%), TOF 2 (1.6%), TR 4 (2.4%), MR 2 (1.6%), and PS, AR, Epstein anomaly less than 1% for each. The combination of PDA with PFO was found as the most common combination of cardiac lesions in Down syndrome children.

Conclusion: The frequency of CHDs in Down syndrome children in Albaha area was higher than other rejoins in Saudi Arabia and reported in studies worldwide. The prevalence of CHDs was slightly higher in children born to consanguineous parents. Atrioventricular septal defect was found as the most common defect.

Keywords: Down syndrome; Congenital heart diseases

Abbreviations:

DS: Down Syndrome; CHD: Congenital Heart Disease; KFH: King Fahad Hospital; NICU: Neonates Intensive Care Unit; PFO: Patent Foramen Ovale; PDA: Patent Ductus Arteriosus; VSD: Ventricular Septal Defect; PS: Pulmonic Stenosis; PA: Pulmonary Atresia; AVSD: Atrioventricular Septal Defect; PHN: Pulmonary Hypertension; TOF: Tetralogy of Fallot; ASD: Atrial Septal Defect; PFO: Patent Foramen Ovale; TR: Tricuspid Atresia; MR: Mitral Regurgitation; LV: Left Ventricle; RV: Right Ventricle; PA: Pulmonary Atresia; HLHS: Hypoplastic Left Heart Syndrome; TGA: Transposition of Great Arteries

Introduction

Down syndrome was described in 1866 by John Langdon Down. The incidence of DS in world literature varies from 1/600 to 800 [1] and 1/554 of live birth in Saudi Arabia [2]. Down syndrome is the common chromosomal anomaly which cause mental retardation in children [1,3]. Clinically it is diagnosed by the presence of a characteristic features including large protruding tongue, up slanting palpebral fissures, , short neck, skin fold, brush field spots of iris, short hands with simian creases, hypotonia, and gap between first and second toes. Clinical manifestations cannot specify the type DS, therefore the chromosomal study is required [4]. There are three karyotype types of Down syndrome; Trisomy 21 which represents in (95%) of cases, Translocation Down syndrome, and Mosaicism [5]. Down syndrome can be associated with other congenital defects problems. Prevalence of Congenital Heart Diseases (CHD) increased in the last 15 years, it was reported in Europe 8.2 per 1000 live birth [6]. The prevalence of CHD is about 40-50% in literatures (between 30-65%) in Down syndrome children [7,8], while it is between 40.9-86.8% in Saudi Arabia [9-12]. Atrio-ventricular septal defect (AVSD) is the most common cardiac anomalies associated with DS 54%, followed by ventricular septal defect (VSD), patent ductus arteriosus, and atrial septal defect (ASD) [7]. Pulmonary hypertension (PHN) due to volume overload is the most common complication [3], and also been found in DS patients with normal heart [13]. Echocardiography is easy non-invasive procedure to diagnose CHD

and to measure the pulmonary artery pressure. Early diagnosis and management is important to improve quality of life in DS patients [14,15].

Objective

The aim of the study is to determine the prevalence and the most common types of CHDs in Down syndrome patients at Albala area, Saudi Arabia, and to compare it with other previously reported studies from Saudi Arabia and international studies.

Methods

This is a hospital based, retrospective cross sectional study, was conducted in the Cardiology Unit, Pediatric and Neonatology Department, King Fahad Hospital Albaha, Kingdom of Saudi Arabia, from Jan 2010 to Jan 2016. It was approved by the research and ethical committee in the hospital. All patients aged between 0 to 12 years old with down syndrome features was included in our study. All cases were evaluated by full clinical examination looking for features of Down syndrome. The diagnosis was confirmed by chromosomal study. The age, sex, consanguinity, and family history of a previous Down syndrome child or other anomaly. Chest X-ray, electrocardiogram, and echocardiography were done for all patients. The diagnosis of CHD was confirmed by echocardiography (Philips IE33 echocardiography machine, Philips, Bothel, WA, USA), with S 10-12 and S 3-8 chest wall transducer. For sedation, oral Chloral Hydrate 50 mg/kg body weight was used, and in some cases IV sedation was needed. Echocardiogram included M-mode, two dimensional, pulsed wave, continuous wave, and color Doppler was done for all patients. Ejection fraction and fractional shortening of left ventricle were calculated. Standard transthoracic views; an apical 4-chamber, parasternal long and short axes were obtained to evaluate the systolic and diastolic LV function. The evaluation of Pulmonary hypertension was classified based on the recommendations of American College of Cardiology and American Heart Association 2009 meeting, as mild: 30-40 mm Hg, moderate: 40-60 mm Hg, and severe ≥60 mm Hg. The mean pressure of the pulmonary artery was considered normal when it was ≤25 mm Hg [16].

Results

As seen in **Table** 1, total of 150 Down syndrome children were screened by echocardiography for congenital heart anomalies. 125 (83.3%) pts were diagnosed to have congenital heart diseases (CHDs) while 25 (16.7%) pts had normal heart. Consanguinity were documented in 67 (53.6%) cases, and no consanguinity were documented in 58 (46.4%).

Patients distribution	Total	male	female	Consanguinity	No consanguinity	PHN	No PHN
All DS patients seen in the study	150 (100%)	66 (44%)	84(56%)	81 (54%)	69 (44%)	20 (13.3%)	130(86.6%)
DS With CHD	125 (83.3%)	54 (43.2%)	71(56.8%)	67 (53.6%)	58 (46.4%)	18 (14.4%)	107(85.6%)
DS with normal heart	25 (16.7%)	9 (36%)	16 (64%)	14 (56%)	11 (44%)	2 (8%)	23 (92%)
DS: Down Syndrome, PHN: Pulmonary Hypertension, CHD: Congenital Heart Disease.							

Table 1: Distribution of all DS patients screened in this study.

Among the 25 cases of DS children without CHDs, consanguinity was documented in 14 (56%) pts while no consanguinity was seen in 11 (44%) pts. Regarding consanguinity no significant difference was seen between both groups (p-value 0.04). Patients age was between one day and twelve, mean age 2.7 yr. In the DS pts had cardiac defects females were 71(56.8%) and males were 54 (43.2%). Full clinical examination was done for all patients and detailed history about any gestational disease was checked. The upper airway examination was performed on 28 (22.4%) patients older than five years to detect the upper airway obstruction.

Types of CHD	pts number	%	Male		Female	
With CHD	**125**	**100%**	**54**	**43.2%**	**71**	**56.8%**
AVSD	61	48.8%	26		35	
PDA	23	18.4%	7		16	
VSD	16	12.8%	7		9	
ASD	14	11.2%	9		5	
TR	4	3.2%	2		2	
TOF	2	1.6%	2			
MR	2	1.6%			2	

AR	1	<1%	1			
PS	1	<1%			1	
EBETEIN	1	<1%			1	
CHD with PHN	18/125	14.4%				
Palliative surgery as 1st step	3/125					
Operated as univentricle	5/125					
Pericardial effusion	2/125					

DS: Down Syndrome; CHD: Congenital Heart Disease; KFH: King Fahad Hospital; NICU: Neonates Intensive Care Unit; PFO: Patent Foramen Ovale; PDA: Patent Ductus Arteriosus; VSD: Ventricular Septal Defect; PS: Pulmonic Stenosis; PA: Pulmonary Atresia; AVSD: Atrioventricular Septal Defect; PHN: Pulmonary Hypertension; TOF: Tetralogy Of Fallot; ASD: Atrial Septal Defect; PFO: Patent Foramen Ovale; TR: Tricuspid Atresia; MR: Mitral Regurgitation; LV: Left Ventricle; RV: Right Ventricle; PA: Pulmonary Atresia.

Table 2: Frequency and distribution of congenital heart diseases in down syndrome patients no. 125 pts.

Electrocardiogram and echocardiogram were done for all patients. Chromosome analysis was done in 57 pts (38%), where 56 (98.2%) pts were diagnosed as trisomy 21, one case was diagnosed as translocation Down syndrome. As seen in Table 2, AVSD was the most frequent lesion 61 (48.8%), followed by PDA 23 (18.4%), VSD 16 (12.8%), PFO 11 (8.8%) then ASD 3 (2.4%), TOF 2 (1.6%), TR 4 (2.4%), MR 2 (1.6%), and PS, AR, Epstein anomaly less than 1% for each.

The most common combination of cardiac lesions in Down syndrome cases included in this study was PDA with PFO. Only 5 patients had been seen with large PDA who needed transfer for PDA ligation. Female patients were more affected in all types of CHD. Diagnosed patient with CHD needed surgical intervention were transferred to higher cardiac center in Riyadh and Jeddah.

Types of CHD	Our study n=125	Almadina [12]	Aseer [13] n=57	Riyadh [10] n=54	Jeddah [11] N=92
percent	83.3%	40.9%	61.3%	49%	86.8%
AVSD	48.8%	33.3%	22.6%	15%	12%
PDA	18.4%	8.8%	14%	7%	47.8%
VSD	12.8%	22.2%	33.3%	43%	29%
ASD	11.2%	13.3%	21.1%	25%	41%
TR	3.2%		3.2%		33.7%
TOF	1.6%	2.2%	5.3%	4%	1.5%
MR	1.6%				
AR	1%				
PS	1%		1.8%	1.9%	1.5%
EBETEIN	1%				
CHD with PHN	14.4%				9.7%

CHD: Congenital Heart Disease; KFH: King Fahad Hospital; NICU: Neonates Intensive Care Unit; PFO: Patent Foramen Ovale; PDA: Patent Ductus Arteriosus; VSD: Ventricular Septal Defect; PS: Pulmonic Stenosis; PA: Pulmonary Atresia; AVSD: Atrioventricular Septal Defect; PHN: Pulmonary Hypertension; TOF: Tetralogy of Fallot; ASD: Atrial Septal Defect; PFO: Patent Foramen Ovale; TR: Tricuspid Atresia; MR: Mitral Regurgitation; LV: Left Ventricle; RV: Right Ventricle; PA: Pulmonary Atresia.

Table 3: Distribution of CHD in Down syndrome pts in some regions in Saudi Arabia.

Palliative surgery as first step done for 3 (2.4%) and later complete correction done. Univentricular repair was needed in 5 (4%) due to unbalanced AVSD. Pulmonary hypertension developed in 18 (14.4%) cases, and 3 (2.4%) was inoperable due to severe pulmonary hypertension. Faintness was developed in five patients 5 (4%), chest pain in 3 (2.4%) and occasional palpitation in 2 (1.6%). Upper airway obstruction was diagnosed in 28 pts older than 5 years due to large tonsils or adenoids. Hypothyroidism was diagnosed in 9 (6.3%) pts. PHN in DS children with CHD was seen in 18 (14.4%) cases, which was more than DS patients without CHD, the odds ratio (OR: 1.9) (CI 95% 2.77-8.17) p=0.001.

Discussion

Probably related with the high altitude of Albaha, in our study we found that 83.3% of the Down patients had associated CHD, which was similar to other areas in Saudi Arabia, 83.3% in Jeddah [10], 61.3% in Aseer [12], 49% in Riyadh [9], and 40.9% in Almadina [9]. But It was higher than the occurrence reported in the international studies from 19 to 43% [17], 78% in Turkey [18], 60% in Oman [19], 43.5% in Germany [7], 40% in Egypt [1], and between 30-65% in Netherlands [6]. AVSD was found as the highest prevalence 61 (48.8%) pts, VSD 16 (12.8%) pts, and ASD 14 (11.2%) pts. in third (Table 2). Our results were going with the international studies in which AVSD was varies from 40-80% [17] and VSD 33.3% [20]. As seen in Table 3 in Saudi Arabia, like our study, the AVSD was the most common CHD type diagnosed in Almadina [11], VSD the most common was diagnosed in Aseer [12] and Riyadh [9], while PDA was the most common in Jeddah [10].

We found that patients with DS and CHD have a higher risk of developing PHN 14.4% compared with DS patients without CHD 2%. The prevalence of PHN in neonates with DS was 1.2 to 5.2%, while the prevalence in the general population is 0.1% [7]. The patients with DS develop PHN even in the absence of CHDs. Upper airway obstruction is a common associated problem with DS and may cause obstructive sleep apnea and can develop pulmonary hypertension [13]. In our study 28 pts (22.4%) were diagnosed as upper airway obstruction, due to tonsils enlargement in 21 pts 75%, and tonsils and adenoids enlargement in 7 pts (25%), similar to the international studies [21]. Our patients were evaluated at high altitude area about 2400 meter above sea level, with lower partial pressure of oxygen which could increase the incidence of PHN.

Limitations

The patients require a long time of follow-up to evaluate their outcomes and determine complications. Furthermore, conducting a chromosomal study for our patients and their parents was very difficult. The transfer of patients to more advanced cardiac centers was also delayed because of the limited availability of beds in these centers.

Conclusion

Compared with the general population, DS patients have a high prevalence of CHD and PHN. The risk of PHN increases significantly when congenital heart defects are present. AVSD is the most common type of CHD associated with DS patients. The pathology of PHN in DS patients without CHD needs to be explained. Echocardiography should be considered as a non invasive tool for the evaluation and screening of all DS patients. Advanced cardiac and genetics centers in Southwest Saudi Arabia are urgently needed to improve the outcomes.

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