

Frequency of Associated Congenital Heart Defects in Down Syndrome

SYED NAJAM HYDER¹, LUBNA HUMAYUN², ALI HASAN³

¹Associate Professor of Pediatric cardiology The Children Hospital and Institute of Child Health, Lahore.

²Assistant Professor Department of Hematology and Pathology, University of Lahore.

³Clinical observer

Correspondence to Dr. SN Hyder, Email: dmajamhyder@gmail.com. Cell: 92333-4262250.

ABSTRACT

Background: Children with Downs seem commonly with defects in their heart.

Aim: To find out the occurrence of congenital heart disease in children with Down.

Methods: This retrospective study planned at the cardiology unit in the children's hospital, Lahore, in 2017. One hundred and seven babies with Down enrolled in this study until 14 years of age.

Results: Heart diseases seem in 63 patients from 107 that revealed 59% with male to female ratio of 1.7:1. Among the isolated single lesions ventricular septal defect found in 60.3% patent ductus arteriosus in 13.7%, complete atrioventricular defects in 8.6%, followed by ASD in 6.8%, Pulmonary atresia with VSD in 6.8% ,Tetralogy of Fallot and TGA in 1.7% of patients. Among the mixed lesion (4.7%) VSD+ASD found in two patients while COA+ PDA, Univentricular heart with TGA and PA+VSD +DORV found in each patients.

Conclusion: Heart lesion in down babies seen to be 59%. In non-cyanotic lesion, ventricular septal defect was common, while in category of cyanotic heart lesion the pulmonary atresia and VSD was found and in mix heart lesion VSD with ASD was found.

Keywords: Down syndrome, Transposition of great arteries, Pulmonary atresia, Tetralogy of Fallot's,

INTRODUCTION

Down syndrome is the chromosomal disorder produced by the additional copy of chromosome 21^{1,2,3,4}. It commonly effects the IQ and is associated with mental retardation¹. In developed world the frequency differs from 1 to 732 live birth^{1,5}. Heart abnormalities related with down's comprise mostly in case of acyanotic lesions are atrioventricular septal defect (AVSD), ventricular septal defect (VSD), patent ductus arteriosus (PDA), and in cyanotic lesion are tetralogy Of Fallot's (TOF) transposition of great arteries (TGA), double outlet right ventricle (DORV), pulmonary atresia /stenosis and univentricular heart⁶.

Heart abnormalities are the major factors in mortality in Down syndrome. Some literature showed that 40%-60% patients with Down syndrome have cardiac anomalies⁷. Cardiac associated abnormalities in Down syndrome varies in incidences from one country to other. In USA and Europe regarding types of heart anomalies atrioventricular septal defect found the most common congenital heart diseases. Whereas Ventricular septal defect found to be more common in Asian Countries, and atrial septal defect in Latin America and Korea^{6,8,9}.

Therefore, this study was piloted to find the occurrence of various types of congenital heart anomalies in children with Down syndrome at our set up with confirmation of karyotyping.

MATERIAL AND METHODS

A institution-based study was done in the cardiology unit of the children hospital Lahore. 107 babies with Down enrolled in this study until 14 years of age. All patients with Down syndrome came for echocardiography on the basis of the phenotypic appearance were enrolled from January to December 2017 irrespective of any sign of diseases.^{2-D}

Received on 25-05-2019

Accepted on 18-08-2019

echocardiography done by proper consultant cardiologist along with routine chest x-ray, ECG and karyotyping.

Exclusion criteria was:

- PFO.
 - PDA with premature baby below 4 week of life
 - And multiple CHD placed wide with dominant defect
- Data was analysed by SSPS 22 and frequency was applied.

RESULTS

Heart diseases seem in 63 patients from 107 that revealed 63(59%) with male to female ratio of 1.7:1. Patients age were grouped into four categories. Group-1 involved upto 2 months of age, while group-2 comprised from two months to 1 years of age, group-3 involved one to five years of age and group-4 above five years of age. Majority of patients belong to below five years (Table-1).

Mostly the heart anomalies were found below two months of age that is 80%, between three months to one year 65% patients had congenital heart defects, below five years 57% patients had CHD while above five years 22% patients found to had CHDs. Heart anomalies were divided into cyanotic, non-cyanotic and further into isolated and mixed lesions.

In the isolated group ventricular septal defect (VSD) found to be the common heart lesion i.e. 60.3%, then PDA i.e., 13.7% and complete atrioventricular septal defect (CAVSD) (8.6%). In case of cyanotic cardiac lesion common lesion was Pulmonary atresia with VSD i.e. 5.17% then TOF 3.4% followed by TGA 1.7%. (Table-2)

In case of mixed lesions VSD and ASD found in 1.8%, Coarctation and PDA, DORV and pulmonary atresia and univentricle with TGA were found 0.9% each (Table-3).

Regarding Karyotyping findings 47,XY+21 found in 64 patients i.e. 59.8%, 38 patients were 47,XX+21, one patient with 46,XXt (14q:21q), three patients had 46,XYt (14q:21q) and one patient has 46,XYt (13q:21q) (Table-4).

Table 1: Age breakdown of Study population (n=107)

Age	Frequency	%age
0-2 mon	7	6.5
3 mon- 1 year	40	37.2
1 year -5 years	48	44.8
≥5 years	11	10.2

Table 2: Frequency distribution in individual lesion Isolated cardiac lesions (n=105)

Acyanotic lesions (n=3)	%age	Cyanotic lesions (n=2)	%age
VSD	60.5	PA+ VSD	5.17
PDA	13.7	TOF	3.4
CAVSD	8.6	TGA	1.7
ASD	6.0		

Table 3: Frequency distribution in mixed lesion

Acyanotic lesions (n=3)	%age	Cyanotic lesions (n=2)	%age
VSD+ ASD	1.8	PA+VSD+DORV	0.9
PDA+CoA	0.9	Univentricle +TGA	0.9

Table-4 . Karyotypes (n=107)

	Frequency	Percentage
47,XY+21	64	59.8
47,XX+21	38	35.5
46,XXt (14q:21q)	1	0.93
46,XYt (14q:21q)	3	2.8
46,XYt (13q:21q)	1	0.93

DISCUSSION

The occurrence of heart disease in Down's syndrome is well recognised. It varies from 35% to 65%^{1,10}. The presence of heart anomalies in our study was 59% which is close to Korea⁶ and Khyber Pakhtunkhwa study¹. One study from Brazil showed higher frequency heart defects i.e., 81%¹². Some study showed lower frequencies as study from Libya and Netherlands i.e., 45.10%³ and 43%¹³. This difference at various geographic part may be due to different genetic make-up of nation and embryological mechanisms¹.

In our study out of 107 patients with Down the male to female ratio was 1.7:1. Same ratio was also found in Khyber Pakhtunkhwan province.¹ While in Brazil where male to female ratio found to be about 1:1.3¹².

The common type of heart anomalies in our study was VSD i.e., 35(60.5%). It is comparable to Peshawar¹ and the Libyan and Guatemala population^{3,13}. Similar result found in Mexico^{3,7} Korea.⁶ and study from Turkey^{3,14}. Multiple cardiac defects found ie 4.6% in our study.

In acyanotic category isolated and mixed lesions the common heart anomalies was ventricular Septal defect (VSD), followed by atrial septal defect with ventricular septal defect (ASD+VSD) in 2(1.8%) and coarctation of aorta and patent ductus arteriosus (CoA + PDA) in 0.8%. In case of the cyanotic cardiac anomalies pulmonary atresia with VSD was more common ie 5.17% followed by tetralogy of fallots (TOF) and in case of mixed lesions univentricle heart with atrial septal defect was found ie 0.9%. This is supported by the Sudanian study¹⁵. The results were quite comparable to Indian study also¹⁶.

Similarly regarding karyotyping findings 95.3% patients had non-dysjunction, 3.7% patients had translocation at 14q:21q and 0.93% had translocation at 13q:21q.

Drawback: One centre does not reflect the population base incidence.

CONCLUSION

The heart anomalies found in 63% children with Down. In case of acyanotic heart lesion ventricular septal defect while in cyanotic cardiac lesion pulmonary atresia and in mix type VSD and ASD were found common in our center.

IBR approval: Approval granted from The Children Hospital, Lahore.

Conflicts of interest: Nil

Funding: None

Conflict of interest: No conflict.

REFERENCES

- Khan, I. and Muhammad, T. (2012). Frequency and pattern of congenital heart defects in children with down's syndrome. *Gomal. J. Med. Sci.*, **10**(2): 241-3.
- Kolgeci, S., Kolgeci, J., Azemi, M., Shala-Begiraj, R., Gashi, Z. and Sopjani, M. (2013). Cytogenetic Study in Children with Down syndrome among Kosova Albanian Population Between 2000 and 2010. *Mat. Soc. Med.*, **25**(2): 131-135.
- Elmarpy, Z., Rayani, A., Shah, A., Habas, E. and Aburawi, E. (2008). Down syndrome and congenital heart disease: why the regional difference as observed in the Libyan experience? *Cardiovasc. J. Afr.*, **22**: 306-309
- Aburawi, E.H. The burden of congenital heart disease in Libya. (2006) *Libyan. J. Med.*, **1**: 120-122.
- Sherman, S.L., Allen, E.G., Bean, L.H. and Freeman, S.B. (2007). Epidemiology of down syndrome. *Ment. Retard. Dev. Disabil. Res. Rev.*, **13**(3): 221-7.
- Kim, M.A., Lee, Y.S., Yee, N.H., Choi, J.S., Choi, J.Y. and Seol, K. (2014). Prevalence of Congenital Heart Defects Associated with Down syndrome in Korea. *J. Korean. Med. Sci.*, **29**: 1544-1549.
- Figuroa, J.R., Magaña, B.P., Hach, J.L.P., Jiménez, C.C. and Urbina, R.C. (2003). Heart Malformations in Children with Down syndrome. *Rev. Esp. Cardiol.*, **56**(9): 894-9.
- Hamerton, J.L., Briggs, S.M., Giannelli, F. and Carter, C.O. (1961). Chromosome studies in detection of parents with high risk of second child with Down's syndrome. *Lancet*, **2**: 788-91.
- Castilla, E.E., Rittler, M., Dutra, M.G., Lopez-Camelo, J.S., Campaña, H., Paz, J.E. and Orioli, I.M. (1998). Survival of children with Down syndrome in South America. ECLAMC-Downsurv Group. Latin American Collaborative Study of Congenital Malformations. *Am. J. Med. Genet.*, **79**: 108-11.
- Bower, C. and Ramsay, J.M. (1994). Congenital heart disease: a 10 year cohort. *J. Pediatr. Child. Health.*, **30**: 414.
- Abbag FI. Congenital heart diseases and other major anomalies in patients with Down's syndrome. *Saudi Med J* 2006; **27**: 219-22.
- Mourato, F.A., Villachan, L.R.R. and Mattos, S.S. (2014). Prevalence and profile of congenital heart disease and pulmonary hypertension in Down syndrome in a pediatric cardiology service. *Rev. Paul. Pediatr.*, **32**(2): 159-63.
- Vida, V.L., Barnoya, J., Larrazabal, L.A., Gaitan, G., Garcia, F.M. and Castaneda, A.R. (2005) Congenital cardiac disease in children with Down's syndrome in Guatemala. *Cardiol. Young.*, **15**: 286-290.
- Nisli, K., Oner, N., Candan, S., Kayserili, H et al. (2008). Congenital heart disease in children with Down's syndrome: Turkish experience of 13 years. *Acta. Cardiol.*, **63**: 585-589.
- ALI, S.K. (2009). Cardiac abnormalities of Sudanese patients with Down's syndrome and their short-term outcome. *Cardiovasc. J. Afr.*, **20**: 112-115.
- Sharma, B.M., Khera, M.S., Sondhi, L.C.V. and Devgan, C.A. (2013). A study to determine the prevalence of pulmonary arterial hypertension in children with Down syndrome and congenital heart disease. *Medical Journal Armed Forces India*, **69**: 241-245.

