Frequency of Associated Congenital Heart Defects in Down Syndrome

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ABSTRACT

Background: Children with Down's 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 seen 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 patient.
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. It commonly effects the IQ and is associated with mental retardation. In developed world the frequency differs from 1 to 732 live birth. 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 heart.

Heart abnormalities are the major factors in mortality in Down syndrome. Some literature showed that 40%-60% patients with Down syndrome have cardiac anomalies. 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.

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.

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 bellow five years (Table-1).

Mostly the heart anomalies were found bellow two months of age that is 80%, between three months to one year 65% patients had congenital heart defects, bellow 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 furthur into isolated and mixed lesions.

In the isolated group ventricular septal defect (VSD) found to the common heart lesion ie 60.3%, then PDA i.e., 13.7% and complete atrioventricular septal defect (CVA) (8.6%). In case of cyanotic cardiac lesion common lesion was Pulmonary atresia with VSD ie 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). Reguarding Karotyping findings 47,XY+21 found in 64 patients ,47,XX+21, one patients with 46,XXt (14q;21q), three patients had 46,XOt (14q;21q) and one patients has 46,XYt (13q;21q) (Table-4).
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


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

<table>
<thead>
<tr>
<th>Age</th>
<th>Frequency</th>
<th>%age</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2 mon</td>
<td>7</td>
<td>6.5</td>
</tr>
<tr>
<td>3 mon-1 year</td>
<td>40</td>
<td>37.2</td>
</tr>
<tr>
<td>1 year-5 years</td>
<td>48</td>
<td>44.8</td>
</tr>
<tr>
<td>≥5 years</td>
<td>11</td>
<td>10.2</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Acyanotic lesions (n=3)</th>
<th>%age</th>
<th>Cytanic lesions (n=2)</th>
<th>%age</th>
</tr>
</thead>
<tbody>
<tr>
<td>VSD</td>
<td>60.5</td>
<td>PA+ VSD</td>
<td>5.17</td>
</tr>
<tr>
<td>PDA</td>
<td>13.7</td>
<td>TOF</td>
<td>3.4</td>
</tr>
<tr>
<td>CAVSD</td>
<td>8.6</td>
<td>TGA</td>
<td>1.7</td>
</tr>
<tr>
<td>ASD</td>
<td>6.6%</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 3: Frequency distribution in mixed lesion

<table>
<thead>
<tr>
<th>Acyanotic lesions (n=3)</th>
<th>%age</th>
<th>Cytanic lesions (n=2)</th>
<th>%age</th>
</tr>
</thead>
<tbody>
<tr>
<td>VSD+ ASD</td>
<td>1.2</td>
<td>PA+VSD+DORV</td>
<td>0.9</td>
</tr>
<tr>
<td>PDA+CoA</td>
<td>0.9</td>
<td>Univentricle + TGA</td>
<td>0.9</td>
</tr>
</tbody>
</table>

Table 4 . Karyotypes (n=107)

<table>
<thead>
<tr>
<th>Karyotypes</th>
<th>Frequency</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>47,XY+21</td>
<td>64</td>
<td>59.8</td>
</tr>
<tr>
<td>47,XX+21</td>
<td>38</td>
<td>35.5</td>
</tr>
<tr>
<td>46,XXI (14q;21q)</td>
<td>1</td>
<td>0.93</td>
</tr>
<tr>
<td>46,XYI (14q;21q)</td>
<td>3</td>
<td>2.8</td>
</tr>
<tr>
<td>46,XYI (14q;21q)</td>
<td>1</td>
<td>0.93</td>
</tr>
</tbody>
</table>

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 Koreak and Khyber Pakhtunkhwa study.1 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%6 and 43%.13. This difference at various geographic part may be due to different genetic make-up of nation and embryological mechanisms.1

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 1:1:13,12.

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.13,14. Similar result found in Mexico3-5 Korea,6 and study from Turkey14. Multiple cardiac defects found is 4.6% in our study.

In acyanotic catagory 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 i.e 5.17% followed by tetrology of fallots (TOF) and in case of mixed lesions univentricle heart with atrial septal defect was found is 0.9%. This is supported by the Sudanian study15. The results were quite comparable to Indian study also16.

Similarly regarding karotyping 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.