STUDY OF INCIDENCE OF CONGENITAL HEART DISEASES IN CHILDREN OF AGE GROUP 1 MONTH TO 12 YRS
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ABSTRACT: BACKGROUND: This study was designed to determine the reasons for the variability of the incidence of congenital heart disease (CHD). The relative frequency of different major forms of CHD also differs greatly from study to study. METHODS: Cross sectional study carried over a period of 2 months in the patients who were admitted in department of paediatrics, GGH, Guntur. A total of 50 patients were included in the study. RESULTS: most common congenital heart disease is ventricular septal defect. And the common cyanotic lesion is tetralogy of fallot. CONCLUSION: in most of congenital heart diseases definitive diagnosis can be made by echocardiography.

INTRODUCTION: Congenital heart disease is the general term used to describe abnormalities of heart or great vessels that are present from birth. Disorders that arise from faulty embryogenesis during gestational weeks 3 to 8, when major cardiovascular structures begin to function. The cause of congenital heart disease may be either genetic or environmental. The reported incidence of CHD IS 8-10/1000 live births.¹²

GENETICS: The main cause of congenital heart disease is sporadic genetic abnormalities which include mutation, small chromosomal deletions, addition or deletion of an entire chromosome.³ GATA 4, TBX 5 and NKX 2-5 are the transcription factors mutated in some patients with atrial and ventricular septal defects Mutations in α-Myosin heavy chain causes atrial septal defect. Deletion of a small chromosome gene TBX1 causes tetralogy of Fallot. Other important genetic causes include chromosomal aneuploidies particularly Turner syndrome and trisomy's of13, 18 and 22. Mutations in JAGGED 1, NOTCH 1 and 2 lead to bicuspid and aortic valve defects and tetralogy of Fallot.

ENVIRONMENT: Antenatal environmental factors include maternal infections (Rubella), drugs (Alcohol, lithium, hydantoin and thalidomide) and maternal illness (Diabetes, phenylketonuria and systemic lupus erythematosus).⁴

The varied structural abnormalities in congenital heart diseases fall primarily into three major categories
  - Malformations causing a left to right shunt.
  - Malformations causing right to left shunt.
  - Malformations causing an obstruction.

LEFT TO RIGHT SHUNTS: The most commonly encountered left to right shunts include;
  1. Atrial septal defect.
  2. Ventricular septal defect.
4. Atrioventricular septal defects.

**RIGHT TO LEFT SHUNTS:** These are the cyanotic group of diseases. Here are the commonly encountered right to left shunts.
1. Tetralogy of Fallot.
2. Transposition of great arteries.
3. Persistent truncus arteriosus.
4. Tricuspid atresia.

**OBSTRUCTIVE CONGENITAL ABNORMALITIES:** Congenital abnormalities to blood flow may occur at the level of heart valves or within a great vessel. The common examples are;
1. Coarctation of aorta.
2. Pulmonary stenosis and atresia.
3. Aortic stenosis and atresia.

Now, the echocardiography, based on the principle of ultrasound has emerged as a very useful non-invasive diagnostic tool in a wide variety of congenital and acquired cardiac disorders. It has advanced from the uni-dimensional M-mode technique of early 1970s to the two dimensional technique of late 1970. Precise anatomic diagnosis has become a reality with the advent of echocardiography with improved technology and added color flow and Doppler techniques. Identification of cardiac anatomy and associated pathology has become almost as diagnostic as cardiac catheterisation. Two-dimensional and M-mode electrocardiography help in assessing systolic ventricular function.

**AIMS & OBJECTIVES:**
1. To study the prevalence among 1 month to 12 years of age.
2. To study various presentations of congenital heart diseases.
3. To study the age and sex distribution of congenital heart diseases.
4. To know the incidence of congenital heart disease.
5. To study the effects of consanguinity on congenital heart diseases.

**MATERIALS AND METHODS:** The present clinical study was conducted in Guntur Medical College Government General Hospital, Guntur from May 2013 to June 2013. It is a cross-sectional study. All the children from 1 month to 12 years of age group with risk factors like consanguinity associated with congenital abnormalities and other genetic factors were included. The history of familial predisposition of heart diseases, environmental and maternal factors including diabetes mellitus, SLE, maternal ingestion of drugs like lithium, ethanol, warfarin, thalidomide, anti-metabolites, anti-convulsants etc was taken. Our study excludes all neonates i.e. age group up to 30 days, documented hydrops foetalis (or) sustained arrhythmias.
RESULTS:

<table>
<thead>
<tr>
<th>Age</th>
<th>No. of Patients</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;1 yr</td>
<td>18</td>
<td>36%</td>
</tr>
<tr>
<td>1-4 yrs</td>
<td>6</td>
<td>12%</td>
</tr>
<tr>
<td>4-8 yrs</td>
<td>10</td>
<td>20%</td>
</tr>
<tr>
<td>8-12 yrs</td>
<td>16</td>
<td>32%</td>
</tr>
</tbody>
</table>

Graph 1: Most of the patients were below 1 yr which constitutes 36% followed by frequency in 8 – 12 yrs age group.

Graph 2: From the above study, it is seen that male patients predominated over females. 64% were males and 36% were females.
Graph 3: Most of the patients (76%) were suffering from acyanotic heart diseases and the rest (24%) were suffering from cyanotic heart diseases.

<table>
<thead>
<tr>
<th>Type of Heart Disease</th>
<th>No. of patients</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cyanotic</td>
<td>12</td>
<td>24%</td>
</tr>
<tr>
<td>Acyanotic</td>
<td>38</td>
<td>76%</td>
</tr>
</tbody>
</table>

Graph 4: In our study, there are about 32% of cases with Ventricular Septal Defect being the most common disorder among congenital heart diseases.

<table>
<thead>
<tr>
<th>Various types of Congenital Heart Diseases</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>VSD</td>
<td>16</td>
<td>32%</td>
</tr>
<tr>
<td>ASD</td>
<td>8</td>
<td>16%</td>
</tr>
<tr>
<td>PDA</td>
<td>5</td>
<td>10%</td>
</tr>
<tr>
<td>TOF</td>
<td>9</td>
<td>18%</td>
</tr>
<tr>
<td>TGA</td>
<td>3</td>
<td>6%</td>
</tr>
<tr>
<td>Multiple Lesions</td>
<td>4</td>
<td>8%</td>
</tr>
</tbody>
</table>
Presenting with murmur  |  Number  |  Percentage  
---|---|---
Positive  | 37  | 74%  
Negative  | 13  | 26%  

Data based on presence of murmurs

**Graph 5:** About 74% of them were present with murmurs while 26% did not show any type of murmur.

| Maternal Age  |  Number  |  Percentage  
---|---|---
< 20 yrs  | 11  | 22%  
20 – 30 yrs  | 33  | 66%  
>30 yrs  | 6  | 12%  

Data based on maternal age

**Graph 6:** Majority of mothers of patients belonged to 20-30 yrs of age group i.e 66%.
### Study of consanguineous history

<table>
<thead>
<tr>
<th>History of Consanguinity</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive</td>
<td>4</td>
<td>8%</td>
</tr>
<tr>
<td>Negative</td>
<td>46</td>
<td>92%</td>
</tr>
</tbody>
</table>

**Graph 7**: This shows that about 92% of them did not have history of consanguinity.

### Presence of extracardiac congenital anomalies

<table>
<thead>
<tr>
<th>Extracardiac congenital anomalies</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive</td>
<td>5</td>
<td>10%</td>
</tr>
<tr>
<td>Negative</td>
<td>45</td>
<td>90%</td>
</tr>
</tbody>
</table>

**Graph 8**: Down’s syndrome is seen predominantly among 10% of people with other congenital anomalies.
<table>
<thead>
<tr>
<th>History of maternal illness</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive</td>
<td>3</td>
<td>6%</td>
</tr>
<tr>
<td>Negative</td>
<td>47</td>
<td>94%</td>
</tr>
</tbody>
</table>

Data based on history of maternal illness in the first trimester of pregnancy

Graph 9: About 6% of mothers have shown history of maternal illness in our present study.

DISCUSSION: The patients were classified into 4 classes based on their age. Less than 1 yr – 4 yrs, 4 – 8 yrs, 8 – 12 yrs. The majority of patients belonged to <1yr age group i.e about 36% followed by age group of 8 – 12 yrs with 32%. There was less number of patients suffering from congenital heart disease between 1 – 4 yrs age group.

Sex of Patients: Among our study population, male patients predominated over females i.e. there were 64% male children while only 36% are female. In a study conducted by Daljit Singh et.al, the prevalence of male to female ratio of congenital heart disease in India has been reported as 1.1: 1 and 1.25: 1.6. Here our result is 1.7: 1, which coincides to the research value conducted by Daljit Singh et.al

Type of congenital heart Disease: In the present study there are 24% patients of cyanotic type of which tetralogy of fallot Is most commonest followed by TGA with 6% and 76% constituted acyanotic group of which ventricular septal defect was the most common followed by Atrial Septal Defect with 16% and then PDA with 10%, which is comparable to the study by Kasturi L. et.al wherein cyanotic heart disease constituted 19% and acyanotic 81%. Apart from these 8% patients are seen with multiple lesions i.e several combinations of various congenital heart anomalies.

Presenting with Murmurs: It is well known that murmurs are not seen in all cases of congenital heart disease. In the present study, there are 37 children presenting with murmur which constitutes 74% while 26% presented without any murmur.
Maternal Age: In the present study, 22% of mothers belonged to <20 yrs of age while 66% are between 20 – 30 yrs of age and 12% are above 30 yrs. As we know that some congenital heart anomalies depend on age of mother. For example like that of down syndrome (Trisomy 21). Hence, maternal age is of high value.

History of Consanguinity: Consanguinity plays a major role in the incidence of major congenital malformations in children. In our present study, 8% of patients provided a positive history of consanguinity while 92% provided a negative history.

Extra-cardiac Anomalies: It is well known that many extracardiac anomalies are associated with congenital heart diseases. In the present study, 10% cases include extra-cardiac anomalies like Down syndrome, Rubella syndrome, Mental retardation and VACTERL symptoms. In a study conducted by Kasturi et al., 20% of cases with congenital heart disease had extracardiac anomalies. In a study conducted by Joshi et al., 10% cases of congenital heart disease had syndromes and other associated somatic anomalies of which Down syndrome is the commonest.

History of maternal illness in the first Trimester: There are about 6% cases presenting with maternal illness while 94% are normal during their gestational period.

CONCLUSION: This is a hospital based cross sectional study with 50 children of age group 1 month to 12 years suspected of congenital heart disease and definitive diagnosis is made by 2-D Echocardiography. Majority of cases presented belong to <1 yr age group followed by 8 – 12 yrs age group. Male predominance over female is seen in the presentation of congenital heart disease. Among the cases taken for the study, acyanotic heart anomalies are more common cardiac anomalies. Among which Ventricular Septal Defect is the commonest defect and 8% cases were observed with multiple lesions i.e. presenting with a mixed variety of congenital heart anomalies.

Presence of murmurs is also evaluated in each and every case, which gives a clinical suspicion of congenital heart disease. Most of them are presenting with murmur. There are about 10% cases with extracardiac anomalies down syndrome being the commonest. Consanguineous history is of high importance in the cases of congenital heart disease where 8% cases presented with positive history of consanguinity. About 6% cases showed history of maternal illness in the first trimester as organogenesis occurs in the first trimester of gestation. It is important to take the history of maternal illness during first trimester. To summarise, children with congenital heart disease have unique presentation and carry poor outcome unless diagnosed early and managed properly.

RECOMMENDATIONS:
1) It is the duty of government to increase the public awareness about congenital heart disease in developing countries like India so as to encourage early presentation and enhance early diagnosis.
2) Information regarding the adverse effects of consanguineous marriages should be educated to the people by the government.
3) Government should take it as a responsibility that each and every government hospital is provided with the facility of echocardiography, the diagnostic tool of congenital heart disease’s.
REFERENCES:
5. Structural abnormalities of great arterial walls in congenital heart disease.
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