Original Research Paper

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CONGENITAL HEART DISEASE IN DOWN SYNDROME – SPECTRUM IN A TERTIARY CARE HOSPITAL.

Paediatrics

KEYWORDS: Down syndrome, Congenital Heart Disease, Echocardiography

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ABSTRACT

Background-Congenital heart disease (CHD) is common entity seen in Down syndrome (DS). Approximately 40 to 60% of children with DS have heart defects and among those with CHD, 4-10% are associated with DS. Therefore Down syndrome patients are routinely advised echocardiography (ECHO) to rule out CHD. **Material and method**-This is a prospective hospital based study, over a period of two years. Confirmed Down syndrome patients having congenital heart disease were included in this study. The age and sex of patients, age of the mother, their extra cardiac anomalies and type of cardiac defects confirmed by echocardiography were documented and analysed. **Observation**-Echocardiography was done in 65 confirmed cases and among them 36(54.55%) had some form of congenital heart disease. This study showed that the most common form of CHD in these Down's children was Ventricular Septal Defect (VSD) (25%) followed by Atrial Septal Defect (ASD) (22.2%). Patent ductus arteriosus (PDA) was seen in 7 cases (19.5%) and Tetrology of Fallot (TOF) was seen in 2 cases (5.5%). Conclusion-Fetal Echocardiography can be used as a screening tool for diagnosing Down's syndrome baby having CHD in younger age group of mothers with high parity. All clinically suspected new born should undergone ECHO to exclude cardiac defects at the earliest to reduce morbidity and mortality.

INTRODUCTION

Down's syndrome is a chromosomal disorder associated with characteristic body features, hypotonia, mental disability, heart defects, hypothyroidism, increased risk of leukaemia and duodenal atresia. It is also known as trisomy 21.

In 1866, Doctor John Langdon down was the first, who diagnosed a case of Downs Syndrome. In the 1930s, Waardenburg and Bleyer hypothesized that DS might be due to chromosomal abnormalities. But it was not until 1959 when Jerome Le Jeune and Patricia Jacobs discovered trisomy (triplication) of the 21st chromosome, the smallest human chromosome which is the cause of Down syndrome (1). The parents of the affected individual are typically genetically normal (2). The possibility increases with age of the mother, from less than 0.1% in 20 yrs to 3% in those age 45yrs (3). Down syndrome can be identified during pregnancy by prenatal screening and diagnostic testing. Postnatally it can be diagnosed by typical phenotype and confirmed by karyotyping. Down syndrome affects 1:700 live births (4, 5). Congenital heart disease (CHD) is common entity seen in Down syndrome. Approximately 40 to 60% of children with DS have heart defects and among those with CHD, 4-10% are associated with DS (6). Therefore Down syndrome patients are routinely advised echocardiography to rule out CHD.

MATERIALS AND METHODS:

This is a prospective hospital based study carried out in the department of Pediatrics, IMS and Sum hospital, Bhubaneswar, Odisha over a period of 2 years from July 2014- June 2016. The suspected Down syndrome children basing on clinical findings were advised for karyotyping. Only those children who were Trisomy 21 by conventional karyotyping with G banding technique underwent echocardiography. Those Down syndrome patients having congenital heart disease were included in this study. The age and sex of patients, age of the mother, their extra cardiac anomalies and type of cardiac defects confirmed by echocardiography were documented and analysed.

OBSERVATION

Basing on the typical phenotypical characteristics and clinical examination 72 patients were suspected for Down syndrome and advised for karyotyping. Out of them 65 were confirmed by conventional karyotyping by G banding technique and followed up. Echocardiography was done in 65 confirmed cases and among them 36 had some form of congenital heart disease.

Among these 36 patients having CHD there were 20 (55.6%)were males and 12 (44.4%)females with male female ratio is 1.25:1(fig 1).Twenty patients (55.6%)were in 0-6 months age group,12 patients (33.3%) were between 6 months and 1 year and only 4 patients (11.1%) presented between 1-5 yrs of age(fig 1).According to maternal age, maximum children(41.7%) belong to mother having age group 30-35 years but 50% mothers of below 30 years also had Down babies(fig 2). The index study showed that the most common form of CHD in these Down's children was VSD (25%) followed by ASD (22.2%). Patent ductus arteriosus was seen in 7 cases(19.5%) and TOF was seen in 2 cases (5.5%)(fig 3).There was developmental delay in all children (100%).Recurrent respiratory tract infection was a common problem seen in 66.6% of cases. Hypothyroidism was also not uncommon and seen in19.4% cases (Table 1).

DISCUSSION

Based on the previous data the frequency of CHD in Down syndrome varies from 35-65%, (7-11). In our study 65 confirmed Down syndrome baby underwent echocardiography and approximately 55% had CHD which is similar to the internationally approved data. Thus there is a high incidence of congenital heart disease in Down syndrome.

Advanced maternal age is associated with high incidence of trisomy. After 35 years the risk of conceiving a child with Down syndrome increases significantly. However younger women though having a low risk (12). In our study 50% of mother having Down syndrome children were less then 30years of age. This may be because of early age of marriage, multiparty among females in this part of our country. This finding is similar to studies by Ruben Figueora et al (13). Most of the Down's baby underwent echocardiography during infancy with 55.5% of patients presenting below 6 months of age. The male to female ratio of CHD in Down's baby 1.2:1. There was no significant gender difference in frequency in CHD in others studies also.

In this study the commonest congenital heart defect was ventricular septal defect which comprises (25%) of CHD seen, which was almost similar to other Indian study by Kava MP et al (14). In most European

countries, Sudan, Turkey and USA atrioventricular canal defect was the most common cardiac defect (15-17).

While studies from Guatemala and Saudi Arabia reported that PDA was the most common form of CHD in DS with frequency of 28.6% and 47.8% respectively (18-19). Recent study from Singapore reported that VSD is the most common subtype of CHD followed by PDA, ASD, and AVSD with the frequency of 39.2%, 34.5%, 23.4% and 15.6% respectively (19). The pattern in our study was almost similar to it except that we had a slight higher number of ASD then PDA. Some publications have suggested that ethnicity and various climatic factors, such as high altitude with lower partial pressures of oxygen may lead to a increase incidence of PDA. (20-22).

Down syndrome babies may have multiple cardiac defects too. In our study multiple cardiac defects were seen in 11.1%. Tetralogy of Fallot was reported in 5.5% of cases, was similar to studies reported by Munsi et al, (23) in Caucassian (4%) (16) and Saudi Arabian children (5.3%) (24).

The frequency and distribution of CHD in DS vary in different geographical regions, the cause for this difference is not clear, although some consistency was observed between certain global areas. This variation in geographical distribution may be caused by numerous factors, one of which could be the genetic make-up of each nation or global region or it could be due to specific embryological mechanisms (23).

The developmental delay was found in all babies (100%) diagnosed to be trisomy 21. History of recurrent respiratory tract infection was seen is 66.6% children. In our study congenital hypothyroidism is not uncommon and found to be 19.4% of children which is quite high in contrast to other study (23), where 5.4% cases of Down's have hypothyroidism.

Conclusion

Fetal Echocardiography is commonly advised as a screening method for high risk mothers. But it can be used liberally too as a tool for diagnosing Down syndrome baby having CHD in younger age group of mothers with high parity. As the presence of CHD in trisomy 21 is very high, all clinically suspected new born should undergone echocardiography study to exclude cardiac defects at the earliest to reduce morbidity and mortality.

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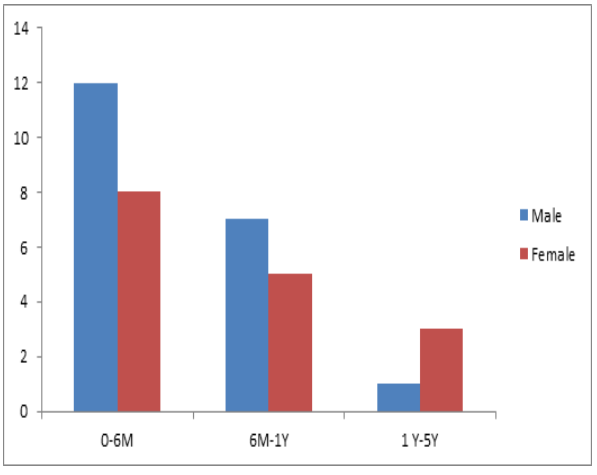

Figure 1. Sample distribution according to Age and Sex

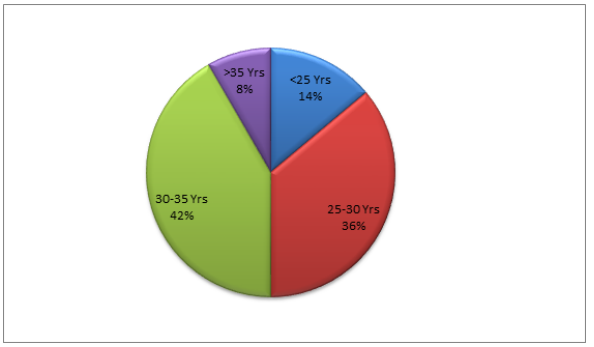

Figure 2. Sample Distribution according to age of Mothers

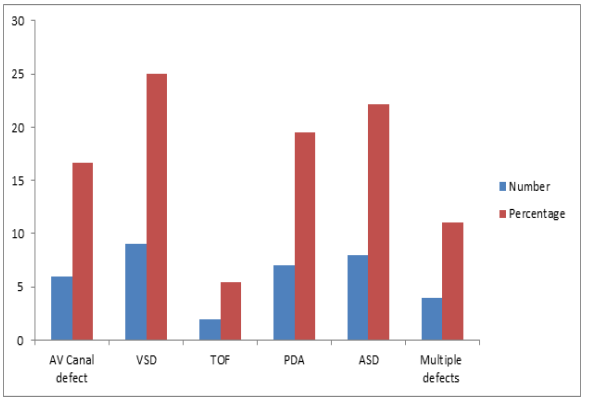

Figure 3. Sample distribution according to congenital heart disease

Table -1. Sample distribution according to Non-cardiac manifestation

Clinical findings	Number	Percentage
Delayed Development	36	100
Recurrent RTI	24	66.6
Hypothyroidism	7	19.4
Duodenal atresia	2	5.5
Ophthalmological manifestation	3	8.3

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