

Chromosomal study in congenital Heart diseases in 39 patients

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Abstract:

Congenital heart disease (CHD) is the most common form of human birth defects accounting for about 30% of the total anomalies. The prevalence rate of CHD is 9 per 1000 live births. The etiology for the majority of these anomalies remains unknown but genetic factors are being recognized as playing an increasingly important role. The aim of this study is to evaluate the CHD in Raichur district, Karnataka and Hyderabad, Andhra Pradesh. The study includes 39 patients with congenital heart defects & confirmation of the heart defects are accomplished by echocardiography. After written consent was obtained from the family members, 39 CHD patients were subjected for conventional cytogenetic studies. Chromosomal analysis was done by using GTG-staining banding. Out of 39 patients, 1 patient was with numerical abnormality (Trisomy 21) and remaining 38 patients were with normal chromosomes.

Key words: Congenital heart diseases, chromosomal analysis, Trisomy 21

Introduction:

Congenital heart disease (CHD) or congenital heart defect is defined as defect in the structure of the heart and great vessels which is present at birth. Many types of heart defects exist, most of which either obstruct blood flow in the heart or vessels near it, or cause blood to flow through the heart in an abnormal pattern^[1]. CHD is the most common birth defect & the leading cause of mortality in the 1st year of life with a prevalence of 9% in live births & 10% in aborted fetuses. In majority cases CHD occurs as an isolated malformation & however in 33% cases have associated anomalies^[2].

Prevalence of CHD in population based studies world wide is found to be range between 1-150/1000 live births & in India it is found to range from 2.2 to 50.89/1000 live births^[3]. Since in a large number of births in our country takes place at home & mostly unsupervised by a qualified doctor & a large number of CHD are critical & leading early death, we have no data on CHD prevalence^[4]. Approximately 10%

infant mortality in India may be because of CHD. Despite advance in medical & surgical care the etiology of the CHD is still not completely understood^[5]. The cause for CHD can be categorized into 3 major groups such as chromosomal, single gene disorders (10-15%) and multiple factors (85-90%). Hence the identification of the chromosomal abnormalities can help the proper management of CHD.

Material and Methods:

For the present study material has been collected from Innova children's hospital, Hyderabad & Sangameswara polyclinic, Raichur. Total 39 patients were enrolled for this present study and they are suffering with various types of CHD. The age group of the patients ranges from 1 year to 15 years and all these patients were clinically diagnosed by 2D Echo cardiography. After taking the written consent from the parents 2-3ml of blood sample collected in heparinized vacutainers in sterile conditions. The collected samples were processed for karyotyping. Total 3 steps are involved in karyotyping. In 1st step blood lymphocyte culture has been done for 72 hours in RPMI medium. In 2nd step lymphocyte are harvested by adding colchicine and fixation has been done with suitable fixative. In 3rd step slides are prepared & GTG staining (G-banding) has been done. Total 20 metaphase plates were observed and karyotype was prepared.

Results:

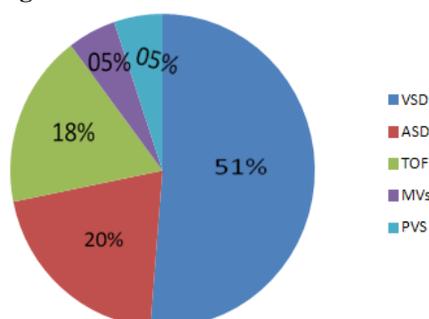
The study group consists of 39 patients among whom 19 are male & 20 patients were females whose age is ranging from 1 year to 15 year with various types of CHD distribution & No of case are explained in Table No 1. Among 39 cases in 1 case Trisomy 21 (Mosaicism) Fig No.1 has been observed and remaining 38 cases chromosomes are normal. Out of 39 cases in 12 cases parents had a consanguineous marriage.

Table No 1 Distribution of the heart defect

Type of CHD Patients	No of
VSD	20(51%)
ASD	08(20%)
TOF	07(18%)
MVS	02(05%)
PVS	02(05%)

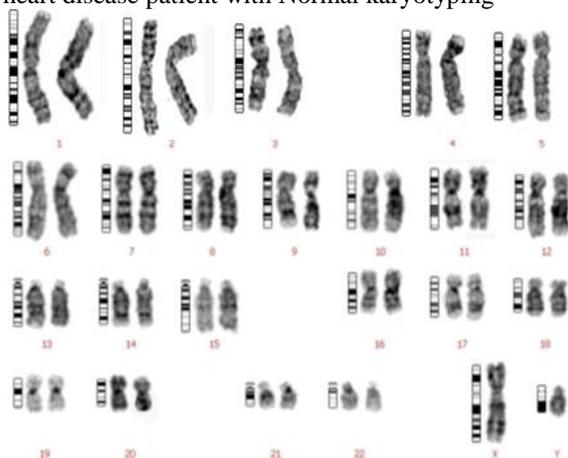
- VSD- Ventricular Septal Defect
- ASD- Atrial Septal Defect
- TOF-Tetrology Of Fallot
- MVS- Mitral valve stenosis
- PVS- Pulmonary valve stenosis

Fig 1: Distribution of CHD



The above figure 1 shows the distribution of the CHD among the tested population.

Fig No 1: G-Banded metaphase of the congenital heart disease patient with Normal karyotyping



Discussion:

The study by H.Gawde, Z.M. Patel, M.I. Khatkhatey, A. D’Souza, S. Babu, R. Adhia and P. Kerkar on a total of 105 patients karyotypic analysis, has been revealed normal karyotyping [6]. Smitha Rame Gowda, Harshavardhan M. Gawde and Nallur B Ramachandra in their study considered a total 136 patients karyotypic analysis. They identified in 18 cases numerical abnormalities, in 3 cases structural abnormalities and in 1 case both numerical and structural abnormality. One of the risk factor for the occurrence of CHD is consanguineous marriages. Smitha Ramegowda and DrNallur B. Ramachandra, revealed total 144 Parental consanguinity in South India, in their study total 144 confirmed CHD cases with an age range of 1 day to 23 years and a control group of 200 families from different localities of Mysore city were taken. Findings revealed first-cousin marriages to be 44.68% and uncle–niece marriages 46.81% [7]. Badaruddoza, Mohd. Afzal, and Akhtaruzzaman, revealed, Parental consanguinity in North India. In their study a total of 1721 infants and children up to the age of 6 years were examined among which 741 children’s around 43% were the offspring of consanguineous marriages. In the present study of 39 cases of CHD one case shown Trisomy 21 (Mosaicism) with 3.3% and in 16 cases about 41% were the offspring of consanguineous marriages [8].

Conclusion:

In the present study Karyotype of CHD has been studied to confirm the diagnosis, to identify the chromosome or chromosomes causing CHD, to study the severity of the CHD, to determine the chance of occurrence of disease in consanguineous marriages and to provide the proper guidance and counseling to the families.

Congenital heart disease (CHD) is the major cause of birth defects, and accounts for more deaths in the first year of life than any other disease or defect thou advance in medical & surgical procedures the etiology of the most cases is unknown. Advance in molecular technology & sequencing of human genome may play important role in identification of gene regulators. However in the present study those cases having normal karyotyping requires more specific molecular biological methods like FISH, PCR for further studies.

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