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# A Prospective Clinical Study of Congenital Heart Disease in Children with Consanguineous Marriages

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**Abstract**
**Objectives**: *1. To study the incidence of Congenital Heart Disease in children born to consanguineously married parents.*
*2. To study the spectrum of Congenital Heart Disease in children born to consanguineously married parents.*
**Material and Methods:** *The study was conducted on 305 consanguineously married parents' children in Dept. of Paediatrics. Govt. General Hospital, Kakinada. from October.'2010 to September.'2011. Age Group selected for this study are 0-5 Years children 2nd Degree, 3rd Degree and 4th Degree consanguineous marriages are included in this study. Maternal gestational diabetes, maternal exposure to teratogenic drugs; Radiations etc. are excluded from the study.*
**Results:** *Out of 305 children born to consanguineously married parents, 36 had congenital anomalies 11.8% of which 16 cases found Congenital Heart Diseases (5.2%) and 20 cases found other Congenital Diseases like Gastroschisis, Club Foot, Cleft lip, Polydactyly, etc (6.55%). The frequency of Acyanotic Heart Diseases are 13 (81.3%) and Cyanotic Congenital Heart Diseases are 3 (18.7%) in 16 cases of Congenital Heart Diseases. Spectrum of Heart Diseases diagnosed are VSD, Dextrocardia, TOF, Complex CHD, ASD, PDA, MR and DORV.*
**Conclusion:** *The study suggests that children born to consanguineous parents are more likely to have congenital heart disease and other anomalies involving various systems. Congenital Heart Diseases found to be more common in children born to 2nd Degree congenital couple.*
**Keywords:** *Consanguineous marriage, Congenital Heart Disease, Gene Copy*

## Introduction

Consanguinity describes a relationship between two people who share common ancestor a "Shared Blood" relationship[1]

The most common form of consanguineous relationship is between first cousins. Usually two unrelated people without carry the same family gene copy. Children of unrelated parents are at low risk of inheriting from each of their parents a copy to the same faulty gene that could result in a genetic condition.

The Closer the biological relationship in between relatives, the more likely they will have the same faulty gene in common. 1st Degree Consanguinity marriage between brother and sister 50% genetic material common. 2nd Degree - Marriage of Fathers own sister or mother own brother 25% genetic material in common. 3rd Degree – Marriage to Father's Sisters Son / Daughter or Mother's brother's Son / Daughter 12.5% genetic material in common. 4th Degree – This type of marriage is between distant relatives in between distant relatives – Minimal risk.

Congenital Heart Diseases occurs in approximately 2-3/1000 infants with Congenital Heart Disease will be symptomatic in the first year of life. Genetic factor causes congenital heart Diseases are A) Single Mutant Genes (AD, AR or X-Linked).

B) Chromosonal Trisomy 21 (50%), Trisomy 13 (40%), Trisomy 18 (90%).

C) Multifactorial Gene Factors believed to be the basis for PDA and some other congenital heart diseases as well [2]. D) Consanguinity.

## Material and Methods

The study was done in the Department of Paediatrics, Govt. General Hospital, Kakinada. for a period of 12 months i.e. from October 2010 to September 2011. 3190 cases of 0-5 Years of age children who are admitted in Govt. General Hospital, Kakinada. Of these 305 children born consanguineous parents were closely monitored and investigated made up the study population. Detailed data was recorded for this study population. Newborns presented with preterm delivery, birth asphyxia, Cyanosis, Vomiting, excessive cry, Abdominal distension, Delayed passage of meconium and urine are thoroughly investigated after detailed clinical examination to rule out major and minor anomalies.

After one month of age, children presenting with failure to thrive, respiratory distress, cyanosis, clubbing, enema and convulsions are examined thoroughly and the following investigations like X-Ray, electrocardiogram, echocardiogram, X-Ray, Barium meal studies are done as required. Infantogram is taken in all new borns. CT Brain and MRI Brain are done whenever required.

1. Chest X-Ray: Standered posteronterior view (for the children < 1 Year) of chest are taken for all cases. In same cases requiring special study lateral views and barium studies are also taken.
2. ECG: 12 lead electrocardiogram is recorded after a good sedation in uncooperative patients V3 R and V4 R right sided leads are also taken in addition to standard chest and limb leads. The recordings are done on a ECG paper moving at speed of 25mm per second and

with a voltage of 1 mllivolt per 10mm standardization. The chamber hypertrophy-ies are determined by the criteria laid down considering the age of the child.

3. 2D. ECHO: Echocardiography is done in all the standard views like parasternal long axis and short axis, apical four-chamber view and suprasternal notch view to enable correct diagnosis.

This opportunity is also utilized to counsel the parents About consequences of consanguineous marriages, the need to undergone antenatal screening after conception and regular antenatal checkups during pregnancy to recognize any anomilies at the earliest in the prenatal period.

## RESULTS

During the study period consanguineous parents children's are 305 (10.5%) and Non-Consanguineous are 2885 (89.5%).

**Incidence of Degree of Consanguinity and Congental Heart Diseases in Study Group As Follows**

| DEGREE OF CONSANGUINITY | TOTAL NO. OF CASES | PERCENTAGES | NO. OF CHD | PERCENTAGES |
|---|---|---|---|---|
| 2ND Degree | 190 | 62.4% | 11 | 5.8% |
| 3rd Degree | 90 | 29.5% | 5 | 5.5% |
| 4th Degree | 25 | 8.1% | 0 | 0.0% |

All Degree 305 – 16 = 5.2%

**Frequency of Acyanotic and Cyanotic Congenital Heart Diseases In The Study Group**

| CONGENITAL HEART DISEASE | NUMBER OF CASES | PERCENTAGES |
|---|---|---|
| ACYANOTIC | 13 | 81.3% |
| CYANOTIC | 3 | 18.7% |

**Incidence of Degree of Consanguinity and Congental Heart Diseases In Stuiy Group**

| TYPE OF CONGENITAL HEART DISEASE | 0-1 Month | 1 Month 1 Year | 1 Year 5 Years | No. Of Cases of CHD | PERCENTAGES |
|---|---|---|---|---|---|
| VSD | 1 | 3 | 2 | 6 | 37.5 % |
| DEXTROCARDIA | 2 | 0 | 0 | 2 | 12.5 % |
| TOF | 0 | 1 | 1 | 2 | 12.5 % |
| COMPLEX CHD | | | | 2 | 12.5 % |
| ASD | 0 | 1 | 0 | 1 | 6.2 % |
| PDA | 0 | 1 | 0 | 1 | 6.2 % |
| MR | 1 | 0 | 0 | 1 | 6.2 % |
| DORV | 0 | 0 | 1 | 1 | 6.2 % |

Most of the children with congenital heart diseases in the study group failure to thrive followed by congestive heart failure, breathlessness, LRTI, Cyanosis, Clubbing and Convulsions.

**Different Modes of Presentation of CHD in The Study Group**

| S. NO. | MODE OF PRESENTATION | NO. OF CASES | PERCENTAGE |
|---|---|---|---|
| 1 | FALURE TO THRIVE | 12 | 75.00 % |
| 2 | CHF | 9 | 56.25 % |
| 3 | BREATHLESSNESS | 7 | 43.75 % |
| 4 | LRTI | 6 | 37.50 % |
| 5 | CYANOSIS | 3 | 18.75 % |
| 6 | CLUBBING | 3 | 18.75 % |
| 7 | CONVULSIONS | 1 | 6.25 % |

## DISCUSSION

Congenital Heart Disease occurs in about 8-10/1000 live births and comprises one of the major diseases in the paediatric age group. Congenital Heart Disease has become an important cause of morbidity and mortality in

infancy and accounts for 2/3 of all major birth defects along with neural tube defects.

In inbred population, parental consanguinity has been observed to aggravate the underlying genetic risk factors for congenital heart diseases suggesting an autosomal recessive component.

The closer the biological relationship between relatives the more likely they will have the same faulty gene in common. So, Children of parents who are blood relatives generally have increased risk of having birth defects 3

This study is carried out to assess the impact of consanguinity on frequency of congenital heart diseases and other congenital anomalies.

Out of 3190 admissions, 305 children are found to be born to consanguineous parents (10.45%).

Out of 305 children, 16 children are found to be having congenital heart diseases, that is with a frequency of 5.24 % amount consanguineous parents.

In the Present study acyanotic heart diseases are found to be most common – 81.25% (13 out of 16) followed by cyanotic heart diseases – 18.75 % (3 out of 16).

VSD is more common (37.57%) in congenital heart diseases.

**Comparison of Type of the CHD with Othter Studies**

| S. No. | Type of CHD | Rame Gowda (Mysore) | BDRI (Chennai) | Present Study (Kakinada |
|---|---|---|---|---|
| 1 | VSD | 4.97 % | 17.8 % | 37.5 % |
| 2 | ASD | 19.6 % | 10.2 % | 6.25 % |
| 3 | TOF | 4.8 % | 3.4 % | 12.5 % |
| 4 | PDA | 9.2 % | 6.3 % | 6.25 % |
| 5 | DORV | - | 2.5 % | 6.25 % |

If we consider relative frequency of degrees of consanguinity in the study group 2nd Degree formed the major group [190 (62.4%)] followed by 3rd Degree [90 (29.45%)] and 4th Degree [25 (8.1%)].

Among all the degree of consanguinity childern born to 2nd Degree consanguineous parents are found to be must affected with congenital Heart Diseases. No Congenital Heart Disease is found in childern of 4th Degree consanguineous parents.

Most of the childern with congenital Heart Disease are admitted during the period of 1 Month to 1 Year, which constitute 50% (8 out of 16).

Male to Female ratio with congenital Heart Disease in the study group is 1.6:1.

Regarding mode of presentation, must of the childern with Congenital Heart Diseases presented with failre to thrive [75% (12 out of 16)] followed by Congestive Heart Failure [56.25% (9 out of 16)

Frequency of anomalies other then Congenital Heart Diseases in the study group is 6.55%.

## CONCLUSIONS

The study suggestive that childen born to consanguineous parents are more likely to have congenital Heart Disease and other anomalies involving various systems.

The study suggests that nearer the blood relation between the parents the more likely that their childern are to be born with congenital anomalies, compared to distant relation.

There is a need to educate the public on deleterious effects of inbreeding, especially in south india, which has high overall consanguinity.

For possible prevention genetic counselling before marriage must be applied for all consanguineous parents.

Off springs of all consanguineous parents should be throughly examined and investigated for congenital anomalies, particularly for congenital heart disease, because they are being missed commonly at the new born period.

**REFERENCES**


1. Bromiker R etal. Association of Parental Consanguinity with congenital malformations among Arab neonates in Jerusalem Clinical Genetics 66 (1) : 63-6.
2. Holmes LB, Multifactorial inheritance in Nelson text book of pediatrics, 18th Edition, Behrman et al, WB Saunders company, phil.2004.
3. Nabulsi MM, Tamim H, Subbagh M, Obeid My, Yunis KA, Bitar FF, Paental Consanguinity and congental heart malformations in a developing countty, AM J Med Genet A 2003:116: 342-7.
4. Moller JH, Amplatz K, Edwards JE: Congenital Heart Disease. Malaazool, Mich, Upjohn, Rudolph AM: Congenital Diswases of the Heart, Chicage, Mosby.
5. Sridaromont S, Fildt RH, Ritter DG Et al: Double outlet right ventricle: Hemodynamic and anatomic correlation... am J Fardial 38: 85-94.
6. Hoffman JI, Kaplan S. The incidence of congenital heart Disease. Jam Coll Cardiol 002 39:1890-900.
7. Mc Keown T, Record Rg: Malformations in a popultion observed for 5 years after birth. Ciba Foundation Symposium on Congenital Malformations, London, Churchill pp2-21.
8. MG Gnanlingham KK Gnanalingham, A Singh in Acta paediatrica, Congenital Heart Disease and parental consanguinity in South India.
9. Kulkarni ML, Mathew K, Consanguinity and its effects on fetal growth and development, J Med Genet 1990 27:348-352.
10. Parental Consanguinity increases congenital heart diseases in south India-Ramegowda University of Mysore -India-2006.
11. Kulkarni ML, Congenital Malformation, Editiorial Indian Pediatric 189 28 5-10.
12. An Praagh R, Weinberg P, Fornan RB, et al: Malposition of the heart. In Adams FH, Emanouilides GC, Riemenschneider TH (eds): Moss'Heart Disease in infants, Childern, and Adolscents, 4th ed. Baltimore, Williams & Wilkins.
13. Lupinetti FM, Kulik TJ, Beekman RH, et al: Correction of total anomalous pulmonary venous connection in infancy. J Thorac Cardiovase Surg 106 : 880-885.
14. Kawabori I: Cynotic congnital heart defects with increased pulmonary blood flow. Pediatr Clin North Am 25: 777-795.
15. Stevenson JG: Acyanotic lesions with increased pulmonary Blood flow. Pediatr Clin North Am 25: 743-785.
16. Pak MK, Gountheroth WG: How to Read Pediaric ECGs 4th ed. Philadelphia, Mosby 2006.
17. Goldmunts E: The Genitic Contribution to Congenital Heart Disease, Pediatric Clin North Am 2004.