# STUDY OF MULTIPLE CONGENITAL ANOMALIES IN THE CHILDREN OF CONSANGUINEOUS COUPLES FROM BANGALORE DISTRICT

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## ABSTRACT

**Aim of the present study:** Consanguinity is a tradition prevalent in India. Our study aims to determine the role of consanguinity on congenital anomalies and identify the different types of anomalies and the percentages of their incidence that we came across in Raja Rajeswari Medical College & Hospital, Bangalore and other hospitals of Bangalore district in offsprings of consanguineous couples.

**Period of study:** A two years study from beginning of 2007 to end of 2008. Materials and Methods: One thousand and two hundred patients were screened for study of anomalies. Anomalies were observed in sixty four (5.33%) subjects of different age groups born to consanguineous couples and constituted the materials for the present study.

**Results:** There were sixty four (5.33%) cases of congenital anomalies in patients of different age groups ranging from new born to adult age groups. Males were 59.37% and females were 40.63%. Higher incidence of anomalies was noted involving the musculoskeletal system and facial anomalies (15.625% each) followed by ear, nose and throat anomalies (14.06%). The least incidence was dental anomaly (01.56%).

**Conclusion:** Consanguinity may result in congenital anomalies due to homozygosity of genes. Counseling of such couples is essential to avoid congenital anomalies in the offspring.


**Keywords:** Consanguinity, consanguineous couples, heredity, heterogeneity, superstitions, birth defects, abnormalities, anomalies.

## INTRODUCTION

Consanguineous marriages have been described as an important factor contributing to increased congenital malformations.[1] Developmental anomalies are common, but their types & incidences differ from one system to another. Some anomalous babies are born dead, some fetuses spontaneously get aborted, some need termination, while some others need constant antenatal check up followed by early treatment after birth[1]. Our study focuses on the incidence of congenital anomalies present in offsprings of consanguineous couples and the various systems involved in the anomalies.

## MATERIALS AND METHODS

1200 patients were examined for the presence of multiple congenital anomalies from different hospitals of Bangalore district and also from the hospital of Rajarajeswari Medical College, Bangalore.

## RESULT

Sixty four cases of multiple anomalies from consanguineous couples were observed. All the cases were of different age groups ranging from new born to adult age groups. Anomalies were observed in 59.37% (n=38) males and 40.63% (n=26) females.

## OBSERVATION

The anomalies noted were classified under eleven systems (Table 1).

**Table 1 : Showing percentages of anomalies in 11 systems (n=64)**

| S. No | System | Number of cases | Percent ages |
|---|---|---|---|
| 1 | Cardiovascular anomalies | 02 | 03.125% |
| 2 | Craniospinal anomalies | 04 | 06.25% |
| 3 | Anomalies of eye | 04 | 06.250% |
| 4 | Musculoskeletal anomalies | 10 | 15.625% |
| 5 | Dental anomalies | 01 | 01.56% |
| 6 | Ear, nose and throat anomalies (ENT) | 09 | 14.062% |
| 7 | Renal anomalies | 07 | 10.937% |
| 8 | Anomalies of skin | 05 | 7.8125% |
| 9 | Gastrointestinal anomalies | 07 | 10.937% |
| 10 | Facial anomalies | 10 | 15.625% |
| 11 | Syndromes (genetic) | 5 | 7.8125% |

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**Table 2: Showing percentages of anomalies in males and females (n=64)**

| S. No | Anomalies in the systems | MALES | | FEMALES | |
|---|---|---|---|---|---|
| | | No. | Percentage | No. | Percentage |
| 1 | Cardiovascular anomalies | 02 | 3.125% | 00 | 00 |
| 2 | Craniospinal anomalies | 03 | 4.68% | 01 | 1.56% |
| 3 | Anomalies of Eye | 01 | 1.56% | 03 | 4.68% |
| 4 | Musculoskeletal anomalies | 06 | 9.375% | 04 | 6.25% |
| 5 | Dental anomalies | 00 | 00 | 01 | 1.56% |
| 6 | Ear, nose and throat anomalies | 06 | 9.375% | 03 | 4.68% |
| 7 | Renal Anomalies | 05 | 7.81% | 02 | 3.125% |
| 8 | Anomalies of skin | 04 | 6.25% | 01 | 1.56% |
| 9 | Gastrointestinal anomalies | 05 | 7.81% | 02 | 3.125% |
| 10 | Facial anomalies | 03 | 4.68% | 07 | 10.93% |
| 11 | Syndromes (genetic) | 03 | 4.68% | 02 | 3.125% |
| | **TOTAL** | 38 | 59.37% | 26 | 40.63% |

Table 2 shows males (59.37%) have a higher incidence of anomalies compared to females(40.63%). In males incidence of musculoskeletal system and ENT anomalies are found to be higher (9.375%) followed by renal & gastrointestinal anomalies (7.81%). In females facial anomalies were found to be higher (10.93%) followed by musculoskeletal anomalies (6.25%).

The following are the various anomalies detected in the patients in 11 systems :

1) Cardiovascular anomalies: 2 cases(3.125%): Atrial Septal Defect-1 and Fallot's tetrology-1
2) Craniospinal anomalies: 4 cases(06.25%): Meningocele - 1, Myelomeningocele - 2, Spina bifida - 1, Anencephaly - 1
3) Anomalies of eye : 4cases (6.25%) : Iridolenticular coloboma-1, Aniridia-1, Retinochoroidal coloboma -1, Congenital cataract-1
4) Musculoskeletal anomalies: 10 cases(15.625%): Sessile polydactyly-1, Pedunculated polydactyly -1, Syndactyly-1, Club foot- 6, Polydactyly of thumb-1
5) Dental anomaly: 1 case(01.56%): Congenital absence of teeth-1
6) ENT anomalies: 9 cases(14.062%): Absence of external auditory meatus-3, Microtia with external auditory canal atresia -3, Deformity of pinna-3
7) Renal Anomalies: 7 cases(10.937%): Agenesis of right kidney-2, Congenital nodular kidney-2, Polycystic kidney -2, Bilateral enlarged echogenic kidney with hypospadias-2
8) Anomalies of skin : 5 cases (7.8125%) : Haemangioma-2, Harlequin baby- 1, Xeroderma pigmentosa-2
9) Gastrointestinal anomalies : 7 cases (10.937%) : Atresia of jejunum associated with dilated stomach and duodenum-2, Omphalocele-1, Gastroschsis-4
10) Facial Anomalies : 10 cases (15.625%) : Unilateral hare lip-5, Bilateral hare lip-2, Cleft palate -2, Facial cleft-1
11) Genetic Syndromes: 5 cases(7.8125%): Rubinstein Taybi Syndrome (16p13-Deletion)-1, Down's syndrome (trisomy 21)-3, Edward syndrome (trisomy 18)-1

The following histories of the mothers were taken: i) History of drug intake during 1st trimester ii) Family history of consanguinity iii) Family history of systemic diseases like hypertension, diabetes and seizures iv) History of still birth, repeated abortions, hydramnios v) History of similar anomalies in previous pregnancies

Routine & relevant investigations of mothers of anomalous babies done during antenatal check up were noted.

**DISCUSSION**

Congenital abnormality refers to any anomaly present at birth.[6,7,8] Consanguinity is defined as a marital relationship among blood relatives. Congenital anomalies associated with consanguinity are due to homozygosity of genes. There may be manifestations of rare traits of genes. The etiology of major multiple congenital anomalies is genetic (30%-40%) and environmental(5%-10%). Etiology of other 50% of multiple congenital anomalies is not known. Chromosomal abnormality constitutes 6% of total genetic causes. Single gene disorder is 25% & Multifactorial causes are 20-30%. Among major multiple congenital anomalies, central nervous system & cardiovascular system are found to be involved more frequently. [9] Consanguinity is said to be the etiological factor for the increased incidence of mental retardation (MR) and multiple congenital anomalies (MCA) which result in increased morbidity and mortality.[10]

The report of incidence of consanguinity in the literature varies between 2 to 60%. And in India, it is 5 to 60%.[11] Incidence of genetic disorders is 2.3% in India[12] and incidence of multiple congenital anomalies is 1.94% - 2.03% as per the analysis of all published studies. [13&14] The commonly occurring multiple congenital anomalies have been found to be defects of neural tube or anomalies of cardiovascular system, musculoskeletal system & gastro intestinal systems.[15-24]

**PRESENT STUDY**

In the present study, incidence of anomalies was 5.33%. They were seen more in males (59.37%) than in females (40.63%). There was second degree of consanguinity in the parents of children with congenital anomalies. Anomalies were noted in offsprings ranging from new born to adult age group of third decade. Higher number of anomalies were found in musculoskeletal system and facial anomalies (15.625%), followed by Ear, nose & throat anomalies (14.06%). The study showed cases of meningocele, myelomeningocele, spina bifida and anencephaly (6.25%). There was bad obstetric history like history of repeated abortions (22.50%) in the mothers in our study. In our study, 65% cases showed multiple congenital anomalies & 35% showed single anomaly. In this study of multiple congenital anomalies, a combination of anomalies of gastrointestinal system, musculoskeletal system and craniospinal defects was found to be more frequent unlike in other studies. This may be due to population or racial differences or due to environmental factors and not following the advice of the treating doctor.

**CONCLUSION**

Detection of anomalies is most advantageous for an obstetrician in early stages of pregnancy so that she can plan for future treatment & also for the decision whether to continue the pregnancy. Ultrasonography has become a useful tool for early diagnosis of anomalies. Our study on multiple congenital anomalies in consanguineous couples gives information regarding incidence of various types of anomalies associated with consanguinity**.** Counseling of such couples is essential to avoid congenital anomalies in the offsprings. Awareness of increased incidence of morbidity and mortality due to consanguinity can be spread to the population by health education through health workers, social workers and NGOs.

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