STUDY OF CONGENITAL MALFORMATIONS IN NEWBORN
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HOW TO CITE THIS ARTICLE:
K. Koteswara Rao, A. Krishna Prasad, D. Manikyamba, K. Adi Reddy, Solomon Saawan P, S. Anusha. "Study of Congenital Malformations in Newborn". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 78, September 28; Page: 13582-13595, DOI: 10.14260/jemds/2015/1943

ABSTRACT: The etiology of congenital malformations has not been clearly defined. It is interesting to note that certain congenital malformations are more prevalent in some areas¹. Neural tubal defects (NTD) are common in Punjab and cleft lip (CL) is common in south India. Gastrointestinal tract defects are detected from south i.e. Mysore, Trivandrum, Hyderabad and Pondicherry. Polydactyly was prevalent in south India and Chandigarh. Incidence of Talipes was highest in Delhi and next in order were Chennai, Pondicherry and Patna. Malformations are controlled by genetic and environmental factors and a thorough analysis would indicate the factors responsible for their genesis and thereby, their means of prevention². Thus it is imperative that in every region the prevalence and peculiarities of malformations should be studied. Therefore, the present study was carried out in Government General Hospital, Kakinada, where consanguineous marriages are very much prevalent. The aim is to study the spectrum, incidence and maternal risk factors associated with congenital malformations.

KEYWORDS: Congenital, Malformations, Birth Defects, Consanguinity, Anomalies.

INTRODUCTION:
STUDY METHOD: The study was undertaken in the NICU, Department of Pediatrics, Govt. General Hospital, Kakinada in collaboration with Department of Gynecology and Obstetrics, Govt. General Hospital, Kakinada. This is a prevalence study. The present study was carried out from September 2014 to August 2015 and 10,720 consecutive births were studied in Government General Hospital, Kakinada.

RESULTS:
• Out of 146 cases, CNS defects were seen in 29 cases and accounted for 19.8%, GIT defects in 25 cases (17.1%), Genitourinary malformations were 15 cases (10.3%), CVS were 14 cases (9.6%), musculoskeletal anomalies were 12 cases (8.2%), Oral clefts were 12 cases (8.2%), syndromes were 10 cases (6.8%), skin defects were 8 cases (5.5%), multiple anomalies were 9 cases (6.2%), others were 12 cases (8.2%).
• Incidence among male babies was 14.8/1000 total births, among females was 12.3/1000 total births. Incidence of malformations was 1.17% among live born babies and 8.6% among still born babies³.
• Out of 24 cases of stillborn babies, 8 cases (36%) were having CNS malformations, 7 cases (32%) were GI anomalies and 3 cases (14%) had multiple defects.
• History of Polyhydramnios was recorded among mothers of 10.3% malformed babies,⁴ prematurity in 8.2% and oligohydramnios in 6.8%. Maternal history of PIH was obtained in 6.2%. There was history of fever in 1st trimester in 4.1%, history of repeated abortions in 1.37% and maternal history of Diabetes and hypothyroidism was observed in 2.7% and 0.68% respectively.
• The parents of 8.3% of malformed babies were married to their first cousins. 13.6% were married to a relative further than first cousin. In the present study, parental consanguinity had been noted in 21.9% of all malformations and 33.4% of Musculoskeletal defects, 30% of syndromes, 20% of G.I defects & G.U defects, 13.8% of C.N.S defects, 33.3% of miscellaneous defects.

CONCLUSION: No definite environmental insult could be established as a cause in any of the cases. It was presumed that the abnormalities were mainly caused by genetic factors. A direct relation between the maternal age and occurrence of Down syndrome was noted. Maternal disorders complicating pregnancy like Polyhydramnios, oligohydramnios, maternal febrile illness, PIH were found to be one of the associated factors in the development of congenital malformations in newborns.

Good and thorough antenatal care regarding identification and treatment of maternal complications can prevent malformations. Whenever possible, a thorough screening of the mother and fetus is to be done to rule out intra uterine infections. People should be counseled about consanguinous marriages and about the relation between higher maternal age and Down syndrome. A good and sound genetic counseling will prevent the recurrence of another congenitally malformed baby. Antenatal diagnosis of congenital malformations by means of ultrasonography, Amniocentesis and chorionic villous biopsy is very essential as mother could to terminate her pregnancy so that a lot of morbidity can be avoided.

MATERIALS AND METHODS:
Source of Data: The present study was carried out from September 2014 to August 2015 and 10,720 consecutive births were studied in Government General Hospital, Kakinada. All of the children are born at this hospital. Out of these, 146 cases were having congenital malformations. The cases represent random mixture of all categories of people as the hospital caters to the needs of people from all strata coming mainly from two Godavari districts. The population consists of people from all walks of life, though majority of them are poor and illiterate.

Inclusion Criteria: All of the children who were born at this hospital during the study period were included in this study.

Exclusion Criteria: Children born outside and brought to this institute were excluded from the study.

Method of Collection of Data: A detailed history of mother regarding risk factors, maternal investigations like ante natal scans along with relevant laboratory investigations and detailed family history was taken and the babies were thoroughly examined for congenital malformations and data was recorded as per proforma. Relevant available investigations were done whenever necessary.

RESULTS: In the present study, the incidence of congenital malformations in newborn was 13.6/1000 total births.
### Sex Distribution:
Out of 10,720 babies born in this period 5,585 were male and 5,135 were female and the incidence was 14.8/1000 births among males and 12.3/1000 among females.

### Incidence in Live born and still born babies:
Congenital malformations were found in 122 cases of 10,440 live born (1.17%) and in 24 cases of 280 still born (8.6%).

| Types       | No. of Births | No. of Malformations | %  |
|-------------|---------------|----------------------|----|
| Live Born   | 10,440        | 122                  | 1.17|
| Still Born  | 280           | 24                   | 8.6 |
**Pattern of malformations in both live born and stillborn babies:** In the present study, highest numbers of malformations were seen (in decreasing order) in CNS, GIT and Musculoskeletal system.

| System            | Still Born | Live Born | Total |
|-------------------|------------|-----------|-------|
|                   | No   | %     | No   | %     | No   | %     |
| C.N.S             | 8    | 36    | 21   | 16.9  | 29   | 19.9  |
| G.I.T             | 7    | 32    | 18   | 14.5  | 25   | 17.1  |
| M.S               | 0    | 0     | 12   | 9.7   | 12   | 8.7   |
| G.U               | 0    | 0     | 15   | 12.1  | 15   | 10.4  |
| C.V.S             | 4    | 18    | 10   | 8.1   | 14   | 9.6   |
| Oral Clefts       | 0    | 0     | 12   | 9.7   | 12   | 8.2   |
| Eye               | 0    | 0     | 5    | 4     | 5    | 3.2   |
| Skin              | 0    | 0     | 8    | 6.5   | 8    | 5.5   |
| Syndromes         | 0    | 0     | 10   | 8.1   | 10   | 6.8   |
| Multiple          | 3    | 14    | 6    | 4.8   | 9    | 6.3   |
| Miscellaneous     | 0    | 0     | 7    | 5.6   | 7    | 4.8   |
| **Total**         | 22   | 100   | 124  | 100   | 146  | 100   |

**Distribution of different types of CNS defects:**

![Meningomyelocele Image]
CNS defects were seen in 29 cases (19.9%) out of 146. Meningomyelocele was the commonest CNS anomaly seen in 10 cases (34.5%) followed by hydrocephalus in 9 cases (31%).

| Malformation                     | No | %  |
|----------------------------------|----|----|
| Meningomyelocele                 | 10 | 34.5|
| Anencephaly                      | 4  | 13.8|
| Hydrocephalus                    | 9  | 31  |
| Spinabifida                      | 1  | 3.5 |
| Microcephaly                     | 2  | 6.9 |
| Sacrococcygeal teratoma          | 1  | 3.5 |
| Dandy Walker Malformation        | 2  | 6.8 |
| **Total**                        | **29** | **100** |

Distribution of different types of G.I defects:
GI defects were seen in 25 cases (17.1%) out of 146. Most common were Anorectal malformations and congenital diaphragmatic hernia in 6 cases (24%) each.

| Malformation                  | No | %   |
|-------------------------------|----|-----|
| Anorectal Malformations       | 6  | 24  |
| Exomphalos                    | 3  | 12  |
| Gastroschisis                 | 3  | 12  |
| Diaphragmatic hernia          | 6  | 24  |
| Duodenal atresia              | 2  | 8   |
| Tracheo-esophageal Fistula    | 5  | 20  |
| **Total**                     | **25** | **100** |

**Distribution of various types of Musculoskeletal defects:** Musculoskeletal defects were seen in 12 cases (8.2%) out of 146. TEV is found in 25% & Polydactyly, Genu recurvatum and CDH were found in 16.7% each. In many of these cases maternal history of Oligohydramnios was obtained.

| Malformation                  | No | %   |
|-------------------------------|----|-----|
| Talipes equinovarus           | 3  | 25  |
| Genu recurvatum               | 2  | 16.7|
| Absence of Depressor Angularis oris | 1 | 8.4 |
| Cong dislocation of hip       | 2  | 16.7|
| Polydactyly                   | 2  | 16.7|
| Phacomelia                    | 1  | 8.4 |
| short neck                    | 1  | 8.4 |
| **Total**                     | **12** | **100** |
**Distribution of various types of Genito Urinary defects:** Genito urinary defects were seen in 15 cases (10.3%). Among the G.U defects one peculiar case of absence of penis was observed. There were 5 cases of hypospadias, 2 cases of hydro ureteronephrosis, one case of epispadias, ambiguous genitalia and dystrophic kidney each.

| Malformation                          | No | %   |
|---------------------------------------|----|-----|
| Hypospadias                           | 5  | 33.2|
| Exstropy Of Bladder +Epispadias        | 1  | 6.7 |
| Congenital Hydrocele                   | 3  | 20  |
| Hydro uretronephrosis                  | 2  | 13.3|
| Dystrophic Kidney                     | 1  | 6.7 |
| Absence of Penis                      | 1  | 6.7 |
| Ambiguous genitalia                   | 1  | 6.7 |
| Ovarian cyst                           | 1  | 6.7 |
| **Total**                             | 15 | 100 |

**Distribution of Cardiovascular Anomalies:** Cardiovascular defects were seen in 14 cases (9.6%), consisting of 35.6% cases of Acyanotic Heart Disease and 21.4% cases of Complex Heart Disease and Cyanotic Heart Diseases each. One each case of atrial myxoma, Hypo plastic Heart, and Intra cardiac tumor were observed. The diagnosis was by antenatal ultrasonography and X-Ray, ECG and 2D ECHO of baby.

| Malformation                          | No | %   |
|---------------------------------------|----|-----|
| Acyanotic Heart Disease               | 5  | 35.6|
| Cyanotic Heart Disease                | 3  | 21.4|
| Intra Cardiac Tumor                   | 1  | 7.2 |
| Hypo plastic Heart                    | 1  | 7.2 |
| Atrial Myxoma                         | 1  | 7.2 |
| Complex Heart Disease                 | 3  | 21.4|
| **Total**                             | 14 | 100 |
Distribution of Oral defects: Oral defects were seen in 11 cases (7.5%) out of 146. Among oral clefts 18.2% were cleft lip, 36.35% were cleft palate and 45.45% had both cleft lip + palate.

| Malformation  | No. | %    |
|---------------|-----|------|
| Cleft Lip     | 2   | 18.2 |
| CL + CP       | 5   | 45.45|
| Cleft Palate  | 4   | 36.35|
| Total         | 11  | 100  |

Distribution of Eye defects: Eye defects were seen in 5 cases (3.4%) out of 146. Among eye defects, Anophthalmia, congenital cataract, corneal opacity, Buphthalmous, congenital retinoblastoma accounted for 20% each.

| Malformation            | No. | %  |
|-------------------------|-----|----|
| Cataract                | 1   | 20 |
| Buphthalmous            | 1   | 20 |
| Corneal opacity         | 1   | 20 |
| Anophthalmia            | 1   | 20 |
| Cong Retinoblastoma     | 1   | 20 |
| Total                   | 5   | 100|
Distribution of Skin Defects: Skin defects were seen in 8 cases (5.5%) out of 146.

| Malformation            | No. | %   |
|-------------------------|-----|-----|
| Capillary Haemangioma   | 3   | 37.5|
| Pre Auricular Tag       | 3   | 37.5|
| Icthyosis               | 2   | 25  |
| **Total**               | **8**| **100**|

Distribution of Syndromes: Syndromes were seen in 10 cases (6.8%) out of 146. Among these syndromes, 3 cases of Down syndrome had been noted, of which two are male and one female. In two cases maternal age was more than 30 years.\(^\text{16}\)

| Malformation                          | No. | %   |
|---------------------------------------|-----|-----|
| Down Syndrome                         | 3   | 30  |
| Edward Syndrome                       | 2   | 20  |
| Pierre Robin Syndrome                 | 2   | 20  |
| Short Rib Polydactyl Syndrome         | 1   | 10  |
| Arthrogryposis Multiplex Congenita    | 1   | 10  |
| Corneliea de Lange Syndrome           | 1   | 10  |
| **Total**                             | **10**| **100**|
Distribution of Multiple Anomalies: There were 9 cases (6.2%) with multisystem involvement. Two cases were Dandy Walker Cysts with other anomalies, two cases were CHD with limb defects, two cases were ascites with microcephaly and others were cloacal extropy, corpus callosal agenesis and Anophthalmia one each.

| Malformation                                      | No. | %    |
|--------------------------------------------------|-----|------|
| Dandy walker cyst + Bladder Outlet Obstruction   | 1   | 11.10|
| Corpus Callosal Agenesis + Bilateral HDN          | 1   | 11.10|
| Dandy Walker Cyst + Haemangioma                  | 1   | 11.10|
| Cloacal Extropy + CTEV                           | 1   | 11.10|
| Anophthalmia + Bilateral HDN                     | 1   | 11.10|
| Fetal Ascites + Microcephalus                    | 2   | 22.25|
| CHD + Limb Defects                               | 2   | 22.25|
| **Total**                                        | **9** | **100** |

Distribution of Miscellaneous Defects: Distribution of Miscellaneous defects seen in 7 cases (4.8%).

| Malformation             | No. | %    |
|--------------------------|-----|------|
| Cystic hygroma           | 2   | 28.55|
| Retro peritoneal teratoma| 1   | 14.3 |
| Posterior choanal atresia| 2   | 28.55|
| Dentigerous cyst         | 1   | 14.3 |
| Prenatal teeth           | 1   | 14.3 |
| **Total**                | **7** | **100** |
Association of Maternal Factors in Congenital Malformations:

| Maternal History                  | No. | %   |
|----------------------------------|-----|-----|
| Fever in 1st Trimester           | 6   | 4.1 |
| Polyhydramnios                   | 15  | 10.3|
| Oligohydramnios                  | 10  | 6.8 |
| H/o Repeated Abortions           | 2   | 1.37|
| Diminished Fetal Movements       | 3   | 2.1 |
| Preeclampsia                     | 9   | 6.2 |
| Prematurity                      | 12  | 8.2 |
| Gestational Diabetes             | 4   | 2.7 |
| Hypothyroidism                   | 1   | 0.68|

Certain adverse maternal factors were associated with malformations especially fever during 1st trimester, Polyhydramnios, oligohydramnios and eclampsia. All cases of Anencephaly had maternal history of Hydramnios in this study. Maternal history of hydramnios, oligohydramnios and preeclampsia were present in 10.3%, 6.8% and 6.2% of cases respectively. There was no history of maternal smoking and alcoholism in this study.

**Parental Consanguinity:** In the present study, parental consanguinity had been noted in 21.9% of all malformations and 33.4% of Musculoskeletal defects, 30% of syndromes, 20% of G.I defects & G.U defects, 13.8% of C.N.S defects, 33.3% of miscellaneous defects. The parents of 8.3% of malformed babies were married to their first cousins. 13.6% were married to a relative further than first cousin.

**DISCUSSION:** Congenital malformations are prevalent throughout the world in all socio-economic strata in different ethnic groups. In the present study, the incidence of malformations in newborn is 13.6/1000 total births. The present study was carried out in children brought to Government General Hospital, Kakinada. Those delivered at home by midwives are and not brought to this institute are not included. Thus, the figures might not be exact, still they gives a picture of the incidence. The present study is a prospective study and gives a reliable picture.
It is possible that certain adverse environmental factors due to rapid industrialization and increased population flow over the area covered in the study might have been responsible for the higher incidence of malformations\cite{18}. At present the high incidence in this area could probably be attributed to the high prevalence of consanguineous marriages. The rate of malformed babies was higher among stillborn. Incidence of congenital malformation was significantly higher among male babies, preterm babies, still births, LBW babies and in consanguineous marriages. In many of the cases of musculoskeletal malformations\cite{19}, maternal history of Oligohydramnios was obtained and this might have caused the deformity in these cases.

CONCLUSION: 146 malformed babies were detected among 10,720 newborns. Of these, CNS malformations were 29 (19.9%), Gastrointestinal were 25 (17.1%), Genitourinary malformations were 15 (10.3%), CVS were 14 (9.6%), musculoskeletal anomalies were 12 (8.2%), Oral clefts were 12 (8.2%), syndromes were 10 (6.8%), skin defects were 8 (5.5%), multiple anomalies were 9 (6.2%), others were 12 (8.2%). Parental consanguinity was noted 21.9%. No definite environmental insult could be established as a cause in any of these cases. It was presumed that these abnormalities were mainly caused by genetic factors. A direct relation between the maternal age and occurrence of Down syndrome was noted. Maternal disorders complicating pregnancy like Polyhydramnios, oligohydramnios, maternal febrile illness, PIH were found to be one of the associated factors in the development of congenital malformations in newborns.

Most of the congenital malformations are preventable. Good and thorough antenatal care regarding identification and treatment of maternal complications can prevent malformations. Administration of folic acid during the periconceptional period will go a long way in preventing NTD like Anencephaly\cite{20}. Scrupulous avoidance of teratogenic drugs and unnecessary exposure to radiation during antenatal period should be adhered to one and all \cite{21}. Wherever possible a thorough screening of the mother and fetus to be done to rule out intra uterine infections.

People should be counseled about consanguinous marriages and about the relation between higher maternal age and Down syndrome. A good and sound genetic counseling will prevent the recurrence of another congenitally malformed baby. Antenatal diagnosis of congenital malformations by means of ultrasonography, Amniocentesis and chorionic villous biopsy is very essential as mother could to terminate her pregnancy so that a lot of morbidity can be avoided.

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FINANCIAL OR OTHER COMPETING INTERESTS: None