Original Research Article

Pattern of congenital anomalies in neonates at tertiary care centre in Hyderabad, India: a hospital based prospective observational study

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ABSTRACT

Background: With advancements of perinatal, neonatal care congenital defects were the most common cause of morbidity and mortality in developed world. It is one of the common causes of morbidity and mortality in India. Its incidence also influenced by many preventable risk factors. Hence, we are carried out this study to know the changing pattern of congenital anomalies and to know the effect of environmental risk factors on congenital anomalies.

Methods: Prospective observational study conducted at Niloufer hospital Hyderabad during period from November 2017 to 2018. We included intramural and extramural babies. Analysed data by appropriate statistical methods.

Results: Most common system involved was Central nervous system (CNS) with 25 cases out of 112 cases followed by Gastrointestinal system (GIT)and Cardiovascular system (CVS). Meningomyelocele, anorectal malformations and acyanotic heart diseases were most common type of congenital anomalies. Thirty seven to forty weeks gestational age group babies were most commonly have congenital anomalies than other gestational age group babies. Low birth weight babies had higher percentage of congenital anomalies (2.64%). Congenital anomalies were more in the male sex (2.53%) as compared to female babies (1.73%). Maternal obesity, consanguineous marriage and previous family history of congenital anomalies associated with increased risk of congenital anomalies with significant p values.

Conclusions: Incidence of congenital anomalies was 2.15%. Most of congenital anomalies were involved in CNS. Birth weight, Gestational age, Male sex, consanguineous marriage, maternal Obesity and previous family history of congenital anomalies were significantly associated with increased risk of congenital anomalies.

Keywords: Congenital anomalies, Mortality, Neonates

INTRODUCTION

Congenital anomalies represent defects in morphogenesis during early fetal life. Congenital anomalies occur all over the world with similar frequency. According to WHO document of 1972, the term congenital malformations should be confined to structural defects at born.¹ Congenital anomalies account for about 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.²³ With advancements in perinatal and neonatal care, other causes of perinatal mortality have been controlled and as in the west, the time is not far when the leading cause of perinatal mortality would be congenital anomalies, in our country also.

Hence a study on congenital anomalies was undertaken. Evaluation is a continuous process that's why a deep insight into the evolving congenital anomalies and dysmorphology needed. Objectives of our study were to study the pattern of congenital anomalies and to study the association between congenital anomalies and maternal risk factors.
METHODS

The study was conducted at Niloufer Hospital, Hyderabad from November 2017 to November 2018. A detailed antenatal and family history of the mothers was taken.

Inclusion criteria

- Intramural and extramural babies.

Exclusion criteria

- Babies who were still born as unable to find out various anomalies perfectly and cases admitted in a unit after newborn period.

Details regarding birth history were noted. A detailed clinical examination of the newborn was conducted. Though clinical examination is the main criteria for diagnosis appropriate lab and radiological investigations were done when indicated. It is a prospective observational study done in new-born unit, Niloufer Hospital, Hyderabad. The congenital anomalies were analysed in relation to age of the mother, parity of the mother, birth weight of the baby, sex of baby and gestational age of the baby. Influence of risk factors of mothers like obesity, consanguineous marriage on congenital defects also studied. The anomalies were divided according to the major system involved and their frequency was studied.

Statistical analysis

Results were analysed by using simple statistical techniques and methods. Using student’s t-test compared the two groups. More than two groups were compared by ANOVA of F test. P<0.05 was considered significant.

RESULTS

In the present study total number of admissions in the newborn unit were 5198. Total number of babies with congenital anomalies was 112 (2.15%). Central nervous system (CNS) was the most common system involved with 23.32% followed by gastrointestinal system (GIT) and cardiovascular system (CVS) with 17.85 and 16.07% respectively (Table 1).

Table 1: Distribution of congenital anomalies according to major system involved.

| System involved                | Number of cases (112) | %   |
|--------------------------------|-----------------------|-----|
| Central nervous system         | 25                    | 22.32|
| Gastrointestinal system        | 20                    | 17.85|
| Cardiovascular system          | 18                    | 16.07|
| Musculoskeletal system         | 12                    | 10.7 |
| Genitourinary system           | 07                    | 6.25 |
| Syndromes/chromosomal anomalies| 15                    | 13.39|
| Eye                            | 03                    | 2.67 |
| Others                         | 10                    | 8.92 |
| Skin                           | 2                     | 1.78 |

Table 2a and 2b explains the different type of congenital anomalies in different systems. Meningomyelocele, Anorectalomal formations and acyanotic congenital heart diseases were the most common type of anomaly in CNS, GIT and CVS systems respectively.

Table 2a: Different types of congenital anomalies in various systems.

| System affected          | Type of congenital anomaly          | Total no. of anomalies | Percentage for total 112 anomalies |
|--------------------------|-------------------------------------|------------------------|-----------------------------------|
| Central nervous system   | Meningomyelocele                    | 9                      | 8.03                              |
|                          | Hydrocephalus                       | 8                      | 7.14                              |
|                          | Hydrocephalus meningomyelocele microcephaly | 4                  | 3.57                              |
|                          | Meningocele                         | 3                      | 2.67                              |
|                          |                                     | 1                      | 0.89                              |
| Gastrointestinal system  | Anorectanalomalies                  | 5                      | 4.46                              |
|                          | Hepatosplenomegaly                  | 4                      | 3.57                              |
|                          | Cleft lip and palate                | 3                      | 2.67                              |
|                          | Esophageal atresia                  | 2                      | 1.78                              |
|                          | Tracheoesophageal fistula           | 1                      | .89                               |
|                          | Diaphragmatic hernia                | 1                      | 0.89                              |
|                          | Exomphalos                          | 1                      | 0.89                              |
|                          | Umbilical hernia                    | 1                      | 0.89                              |
|                          | Duodenal atresia                   | 1                      | 0.89                              |
|                          | Gastrochisis                        | 1                      | 0.89                              |
| Cardiovascular system    | Acyanotic                           | 14                     | 12.5                              |
|                          | Cyanotic                            | 4                      | 3.57                              |
### Table 2b: Different types of congenital anomalies in various systems.

| System affected                  | Type of congenital anomaly                  | Total no. of anomalies | Percentage for total 112 anomalies |
|----------------------------------|---------------------------------------------|------------------------|-----------------------------------|
| Musculoskeletal system           | CTEV                                        | 3                      | 2.67                              |
|                                  | Limb hypoplasia                            | 3                      | 2.67                              |
|                                  | Polydactily                                | 2                      | 1.78                              |
|                                  | Syndactily                                 | 1                      | 0.89                              |
|                                  | Absent radius and thumb                    | 1                      | 0.89                              |
|                                  | Congenital dislocation of hip              | 1                      | 0.89                              |
|                                  | Genu recurvatum                            | 1                      | 0.89                              |
| Genitourinary system             | Ambiguous genitalia                       | 2                      | 1.78                              |
|                                  | Congenital hydrocele                       | 2                      | 1.78                              |
|                                  | Hypospadias                                | 1                      | 0.89                              |
|                                  | Polycystic kidney                          | 1                      | 0.89                              |
|                                  | Bladder extrophy                           | 1                      | 0.89                              |
| Skin                             | Collodion baby                             | 2                      | 1.78                              |
| Syndromes/chromosomal anomalies  | Down syndrome                              | 4                      | 3.57                              |
|                                  | Pierre robin syndrome                      | 2                      | 1.78                              |
|                                  | Goldenhar syndrome                        | 2                      | 1.78                              |
|                                  | Prune belly syndrome                       | 1                      | 0.89                              |
| Eye                              | Congenital cataract                        | 2                      | 1.78                              |
|                                  | Microphthalmia                             | 1                      | 0.89                              |
| Multiple congenital anomalies    |                                            |                        |                                   |
| Others                           | Congenital cytomegalovirus                 | 2                      | 1.78                              |
|                                  | Hydrops fetalis                           | 1                      | 0.89                              |
|                                  | Congenital rubella syndrome                | 6                      | 5.35                              |

### Table 3: Risk factors and their association with congenital anomalies.

| Risk factor                        | Number of admissions | Number of congenital anomalies | Percentage | P value |
|------------------------------------|----------------------|-------------------------------|------------|---------|
| Maternal age                       |                      |                               |            |         |
| <20 years                          | 2185                 | 36                            | 1.64       |         |
| 20-30 years                        | 2410                 | 65                            | 2.69       | 0.04    |
| >30 years                          | 603                  | 11                            | 1.82       |         |
| Parity                             |                      |                               |            |         |
| <4                                 | 4855                 | 99                            | 2.03       |         |
| ≥4                                 | 343                  | 13                            | 3.79       | 0.03    |
| Birth weight                       |                      |                               |            |         |
| <2.5 kgs                           | 2614                 | 69                            | 2.64       | 0.01    |
| >2.5 kgs                           | 2584                 | 43                            | 1.66       |         |
| Sex                                |                      |                               |            |         |
| Male                               | 2484                 | 63                            | 2.53       | 0.04    |
| Female                             | 2712                 | 47                            | 1.673      |         |
| Parental consanguinity             |                      |                               |            |         |
| Present                            | 380                  | 24                            | 6.31       | 0.03    |
| Absent                             | 4936                 | 88                            | 1.78       |         |
| Maternal nutrition                 |                      |                               |            |         |
| Nourished                          | 3359                 | 81                            | 2.41       |         |
| Under nourished                    | 1409                 | 21                            | 1.49       | 0.04    |
| Obese                              | 428                  | 9                             | 2.1        |         |
| History of any previous anomaly    |                      |                               |            |         |
| Present                            | 74                   | 7                             | 9.7        | 0.02    |
| Absent                             | 5122                 | 105                           | 2.04       |         |
Table 3 explains about the risk factors in causing congenital anomalies all the risk factors significantly associated with congenital anomalies with p value <0.05. Birth weight <2.5 kg was more significantly associated risk factor with p value 0.01. Maternal age, male sex and obesity were less significant associated risk factors with p value 0.04.

Table 4 explains the anomalies according to gestational age, neonates born between 37-40 weeks were most commonly associated may be due to large births in this gestational age.

**Table 4: Frequency of congenital anomalies according to gestational age.**

| Gestational age | No. of congenital anomalies | Percentage |
|-----------------|----------------------------|------------|
| <28 weeks       | 4                          | 3.57       |
| 29-33 weeks     | 10                         | 8.92       |
| 34-36 weeks     | 39                         | 34.82      |
| 37-40 weeks     | 44                         | 39.28      |
| >40 weeks       | 15                         | 13.39      |

**DISCUSSION**

During this study period there were 5198 newborns admitted to the newborn unit, out of these 5198 babies, 112 babies were detected with congenital anomalies and the percentage was 2.15 this is comparable to another studies Tisane A et al, Shamma M et al and Shatanik Sarkar et al they showed as 1.9%, 2-3%, 2.2% respectively.2,4,5

The congenital anomalies were analyzed in relation to age of the mother, parity of the mother, birth weight of the baby, sex of the baby, gestational age of the baby, history of consanguineous marriage, previous history of any anomaly and maternal nutrition.

The congenital anomalies were divided according to the major systems involved and their frequency was studied. In babies born to mothers aged between 20-30 years, percentage of congenital anomalies was high 2.69% and in maternal age above 30 years 1.82% as compared to the percentage of congenital anomalies seen in newborn to the mothers with age groups of <20 years 1.64%. Similar results are obtained by Dr. Taksande A et al and Arjun Singh, et al.2,6

They also have reported that congenital anomalies more in >30 age group. Among the 4855 newborns of less than 4th birth order congenital anomalies were seen in 99 babies with percentage of 2.03. Out of the 343 newborns of 4th and above birth order congenital anomalies were a percentage of 3.79.

Though most of the studies stated that parity of the mother does not seen to influence the incidence of congenital anomalies a higher percentage of congenital anomalies were seen in the birth order of 4 and above in the present study this is similar to other studies like, Dr. Swain et al, R. Kulshrestha et al, Chaturvedi et al, and Grover N. et al.7,10

When congenital anomalies were analyzed with respect to birth weight of the baby higher percentage of congenital anomalies was seen in low birth weight (<2.5kgs) group of babies. In the low birth weight babies (2614), congenital anomalies were seen in 69 babies with percentage of 2.64. Among the babies weighing <2.5 kgs (2584), congenital anomalies were seen in 43 babies with a percentage of 1.66. The higher percentage of congenital anomalies seen in low birth weight babies in the present study and also seen in most of the studies like Taksande A et al, Swain et al, Grover N et al, Vikream Dutta et al, and Bhat and Babu et al.2,7,10,11,12

In the present study out of 5198 admissions male babies were 2484, congenital anomalies were seen in 63 male babies and the percentage was 2.53. Number of female babies were 2712, among these congenital anomalies were seen in 47 female babies with a percentage of 1.73. The higher percentage of congenital anomalies were seen in male babies is also reported by other studies like Dr. Taksande et al, Dr. Shatanik Sarkar et al, Dr. Vikram Dutta et al, Dr. Hemaranjani et al, Dr. Malla B.K. et al, and Fatema K. et al.2,5,11,13-15

Out of 112 cases with congenital anomalies, highest number involved between 37-40 wks. of gestational age with percentage of 41.96 than 34-36 wks. 31.25%. Fatema K et al, have reported that congenital anomalies are highest between 34-36 weeks. of gestational age group.

In the present study highest number of congenital anomalies involved CNS with total 25 cases (22.32%). This frequency also reported by Swain et al, Gupth S et al, Kalaiselvan G et al and Pandya M et al. 7,13,16,17 meningo(myelo)cele was the commonest CNS anomaly detected with a percentage of 8.03(9 cases). Hydrocephalus was seen in 8 babies (7.14%). The high percentage open neural tube defects is similar to reports made by Chaturvedi P. et al and Hemaranjani et al.6,18

Cassell and Golden reported maternal obesity associated with more congenital defects in neonates in our study also we had results of significant association with p value 0.04.19 The role of parental consanguinity for the development of CAs has been addressed by other studies.20,21 Congenital rubella syndrome was seen in 6 cases with percentage of 5.35 this result was similar to other studies like Chandy et al showed that the proportion of suspected cases that were laboratory confirmed increased from 4% to in 2000 to 11% in 2008, Singh S et al have reported that 5% of cases were clinically confirmed and P.15 Singh et al reported anti rubella IgM positive in 2.5%of cases in 0-28 days of age group.14,22
CONCLUSION

Congenital anomalies were higher in babies to mothers between 20-30 years of age (2.69%) and elder mothers with >30 years of age 1.82%. The percentage congenital anomalies was higher in babies of birth order 4 or more than 4 with 3.79%, as compared to babies of birth order <4- 2.03%. Low birth weight babies (2.5 kgs) had higher percentage of congenital anomalies-2.64%. Congenital anomalies were more in the male sex 2.53% as compared to female babies- 1.73%, ambiguous genitalia were seen in two babies. The distribution of the anomalies system wise was in the following order-central nervous system, GIT, CVS, Musculoskeletal system, genitourinary system and skin. Syndromes/chromosomal anomalies were seen in 9 cases with 8.03%. Multiple congenital anomalies were seen in 7 cases 6.25%. Congenital rubella syndromes were seen in 6 cases with 5.35%. Polyhydromnios was present in 2 cases of gastrointestinal tract malformations. Drug intake, radiation exposure, nutritional deficiencies were not seen in any of the congenital anomalies. Incidence of congenital anomalies was 2.15%. Most of congenital anomalies were involved in CNS. Low birth weight babies were commonly affected with male preponderance. Maternal age, nutrition and previous family history of congenital anomalies were also risk factors for congenital anomalies.

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