

## Clinical Study on Congenital Malformations At Birth in A Tertiary Level Hospital in North-East India

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**Abstract:** This was a clinical descriptive study of congenital malformations in new born in a tertiary level hospital in north-east India from the period 1<sup>st</sup> January 2015 to 30<sup>th</sup> April, 2016. Out of 14912 births, 159 had congenital malformation making an incidence of 1.06%. The incidence was more in males, in preterm babies, and more in stillborn. Oligohydramnios was associated with the highest risk of congenital malformation. The most common congenital malformation was cleft lip followed by CTEV.

**Keywords:** Birth, Congenital, Incidence, Malformation, Stillborn

### I. Introduction

Congenital malformation represents defects in morphogenesis during early foetal life. According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth. The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations. Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India.<sup>1</sup> Prevalence studies are important to establish baseline rates, to document the changes that take place over the period of time, and to find out the aetiology. Different registration systems of infants, international organisations, worldwide monitoring and surveillance have been established to enumerate the possible causes and prediction of occurrences of congenital malformations and to establish prevention strategies.<sup>2</sup> Congenital malformation will begin to emerge as one of the major childhood health problems. Treatment and rehabilitation of children with congenital malformations is costly and complete recovery is usually impossible. Approximately, 66% of major malformations have no recognized aetiology and most of them have multifactorial inheritance. These defects can occur for many reasons including inherited genetic conditions, poor diet, toxic exposure of the foetus for example, to alcohol, birth injury and, in many other cases, for unknown reasons. In the present study, we try to determine the overall incidence, types and distribution of various congenital anomalies both in live born and still born foetuses, and also to find out the major associated maternal and foetal associated risk factors.

### II. Materials And Methods

This was a descriptive, cross sectional study of newborns and stillborn babies delivered at Regional Institute of Medical Sciences, Imphal, from the period 1<sup>st</sup> January 2015 to 30<sup>th</sup> April, 2016. Data collection was done in the form of structured form which contained details about variable like date of admission, age, history of chronic illness, drug ingestion, exposure to X-ray, history of consanguineous marriage in other offspring, etc. and that of the baby like live or still birth, gestational age, birth order, existence of congenital anomaly and the type of it. No autopsy examinations were performed. There were a total of 14789 live births and 123 still births during this period. The study material comprised 14912 births (both live and still) and their 14773 mothers(139twin deliveries). Congenital malformations were divided into central nervous system (CNS), musculoskeletal, gastrointestinal, genitourinary, cardiovascular (CVS), syndromes, and miscellaneous disorders.

### III. Results

Out of the total 14912 babies delivered, 14789 were live births and 123 were stillbirths. The number of babies with congenital malformations diagnosed at birth or within the first week of life was 159(1.06%).TABLE 1 gives the frequency and sex distribution of congenital malformations. Out of the 14634 singleton births, 157

(1.07%) were malformed, whereas 2 of the 139 pairs of twins had birth defects. The sex-wise distribution was 56% males and 44% females, giving an M:F ratio of 1.28:1, and there were three cases of ambiguous genitalia. Congenital malformations were seen more in stillbirths as compared to live births, the frequency being 6.50% and 1.02%, respectively. Eight of the 159 malformed babies (5.03%) were still born. TABLE 2 shows the frequency of congenital malformations in relation to foetal and maternal factors. Women less than 20 years had 3.11% babies with congenital anomalies, and the mothers of babies with congenital anomalies were mostly between 20 and 30 years, i.e. 66%, and 31% of the mothers were above 30 years.

**Table 1**

|              | Total Cases | Malformed Babies | Percentage |
|--------------|-------------|------------------|------------|
| Total Births | 14912       | 159              | 1.06       |
| Still Births | 123         | 8                | 6.50       |
| Live Births  | 14789       | 151              | 1.02       |
| Male         | 8325        | 98               | 0.85       |
| Female       | 6584        | 53               | 1.24       |
| Ambiguous    | 3           | 0                |            |

**Table 2**

| BIRTH WEIGHT(GRAMS)  | FOETAL FACTORS |                    | PERCENTAGE |
|----------------------|----------------|--------------------|------------|
|                      | TOTAL CASES    | MALFORMED PATIENTS |            |
| <1000                | 37             | 32                 | 20.12      |
| 1000-15000           | 205            | 34                 | 21.38      |
| 1501-2000            | 254            | 35                 | 22.01      |
| 2001-2500            | 356            | 19                 | 11.94      |
| >2500                | 14060          | 39                 | 24.52      |
| GESTATIONAL AGE      |                |                    |            |
| PRETERM              | 3897           | 97                 | 61.00      |
| TERM                 | 10420          | 59                 | 37.10      |
| POSTTERM             | 456            | 3                  | 1.88       |
| MATERNAL FACTOR      |                |                    |            |
| MATERNAL AGE (YEARS) |                |                    |            |
| <20                  | 289            | 9                  | 5.66       |
| 21-25                | 2098           | 44                 | 27.67      |
| 26-30                | 7708           | 67                 | 42.13      |
| >30                  | 4678           | 37                 | 23.27      |
| PARITY               |                |                    |            |
| PRIMI                | 1089           | 43                 | 27         |
| PARA1-3              | 10898          | 90                 | 56         |
| PARA 4 AND MORE      | 2786           | 26                 | 17         |
| MODE OF DELIVERY     |                |                    |            |
| LSCS                 | 4589           | 36                 | 77         |
| VAGINAL DELIVERY     | 10184          | 123                | 23         |

History of parental consanguinity was present in 4 cases of congenital malformations in our study. Babies with congenital anomalies were of the first order (7.30%), and second order to third order (73.08%). More than four or fourth birth order was associated with 18.16% of the anomalies. There was a history of oligohydramnios in 45 cases and polyhydramnios in 23cases. Also, 23/159 mothers (14.47%) had a history of previous abortions, 6/159 (3.77%) were diabetic mothers and 5/159 (2.79) had a history of congenital heart disease in previous child or malformed babies. TABLE 3 shows the systemic distribution and the incidence of individual congenital malformations. Gastrointestinal malformations were the most common in live births, followed by musculoskeletal system malformations. The incidence of CNS malformations was higher in the stillborn.

**Table 3**

| TYPE OF DEFECT              | TOTAL NUMBER |
|-----------------------------|--------------|
| Musculoskeletal             | 39           |
| Ctev                        | 25           |
| Polydactyly                 | 12           |
| Arthrogryposis              | 1            |
| Hemimelia                   | 1            |
| Gastrointestinal            | 45           |
| Cleft Lip                   | 29           |
| Cleft Palate                | 15           |
| Gastroschisis               | 1            |
| Central Nervous System      | 32           |
| Hydrocephalous              | 24           |
| Meningomyelocele            | 2            |
| Spina Bifida                | 3            |
| Encephalocele               | 3            |
| Genitourinary System        | 21           |
| Hypospadiasis               | 9            |
| Hydrocele                   | 5            |
| Ambiguous Genitalia         | 3            |
| Micropenis                  | 2            |
| Undescended Testis          | 2            |
| Skin                        | 5            |
| Acessory Nipples            | 1            |
| Haemangioma                 | 4            |
| Cardiovascular System       | 10           |
| Acyanotic Heart Disease     | 1            |
| Cyanotic Heart Disease      | 9            |
| Respiratory System          | 2            |
| Diaphragmatic Hernia        | 1            |
| Tracheosophageal AtresiaEye | 12           |
| Anophthalmia                | 1            |
| Congenital Cataract         | 1            |
| Syndrome                    | 3            |
| Down's Syndrome             | 2            |
| Turner's Syndrome           | 1            |



**Figure showing a baby with bilateral Anophthalmia**

#### **IV. Discussion**

Congenital malformations are an important cause of perinatal mortality and childhood morbidity. The number of birth defects is increasing antenatally and during the neonatal period due to advanced diagnostic technology, especially USG.<sup>1</sup> The pattern and prevalence of congenital malformations may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors, including socio-cultural, racial and ethnic factors.<sup>4</sup>

In the present study the incidence of congenital malformation was 1.06% which is comparable with earlier studies from same part of the country<sup>5</sup>. There are other reports from different parts of the world representing different frequency of congenital malformations<sup>2</sup>. Although we got nearly the same result as reported in other studies, the prevalence of congenital anomaly have been more than the present rate, if it was a community based study and not merely in a tertiary care setup.

With regard to the pattern of congenital anomalies in the study, the most common system involved was GIT (gastrointestinal system) (28.30%), followed by musculoskeletal(24.53%), CNS(central nervous system)(20.13%), Genitourinary system(13.21%), CVS(Cardiovascular System)(6.29%), Skin(3.14%), Eye(1.26%), Respiratory System(1.26%), syndromes(1.26%).The prevalence of cardiovascular malformations was found to be low in this study. The results conform to studies conducted in same part of the country<sup>5</sup>. Some studies from other parts of the country have reported higher prevalence of CVS malformations<sup>1</sup>, musculoskeletal malformations<sup>6,5</sup> where as some studies have reported higher prevalence of CNS malformations<sup>2,7</sup>.

More male babies were born with congenital malformations than females which is consistent with other studies<sup>1,4</sup>. It may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life.

Taksande et al<sup>1</sup>, Parmar et al<sup>3</sup>, Baruah et al<sup>5</sup> found increased prevalence of congenital malformations in preterm babies. In this study also, the prevalence of congenital malformations were much higher in preterm babies (61%). Sarkar et al<sup>4</sup> reported a higher prevalence of congenital malformations in low birth weight babies which is in accordance with our study (75%). In this study the highest prevalence of congenital malformations was found in the maternal age group of (26-30) years. Taksande et al<sup>1</sup>, Datta et al<sup>9</sup> found increased prevalence of congenital malformations in maternal age group of 35 years or more, where as Sarkar et al<sup>4</sup> higher congenital malformations in the maternal age group of 25-29 years which is in accordance with our study.

Previous studies<sup>1,4</sup> have demonstrated increased incidence of congenital malformations with increasing birth order, but in this study the highest prevalence of congenital malformations were found in the parity1 to parity3.

This study found increased prevalence of congenital malformations in the babies that were delivered vaginally which is in accordance with other studies<sup>3</sup>. History of consanguineous marriage was found in 4 cases in this study where as some studies have reported increased incidence of malformations in consanguineous marriages (double risk)<sup>8</sup>.

## **V. Conclusion**

Congenital malformations are a major cause of still births and infant mortality which must be identified and surgical correction attempted, if possible. There should be widespread education in the community regarding the common congenital malformations, their outcomes and possible available modes of treatment.

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