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Spectrum of Congenital Malformations in Neonates at a Rural Teaching Hospital — A Cross Sectional Observational Study

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Abstract

Introduction: The global estimates of congenital anomalies in neonates are 6% and few of them are severe enough to cause death. According to World Health Organization (WHO), congenital anomalies attribute to 17-42% of the infant mortality. Apart from causing death, they also contribute to preterm births, childhood and adult mortality, with significant repercussions in families. With the advancement of technology, there has been a decrease in the number of deaths due to other causes and there has been an increasing concern about congenital anomalies. This calls for an inquiry into the recent burden of congenital anomalies and their associated risk factors. Hence, the study was carried out. **Objectives:** 1. To assess the frequency and pattern of congenital anomalies. 2. To determine the factors associated with congenital anomalies. **Materials & methods:** This is an observational study which included all live born neonates with congenital anomaly/ies admitted to R L Jalappa Hospital (RLJH) and still born neonates or aborti with congenital anomaly/ies delivered in RLJH during the study period. A detailed history of the study participants was taken, and all the anomalies were coded as per the ICD coding system. For still born babies, aborti and neonatal deaths, infantogram, gross autopsy and histopathological examination findings were noted. **Statistical analysis:** Data was analysed using "Microsoft excel sheet" and the analysis was done using Statistical Package for Social Sciences (SPSS-16) software. Significance was defined as $p < 0.05$. **Results:** Our hospital had 2,400 deliveries during the

study period, out of which the frequency of congenital anomalies was 1.3%. As per the associated risk factors, 66.6% of the babies had no associated risk factors while the remaining 33.4% of the babies had an associated risk factor. Most commonly seen risk factor in the study was 3rd degree consanguinity (11.1%). As per the system involved, Musculoskeletal system involvement was seen in the majority (63.9%) of the neonates, followed by Cardiovascular system in 11.1%, Central Nervous System (CNS) in 8.3%, Genital system in 8.3%, Lymphatic system in 5.6%, Gastrointestinal system in 2.8%, Cutaneous in 2.8%, Oral cavity in 2.8% and syndromic anomaly in 2.8%. **Conclusion:** The prevalence of congenital anomalies is considerably high and increasing the awareness to prevent them is the need of the hour. Appropriate consideration should be given to reducing the risk factors and genetic counseling should be provided to parents with high risk.

Keywords: Congenital malformations; Neonates; Congenital anomalies

Introduction

Congenital malformations are defined as structural or functional abnormalities of prenatal origin which are present at birth¹. They are also called congenital disorders, birth defects, or congenital anomalies².

The global estimate of congenital anomalies in neonates is 6% and few of them are severe enough to cause death³. The deaths are approximated to be 303,000 new borns per year. According to World Health Organization (WHO), congenital anomalies contribute to 17% – 42% of Infant Mortality⁴. As per the Global Burden of Disease Study 2013, it is among the top ten causes of deaths below five years of age⁵. In India, congenital malformations were found to be the fifth largest cause of neonatal deaths⁵.

The etiology can be pre-conceptual, mainly genetic, or can occur during or after conception, which is most likely due to environmental factors. Multifactorial

inheritance is said to be the underlying culprit in many cases⁶.

The first four major causes of neonatal deaths in India are preterm births, intra partum complications, pneumonia and neonatal sepsis. With the advancement of technology and science, there seems to be a decrease in mortality due to the other causes, and hence an increasing concern about congenital anomalies⁵.

This transition propounds the need for sufficient data on the frequency and risk factors of congenital defects. The burden of birth defects has been described extensively in recent years worldwide^{4,6-11}. However, there is a paucity of recent literature in India as most of the published reports are more than 5 years old^{5,12-14}.

About 60% of the malformations can be prevented through proper education of the mother and by following timely preventive measures^{9,15,16}. Therefore, it is vital to understand the most common factors associated with congenital malformations.

Objectives of the study

- To assess the frequency and pattern of congenital anomalies.
- To determine the factors associated with congenital anomalies.

Materials and methods

- **Study area:** Newborns born in R L Jalappa Hospital (RLJH) in Tamaka, Karnataka, and extramural neonates admitted in RLJH during the neonatal period.
- **Study period:** January 2020 to June 2021 (18 months)
- **Type of study:** Cross-sectional observational study.
- **Inclusion criteria:** All live neonates (intramural and extramural) with congenital anomaly/ies admitted in RLJH and still born neonates or aborti with congenital anomaly/ies delivered in RLJH during the study period.
- **Exclusion criteria:** Babies of parents who have not provided consent.
- **Sample size and sampling techniques:** All neonates satisfying the inclusion criteria admitted in RLJH during the study period were included after getting consent from the parents.
- **Methods and Methodology:** Approval of the institutional ethics committee was taken prior to starting the study.

Post-delivery, after thorough hand washing, the neonate was assessed, and the basic steps of essential newborn care was performed. The newborn was examined systemically from head to toe, and all the findings were noted. Similarly, any outside-born (extramural) baby admitted in RLJH was examined systemically, and findings were noted. If the newborn was found to be having a structural birth defect, informed consent of the parent was taken, and the neonate was included in the present study. A detailed history of the neonate was taken, including family history, antenatal history, birth history and risk factors, and entered in the study proforma. Findings of the antenatal scan were also noted. Relevant interventions were done on a per needful basis.

Statistical analysis

Data was analyzed using "Microsoft excel sheet" and Statistical Package for Social Sciences (SPSS-16) software. The analysis involved preliminary data entry, content analysis, and interpretation. Percentages were used to analyze epidemiological variables, and mean, standard deviations were calculated. Based on the data collected, statistical analysis was performed using the chi-square or Fisher exact test. Significance was defined as $p < 0.05$.

Results

The study was conducted at R L Jalappa Hospital and Research Center (RLJH&RC). Out of 2400 deliveries in our hospital during the study period, 33 neonates had congenital anomalies (Intramural). There were 3 neonates (extramural and admitted in RLJH during the neonatal period) with congenital anomalies. A total of 36 neonates were included in the study.

The 36 neonates in our study included: 34 Live babies, 1 Still birth, 1 Abortus

In our study, there was a male predominance of 61.1%. According to the gestational age, 81% of the neonates were delivered at term gestation (37 weeks to 41 weeks 6 days), whereas, 11% of the neonates were delivered late preterm (34 weeks to 36 weeks 6 days), 5% of the neonates were extreme preterm (< 28 weeks) and 3% were delivered post term (> 42 weeks). Among the mothers, 41.7% of the mothers were primiparous, 41.7% were P2 and 16.7% of the mothers were P3.

History of serious maternal illness was noted in 13.9% of the mothers, of which, diabetes was majorly associated (8.4%). One previous abortion was noted in 8.3% of mothers, while two previous abortions were noted for 5.6% of mothers. The remaining 86.1% of the mothers had no history of previous abortions. None of the mothers in the study group had a history of still birth.

In our study participants, 2.8% had a congenital anomaly in the siblings, while 5.6% of the study participants had a history of congenital malformation in the family, excluding siblings.

The majority of the anomalies in our study were single anomalies (75%), whereas 25% of the neonates had multiple anomalies.

Among our study participants, 66.6% had no associated risk factors. The remaining neonates (33.4%) had at least one associated risk factor. Among the associated risk factors, third degree consanguinity accounted for 11.1% of the neonates, while second degree consanguinity accounted for 8.3% of the neonates. Diabetes was seen in three of the mothers, while one mother had a bicornuate uterus. There was oligohydramnios in one pregnancy, and one of the mothers had hepatitis B infection. This association was statistically significant ($p < 0.001$) — Table 1.

As per the system wise distribution, Musculoskeletal system accounted to majority of the anomalies (63.9%), followed by Cardiovascular system (11.1%), Central Nervous System (CNS) (8.3%), Genital (8.3%), Lymphatic (5.2%), Cutaneous (2.8%), Gastrointestinal (2.8%), Oral cavity (2.8%) and Syndromic (2.8%) — Table 2.

Table 1. Association Between Overall Risk Factors and Presence of Anomalies

Parameters	Risk Factors (n = 36)	p value
Risk Factor***	Frequency (%)	<0.001²
None	24 (66.6)	
3rd Degree Consanguinity	4 (11.1)	
2nd Degree Consanguinity	3 (8.3)	
Diabetes in Mother	2 (5.6)	
Bicornuate Uterus in the Mother	1 (2.8)	
Diabetes in Mother + HepB	1 (2.8)	
Oligohydramnios	1 (2.8)	

Table 2. System-wise Involvement of Anomalies Among Study Participants (n=36)

Anomalies (System-wise)	Frequency
Musculoskeletal	23
Cardiovascular	4
CNS	3
Genital	3
Lymphatic	2
Gastrointestinal	1
Cutaneous	1
Oral cavity	1
Syndromic	1

Discussion

Congenital anomalies in neonates are considered as one of the major causes of neonatal deaths today. The underlying causes of these anomalies continue to remain obscure, and over 50% of them are of unknown etiology⁹. Investigating the risk factors can help us to comprehend the patterns and help in prevention.

In the present study, the frequency of congenital anomalies among intramural neonates during the study period of 18 months was 1.3%. A study conducted in Iran by Sedighi I et al. showed a prevalence of 0.85%¹¹, while another study conducted in Morocco had a prevalence rate of 1.02%⁹. The difference in the prevalence in various studies could be attributed to population differences and risk factors.

In our study, there were a total of 36 cases, of which 34 (94.4%) were live born neonates, one was an abortus at 15 weeks of gestation (2.8%) and the other was a still birth at 23 weeks of gestation (2.8%).

The majority (81%) of the babies in our study were born at term. In a retrospective study done in Ethiopia, around 82% of the babies with congenital anomalies were born at term which is similar to our study⁴. Preterm babies with congenital anomalies were present in 8.3% of neonates in our study.

In contrast, a retrospective study done in Morocco showed 17.4% of babies with anomalies remains preterm⁹. In a case control study from Ethiopia by Abebe S et al, among 251 cases with congenital anomalies, preterms constituted 47.6%⁸. The association between congenital anomalies and gestational age remain unknown.

The majority (86.1%) of the mothers in our study had no history of serious illness and the remaining mothers (13.9%) had an associated serious illness. The most common maternal illness noticed was gestational diabetes in the mother (8.4%). The results of the present study were in conformity to a study published in 2020, to detect birth defects in women with diabetes, where gestational diabetes was found in 5.3% of mothers¹⁷. In another case control study done in South-western Ethiopia, Diabetes was found in 1.6% of neonates with congenital malformations⁸. The findings of these studies were in close proximity to the ones in our study.

Among siblings of our study participants, 2.8% had anomalies. Research done to find out about the lethal congenital malformations in North India showed 25% of babies with congenital malformation in siblings¹⁸. The wide difference seen could be because our study involved all anomalies, both minor as well as major, and their study included only lethal malformations.

Among the family members of our study participants, excluding siblings, 5.6% had congenital anomalies and 94.4% had no congenital anomalies. Another study was conducted using case control model. In their study, among controls, 1.7% had a family history of congenital anomalies, as opposed to 3.8% in that of cases⁸. Even though our study had a slightly higher association of family history with congenital anomalies, the difference is not significant enough.

Among the study participants of our study, 38.9% had risk factors. The most common risk factor was seen in 11.1% of the babies, which is 3rd degree consanguinity. Followed by that, the next common risk factor was 2nd degree consanguinity seen in 8.3% of the babies. In a study done in North India to study the lethal malformations, 10.5% of the parents had consanguinity¹⁸. Our study was conducted in South India. The concept of consanguineous marriages is more in South India compared to North India, which could have attributed to higher percentage of consanguinity in our study¹⁹.

Our study involved the following systems in order.

Musculoskeletal system — 63.9%, Cardiovascular — 11.1%, CNS — 8.3%, Genital — 8.3%, Lymphatic — 5.2%, Cutaneous — 2.8%, Gastrointestinal — 2.8%, Oral cavity — 2.8%, Syndromic — 2.8%

There is a wide variation among different studies based on the system involved.

In few studies, Nervous system was most commonly involved^{4,7-9,18,20,21}.

Few other studies had predominantly Musculoskeletal system involvement^{12,14,18,22}.

One study had a predominant Cardiovascular system involvement⁵.

Most of the studies had either the nervous system or musculoskeletal system as the most common abnormality. Our study also had musculoskeletal cases in the majority and among CNS, we had only sacral dimple as opposed to many other studies which had many other cases like anencephaly or holoprosencephaly or neural tube defects among CNS disorders^{7,8}.

There are few possible explanations for this. In our study, all the mothers had a history of folic acid intake. Hence, this greatly reduces the chances of neural tube defects. The exclusion criteria in our study are those neonates where consent could not be obtained. Most of the CNS anomalies are not compatible with life and there was a need for an autopsy on these infants. There was difficulty in obtaining consent for these babies, hence they had to be excluded from the study population. There could have been more prevalence of lethal anomalies, especially nervous system anomalies in reality.

Limitations

- Genetic analysis of neonates could not be done
- Infant gram and autopsy for a few neonates were missed due to inability to obtain parental consent.

Conclusion

The frequency of congenital malformations in our study was 1.3% of live born neonates, during the study period. The majority of them involved the musculoskeletal system, followed by the cardiovascular system and CNS involvement.

The major factors associated with congenital anomalies were 3rd degree consanguinity, followed by 2nd degree consanguinity.

Other risk factors found to be associated with congenital anomalies were diabetes in the mother, bicornuate uterus, oligohydramnios, Hepatitis B infection. The association between the risk factors and the presence of a congenital anomaly was found to be statistically significant.

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