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## Original Research Article

## Study of congenital malformations in newborn

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## ABSTRACT

First trimester in pregnancy is majorly about “Organogenesis”, formation and development of embryo, and then progression to fetus. Insult to the stages of this organogenesis leads to various anomalies; including malformations. Lack of nutrition, folate deficiency, genetic factors, teratogenic drugs or environmental exposure can lead to such malformed embryo/fetus. Viability of such fetus depends on the system affected, and the severity. Many fetus do not survive major defects, but some may reach to the stage of live births, causing major problems for survival, growth and development. The psychological impact on the couple, along with future financial strain on possible treatments are additional burdens. There have been few investigations, now made available for diagnosing many of such malformations in earlier stages of pregnancy; making us able to detect them, so that with proper counselling of the couple and possible outcome, we can take decisions on continuation or termination of pregnancy.

This was a prospective study, where we studied 200 newborns who were born with congenital malformations, during time period of January 2017 to December 2019. There were total 15687 deliveries in the said time period, making the incidence of congenital malformations to be 1.27%. Most commonly involved system was central nervous system (39%), followed by musculoskeletal system (25%), followed by gastrointestinal system (11%).

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## 1. Introduction

Congenital malformations are defects which can be functional, morphological, biochemical or molecular. Genetic and congenital malformations are majority of the times incurable; but can be diagnosed at an early stage with advancing medical investigations nowadays. Many such malformations need detailed study of past obstetric history, past medical events, presence of medical disorders, family history and even history of possible exposure to teratogenic substances/modalities. Genetic counselling, along with proper investigative modalities may assist in

taking the decision of continuation or termination of pregnancy. With improving survival of neonates, congenital malformations still hold a major threat to the well being and growth and development of the newborns. Neonatal morbidity and mortality would be majorly contributed by such defects with major fall in cases of prematurity and sepsis.

This study majorly focuses on pattern of congenital malformations encountered. The aim is to focus on the systems majorly affected, so that better understanding of causative factors can be brought in light; for further prevention in forthcoming times. Viable or non-viable, these defects do bring a lot of burden on the lives of neonates themselves, along with psychological impact on the couple,

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with financial burden of potential, yet costly management options.

## 2. Materials and Methods

The study was conducted from January 2017 to December 2019. Detailed history including maternal and paternal age, consanguinity, gestational age, detailed obstetric history, history of exposure to teratogenic substance/drug, significant medical history was taken. Details of any previously affected child was also taken. Detailed examination of babies were carried out. Malformed babies who were stillborn, were also examined thoroughly. Newborns were examined in relation to expected gestational age, and their birth weights were noted. Ultrasonography was done in patients who did not have previous report, and presented as emergency patients. X ray was done in few cases of skeletal disorders post-natally. Not all the investigations were possible due to factors like, reference from private hospitals/health centres, late arrival with labour pains, and in few cases, non-cooperation of patient/relative. Malformations with systems involved were noted.

## 3. Results

There were total 15687 deliveries in the said time period, making the incidence of congenital malformations to be 1.27%. Most commonly involved system was central nervous system with 78 cases (39%), followed by musculoskeletal system with 50 cases (25%), followed by gastrointestinal system with 22 cases (11%). Involvement of genitourinary system was seen in 20 cases (10%), cardiac malformations were seen in 11 cases (5.5%), multiple systems were involved in 19 cases (9.5%). (Table 1)

Within the study time period, total live births were 15062, out of which, 117 had congenital malformations (0.77%). There were 625 stillbirths, out of which 83 had congenital malformations (13.28%).(Table 2)

Out of 200 congenital malformed babies delivered, 75 were fullterm (37.5%) and 125 were preterm (62.5%). (Table 3)

## 4. Discussion

Incidence of congenital malformations was 1.27% in this study. There have been studies conducted in our country with nearly same incidence.<sup>1-3</sup> One more study published by Sharma et al, in 2018 had incidence of 2.48%, which was almost double than the present study.<sup>4</sup>

Many of the factors such as polyhydramnios-oligohydramnios, maternal pyrexia, mainly in early pregnancy, maternal diabetes-thyroid disorders-hypertension, previous child/abortion with birth defects increase chances of congenital malformations in current pregnancy.<sup>5</sup>

The incidence noted was much higher in China (5.6%) and USA (3%).<sup>6,7</sup> The probable reason would be under-reporting of many of the malformations in our country. The birth defect registry should be consolidated to detect every malformation, in turn to reduce future neonatal morbidity and mortality.

Most commonly affected system was Central Nervous System, which was involved in 78 cases. Similar frequency was found in studies conducted by Grover in Shimla.<sup>8</sup> (Table 1)

**Table 1:** Involvement of systems

System Involved	No. of cases	%
Central Nervous System	78	39
Musculo-skeletal system	50	25
Gastro-intestinal system	22	11
Genito-urinary system	20	10
Cardiac system	11	5.5
Multiple system involved	19	9.5
Total	200	100

**Table 2:** Live births and still births

Births	Total	Malformed newborns	Percentage
Live	15,062	117	0.77%
Stillbirth/Intrauterine	625	83	13.28%
Fetal Death			
Total	15,687	200	1.27%

**Table 3:** Gestational age/maturity

Maturity	Total Deliveries	No. of malformed cases	Percentage
Full Term	12,209	75	0.61%
Preterm	3478	125	3.59%

Folic acid deficiency has been described as major cause for neural tube defects, thus necessitating folate supplementation starting at least 100 days before the planning of pregnancy.

In present study, Anencephaly (15 cases), Meningocele (14 cases), Hydrocephalus (13 cases) were majorly seen cases amongst those affected with CNS Defects.

Musculoskeletal system was involved in from of defects such as cleft lip and/or cleft palate (15 cases), talipes equino varus (9 cases). 50 babies born in the study time period had Musculo-skeletal system affected.

22 babies were born with gastro-intestinal defects. Omphalocele, Tracheo-esophageal fistula, congenital diaphragmatic hernia were the common defects.

Out of 20 cases involving genito-urinary system, major defect seen was: dysplastic kidneys. All the babies were born live and were referred to pediatric surgery department.

Out of total 200 babies with malformations, 3 cases were diagnosed with Down's Syndrome and 1 case with Backwith Widman Syndrome. All these 4 cases did not have any prior investigation done and presented as emergency patients.

Incidence of congenital malformations was higher in those who were stillborn as compared to those who were born alive. (13.28% vs 0.77%). Majority of the deliveries were preterm as compared to full-term (62.5% vs 37.5%). Thus, it signifies the impact of such malformations on viability as well as nature's mechanism to expel such conceptus even in early pregnancy, i.e., first and second trimester.

## 5. Conclusion

Obstetrics has made advances in detecting many of the anomalies in the modern world. Much of neonatal morbidity and mortality can be prevented by even basic steps of proper nourishment, folate supplementation, pre-pregnancy workup, ruling out of medical disorders, avoiding teratogenic drugs, and even genetic counselling in the couple having high risk for developing anomalous fetus. We have achieved significant reduction in maternal as well as perinatal morbidity and mortality; and yet, we have scope of improvement in from of reduction in congenital malformations.

## 6. Source of Funding

None.

## 7. Conflict of Interest

None.


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