

# A study on Prevalence of Congenital Anomalies in Neonates in a Tertiary Care Hospital.

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## ABSTRACT

**Background:** Congenital anomalies are a major cause of stillbirths and neonatal mortality. The pattern and prevalence of congenital anomalies may vary over time or with geographical location. The aim of this study is to determine the proportion and types of congenital anomalies in live newborns and to study maternal and perinatal risk factors. **Methods:** This prospective study was conducted over a 1 year period (March 2014 to February 2015) in the department of Pediatrics and Obstetrics/Gynaecology at Patliputra Medical College, Dhanbad, Jharkhand during the period of May 2014 to April 2015. All the live born babies born in this hospital during this period were included. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic variables. **Results:** During the study period, 8546 babies were born, of which 246 had congenital malformations, making the prevalence 2.19%. Most of the women (57.8%) belonged to the age group between 21 and 30 years. Congenital anomalies were seen more commonly (3.3%) in the multiparas in comparison with primiparas (1.8%). The predominant system involved was Musculo-skeletal system (33.2%) followed by gastro-intestinal (GI) system (15%). Talipes (17.1%) was the most common one in musculoskeletal group and likewise cleft lip and cleft palate in GI system. Congenital anomalies were more likely to be associated with low birth weight, prematurity, multiparity, consanguinity and cesarean delivery. **Conclusion:** Public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended.

**Keywords:** Congenital anomaly, prematurity, prevalence, risk factors, types.

## INTRODUCTION

According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.<sup>[1]</sup>

However, as per the more recent WHO fact-sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.<sup>[2]</sup> Congenital anomalies are an important cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.<sup>[3,4]</sup> It is not only a leading cause of fetal loss, but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families.

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### Aims and Objectives

This study was undertaken to determine the proportion and pattern of congenital anomalies in live newborns and to study the associated maternal and perinatal risk factors.

## MATERIALS AND METHODS

This prospective study was conducted over a 1 year period (March 2014 to February 2015) in the department of Pediatrics and Obstetrics/Gynaecology at Patliputra Medical College, Dhanbad, Jharkhand during the period of May 2014 to April 2015. All babies borne less than 28 week of age were excluded from this study.

The newborns were examined and assessed systematically for the presence of congenital anomalies. Diagnosis of congenital anomalies was based on clinical evaluation by the paediatrician and other investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc., System wise distribution of the anomalies was performed. For each case, a detailed antenatal and maternal history, including the age of the mothers, parity or the history of consanguinity were obtained by reviewing the maternal and labour ward records and by interviewing the parents.

A marriage has been considered consanguineous, when that is found to have occurred between a male and a female who are blood related, e.g., between brother and sister, between 1st cousins etc., Birth weights >2.5 kg were considered to be normal; Whereas, birth weights <2.5 kg and <1.5 kg were termed as low birth weight (LBW) and very low birth weight (VLBW) respectively. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as premature.

Data was entered into excel data sheet and appropriate statistical analysis was performed.

Proportion was calculated and the association was tested with Chi-square test and Fisher's exact test. P < 0.05 was considered to be statistically significant.

### RESULTS

During the study period, 4580 newborns were born in our institution; of which 213 had congenital malformations, making the prevalence 4.65%. Among all the newborns, 56 babies were born of twin delivery, three of triplet delivery and five of these 59 babies, that were products of multiple

gestations, had one or more congenital anomalies. The congenital anomalies affected significantly higher proportion of male babies (2.9%) than their female counterparts (1.5%).

The predominant system involved was a musculo-skeletal system (34.7%) followed by gastro-intestinal (GI) system (16.9%) and central nervous system (CNS) (13.1%) [Table 1]. CTEV (18.9%) was the most common anomaly seen in the musculoskeletal group and likewise cleft lip (33.3%) and cleft palate (5.5%) in GI system and meningocele (60.7%) in CNS. [Table 1].

**Table 1:** System wise distribution of Congenital anomalies (n=213).

System	Number	Percentage
<b>Musculo-skeletal system</b>	74	34.7
CTEV	14	18.9
Calcaneo-valgus	11	14.8
Polydactyly	5	6.7
Syndactyly	8	10.8
Absence of depressor anguli oris	6	8.1
Absence of pectoralis major	4	5.4
Vertebral anomalies	12	16.2
Pterygium	14	18.9
Osteogenesis imperfecta	2	2.7
Phocomelia	8	10.8
<b>Gastro-intestinal system</b>	36	16.9
Cleft lip	12	33.3
Cleft palate	2	5.5
Tongue tie	4	11.1
Imperforate anus	5	13.8
TEF	3	8.3
Ranula	4	11.1
Gastroschisis	2	5.5
Omphalocele	1	2.7
Duodenal atresia	1	2.7
Malrotation of gut	1	2.7
Annular pancreas	1	2.7
<b>Central nervous system</b>	28	13.1
Meningocele	17	60.7
Encephalocele	3	10.7
Hydrocephalus	3	10.7
Anencephaly	2	7.1
Holoprosencephaly	1	3.6
Microcephaly	2	7.1
<b>Genitourinary system</b>	17	7.9
Hydronephrosis	10	58.8
Ambiguous genitalia	2	11.7
Posterior urethral valve	1	5.8
Polycystic kidney	1	5.8
Hypospadias	1	5.8
Epispadias	1	5.8
Extrophy of bladder	1	5.8
<b>Cardiovascular system</b>	12	5.6
Acyanotic	7	58.3
Cyanotic	5	41.6
<b>Skin</b>	17	7.9
Hemangioma	11	64.7
Skin tag	2	11.7
Aplasia cutis	1	5.8
Blueberry muffin	1	5.8
Piebaldism	1	5.8
Others	1	5.8
<b>Syndromes</b>	7	3.2
Down syndrome	4	57.1
Holt-Oram syndrome	1	14.2
Prune Belly syndrome	1	14.2
TAR syndrome	1	14.2

<b>Respiratory system</b>	3	1.4
Diaphragmatic hernia	1	33.3
Eversion of diaphragm	1	33.3
Choanal atresia	1	33.3
<b>Multiple system affected</b>	19	8.9

Regarding the parity of the mothers, 9,185 were primiparas and rest 3,425 were multiparas. Cases of congenital anomaly were found in 3.3% of multiparas, whereas in primiparas, the proportion was only 1.8%. It has been seen that more than half of the mothers were aged between 20 and 30 years (55.7%) with only 10% of the mothers were over the age of 30 years. The prevalence of congenitally anomalous babies born was 1.9% for mothers <20 years, 2.4% for 20-30 years and 2.2% for >30 years. However, this difference was not statistically significant. In the present study, 5 mothers had a history of consanguinity and two of them showed some congenital anomaly (40%) in their babies, whereas in non-consanguineous couples, the prevalence was only 2.2%. This percentage was about 18 times less than in consanguineous couples and was highly significant. Prematurity and LBW was found to have a higher risk of congenital anomalies. The occurrence was about 4.5 times more in case of preterm delivery as compared with the term ones, making it statistically significant. Mode of delivery was also significantly associated with congenital anomaly and it was more in case of cesarean deliveries.

## DISCUSSION

The pattern and prevalence of congenital anomalies 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 variables.<sup>[5]</sup> With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India.<sup>[6]</sup>

In the present study, the prevalence of congenital malformations in the newborns were 4.65%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.<sup>[7,8]</sup> There are other reports from different parts of the world representing different frequency of congenital malformations.<sup>[9,10]</sup> Although we got nearly the same result as reported in other studies,<sup>[7-11]</sup> the prevalence of congenital anomaly would have been more than the present rate, if we could have included the abortions and stillbirths. Tertiary care hospitals usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal

for true estimation of incidence of congenital anomalies in a population.

With regard to pattern of congenital anomalies in the study, the most common system involved was musculoskeletal system (34.7%), followed by a gastro-intestinal tract (GIT) (16.9%), CNS (13.1%), genitourinary (7.9%), cardiovascular system (5.6%), skin (7.9%) etc. This was comparable with studies conducted by others.<sup>[12-17]</sup> Some studies however recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system,<sup>[9,18]</sup> whereas Suguna Bai et al.<sup>[19]</sup> reported GI malformations as the most common one.

More male babies with congenital anomalies than females were noted in the present study. Male preponderance were similar to the other studies.<sup>[6,7]</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.

An association of LBW with increased risk of congenital malformations is very well- documented.

<sup>[6]</sup> Our finding is in accordance with that. The incidence of congenital anomalies was significantly higher in preterm babies as compared with the full term babies, which is in conformity with the previous studies reported from this country.<sup>[17]</sup> Mode of delivery also showed a significant association with congenital anomalies in this study with caesarean section being more commonly associated than normal delivery.

Suguna Bai et al.<sup>[19]</sup> reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta et al.<sup>[18]</sup> documented statistically insignificant association of increased maternal age and congenital anomalies. The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were born of mothers aged 20-29 years; though, it was statistically insignificant.

Previous studies have reported significantly higher incidence of malformations among the multiparas.<sup>[6]</sup> Our result is consistent with this finding, which indicates a positive correlation between the birth order and the incidence of congenital anomalies.

Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations.<sup>[20]</sup> In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages as seen in studies from Kuwait, Arab<sup>[21,22]</sup> and also India.<sup>[17]</sup>

Despite the high risk of recurrence of congenital malformations, there are no well accepted preventive measures in developing countries like India. It indicates that strong preventive measures for

congenital anomalies in this region are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

## CONCLUSION

This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

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