

## Incidence of congenital anomalies in neonates – a tertiary care experience

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**Introduction:** Congenital malformations represent a defect in the morphogenesis during fetal life. Since the introduction of primary health care, congenital malformations have emerged as one of the commonest causes of perinatal mortality. Hence timely detection and intervention can decrease the morbidity and mortality associated with the congenital anomalies. **Aims and Objectives:** To study the incidence of congenital anomalies among live births, term, and preterms, to classify the congenital anomalies and study the underlying risk factors and to study the immediate outcome of congenital malformations in hospital delivered neonates. **Methods:** This was a prospective study in which total live births in a duration of 6 months from August 2018 to January 2019 were studied. Data were obtained in the form of total live births-preterm and full term. The congenital anomalies were identified within 24 hours and classified according to the system involved. The information regarding maternal risk factors, age of the mother, family history, previous sibling death, antenatal detection, MRI and CT findings were studied. Detailed history, examinations, and investigations were carried out to identify etiological factors. The further outcome in the form of morbidity, mortality, and intervention was studied. **Results:** The total incidence of CM among live birth was 6.1%. The incidence among term neonates was 5.1% and among preterms was 33%. The most common system involved was gastrointestinal followed by musculoskeletal, neurological and cardiac (mention percentages). **Conclusions:** The overall incidence of congenital anomalies among live birth was 6.1%. The incidence in full-term was and incidence in preterms was 5.1% and 33% respectively. Male gender, multiparity, and consanguinity were identified as risk factors. Gastroesophageal anomalies were the most common followed by musculoskeletal, neurological and cardiac.

**Keywords:** Congenital malformations, Perinatal mortality, Morbidity

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## Introduction

The World Health Organization defines the term congenital malformation (CM) as structural defects present at birth [1]. The first trimester, especially between the 3rd and 8th weeks of gestation, is the crucial period for morphogenesis of organs. Any insult in any form during this period can cause congenital abnormality [2]. This is the period where preventive intervention strategy will reduce the incidence of developing CM. The prevalence of CM ranges between 3% and 7% and varies in different geographical, racial, and ethnic parts of the world [3] [4]. In India, CMs have emerged as the third most common cause of perinatal mortality [5]. The prevalence of birth defects in India is 6-7% which translates to around 1.7 million birth defects annually. CM may be minor or major. The minor malformation is defined as structural abnormality present at birth which has minimal effect on clinical function but may have a cosmetic effect, for example, a preauricular tag. Major malformation has a significant effect on function or on social acceptability, for example, ventricular septal defect and cleft lip [6]. Congenital anomalies can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies [7]. Expenditures of hospitalization and treatment procedures for these children impose a large excess burden on the health system and their families [8]. There have been many studies in India on the incidence of congenital anomalies. But there are very few studies in the state of Maharashtra. Hence this study has been conducted to throw light on the overall and individual incidence and of clinically detectable congenital malformations in newborns delivered at a tertiary hospital and their association with fetal factors and maternal risk factors.

## Aims and Objectives

To study the incidence of congenital anomalies among live births, term, and preterms.

To classify the congenital anomalies and study the underlying risk factors.

To study the immediate outcome of congenital malformations in the hospital delivered neonates.

## Methods and materials

**Type of study:** Prospective study

**Duration of the study:** Total live birth in a duration of 6 months from August 2018 to January 2019 were studied.

**Inclusion criteria:** Age less than or equal to 30 days, neonates delivered in the hospital.

**Exclusion criteria:** Age above 30 days, outside deliveries admitted in the hospital

**Data collection:** Consent from the ethical committee was taken. Data were obtained in the form of total live births –preterms and full terms. The congenital anomalies were identified within 24 hours and classified according to the system involved. The information regarding maternal risk factors, age of the mother, family history, previous sibling death, antenatal detection, MRI and CT findings were studied. For the cardiac anomalies, 2D ECHO was done. The congenital anomalies were classified according to WHO-ICD 10 classification [9]. The congenital anomalies are included in the Q 00-Q 99 classification. They were also divided into minor and major anomalies. Detailed history, examinations, and investigations were carried out to identify etiological factors. The further outcome in the form of morbidity, mortality, and intervention was studied.

**Statistical analysis:** Statistical analysis was done using the chi-square test and t-test. P-value <0.05 was considered significant.

## Results

**Table-1: Demographic profile.**

Total deliveries	4512	% of live birth
Livebirths	4453	
Full terms	4302	96.6%
Preterms	151	3.3%
Stillbirth	9	0.1% of the total deliveries

Table 1 shows the percentage of full terms, preterms, and stillbirth over the study period

**Table-2: Total incidence of congenital anomalies.**

Incidence	Per thousand live birth
Live births	6.1
Full-term	5.1
Preterm	33

Table 2 shows the incidence of congenital anomalies among total live births (6.1%), among full terms (5.1%), and preterms (33%).

**Table-3: Genderwise distribution.**

Gender	No. live births	No of pts with congenital anomaly
Males	2393	20
Females	2060	5

P=0.00281 p significant

Out of the total neonates with congenital anomalies 20 were males and 5 were females. Congenital anomalies were significantly more in males compared to females (P<0.05) (table 3)

**Table-4: Maternal risk factors**

Oligohydramnios	4	16%
Polyhydramnios	4	16%
Diabetes mellitus	1	4%

9 mothers of neonates with congenital anomalies had risk factors, of which 4 had oligohydramnios, 4 had polyhydramnios, and 1 had diabetes (table 4)

**Table-5: Maternal age**

Maternal age	No of neonates with congenital anomalies	% of congenital anomalies
<20	2	8
20-25	12	48
26-30	8	32
>30	3	12

Of the mothers of the neonates with congenital anomalies, a maximum of 48% was between the age of 20-25 yr (table 5)

**Table-6: Parity**

Primiparous	4
Multiparous	21

21 mothers of the neonates with congenital anomalies were multipara and 4 were primipara (table 6)

**Table-7: System wise distribution**

Congenital anomalies	No of neonates	% of congenital anomalies
Central nervous system	4	16
Meningomyelocele		
MMC with hydrocephalus		
Congenital hydrocephalus		
Arnold Chiari malformation		
Musculoskeletal	5	20
Polydactyly		
Klippel Feil		
CTEV		
Cardiovascular	3	12
ASD with PDA		
PDA		
VSD		

Anorectal	2	8
Anal atresia		
Anal fistula		
Genitourinary	3	12
Polycystic kidney		
Hypospadias		
Congenital hydronephrosis		
Gastroesophageal	6	24
Tracheoesophageal fistula		
b/l cleft lip and palate		
cleft lip		
Ear	1	4
Microtia		
Multiple anomalies	1	4

The most common congenital anomalies identified were of gastroesophageal (24), followed by musculoskeletal (20), central nervous system (16), cardiovascular (12), genitourinary (12) anorectal (8) and ear anomalies (4) (table 7)

Major anomalies -14 minor anomalies -11

Antenatal detection- all four neurological cases and one heart disease was detected antenatally

Family history –one father and sibling history of cleft lip

Consanguinity –seen in 5 patients

**Table-8: Outcome**

Outcome	No of patients	%
Mortality	2	8
Morbidity	4	16
Intervention /pediatric surgery referral	19	76

The death occurred in 2 patients. 4 patients had increased morbidity and 19 patients were referred for further management (table 8)

## Discussion

Congenital Malformations represent one of the causes of neonatal mortality. Health-care managers must stress on primary prevention in the form of good antenatal care, nutrition, and drugs to decrease the preventable share of CMs. Early detection and timely management are required to decrease mortality.

**Incidence:** The incidence of congenital anomalies among the total live births was 6.1%% per thousand live births (Table 2). Many studies in India have addressed the prevalence of birth defects in the country. Their frequency varies from 1.94% to 2.03% of birth on an average [5].

In the present study, the incidence of CM in live births was comparable to study done by Padmanabhan et al [6] who showed an incidence of 4.08%, however, this incidence was higher when compared with the study by Taksande *et al.*, [10] which showed an incidence of 1.9% in live births. Singh and Gupta [11] show an incidence of 1.5% in live births. There is a variation in the incidence of congenital anomalies in other countries. A study in Iran showed an incidence of 11.2 per thousand live births [8]. Another study done in Kathmandu showed an incidence of 0.6% [12]. Thus it is necessary to identify the overall incidence of congenital anomalies for early detection of the cases. This difference in the incidence can be due to different racial, ethnic, and social factors in various parts of the world

**Incidence among terms and preterms:** In the present study incidence of Congenital malformations was 5.1% in full terms and 33% in preterms. This difference was not statistically significant. 20% of the malformed babies were preterms and 80 % of the malformed babies were full terms. This is comparable to the study done by Padmanabhan et al [6] who showed that 23% of malformed babies were preterm and 77% of babies were full term.

A study by Malla [12] and Dutta *et al* [13] showing similar results (36% preterm and 64% full-term, and 40.6% preterm and 59.4% full-term babies, respectively). Patel et al [5] showed an incidence of 33% in preterms and 17 % among full terms Thus preterm delivery is associated with an increased incidence of congenital anomalies. This can help in proper antenatal care and early detection of congenital anomalies in the preterms.

**Genderwise variation:** In the present study 80% of the malformed babies were males and 20 % of the malformed babies were females. this genderwise difference was statistically significant. In the study done by Gandhi et al (14) no significant genderwise distribution of congenital anomalies was seen. In a study done by Padmanabhan et al (6) male babies were more affected with malformations. 54% of total malformed babies were male and 45% of female babies. A study by Taksande *et al* (10) showing similar results (61% of male babies and 37.4% of female babies). another study like Mohanty et al [15] has shown male preponderance. This can be because of females having extreme congenital or chromosomal anomalies leading to stillbirths or intrauterine deaths.

## Maternal factors

**1) Maternal age:** In the present study incidence of congenital anomalies was higher in the 20-25 age group (48%). This can be because of maximum deliveries in this age group .in a study done by Padmanabhan et al (6) the incidence of malformation was higher (87.4%) in mothers aged 21-30 years, and 9.2% in mother >31. Another study was done by Swain et al [16] however showed a higher incidence in age group >35 years.

**2) Parity:** In the present study the 84% of the mothers with babies having congenital malformations were multiparous and 16% of the mothers were primiparous. Thus the incidence of congenital malformations was higher in multiparity as compared to primiparity. In a study done by Sarkar et al [17], the incidence was higher in multipara as compared to primipara. a study was done by Taksande et al [10]. reported an incidence of malformations among the multiparas as 19.5%.

**3) Maternal complications:** Maternal complications like oligohydramnios can cause neural tube defects. Polyhydramnios can cause gastroesophageal malformations like tracheoesophageal fistula .in the present study 9 mothers had complications in the form of oligohydramnios (4), polyhydramnios (4) and diabetes mellitus (1). an interesting finding noted was that all the mothers of neonates with trachea-esophageal fistula had polyhydramnios.

The infant of the diabetic mother had a PDA with ASD. In the study done by Kokate et al [18], maternal complications noted were polyhydramnios in 8% and diabetes mellitus. Thus early detection of maternal complications is essential as timely intervention can be done.

5) consanguinity Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations. In the present study, consanguinity was seen in 5 mothers whose babies had congenital anomalies (20%).

In the study done by Malla et al [12] only 8.33% of CBDs had consanguinity history and it is common in a certain community only [12] Increased prevalence of CBD in consanguineous couples is due to homozygous expression of recessive genes inherited from common ancestors [16]. Kokate et al [18] showed consanguinity in 50% of the mothers of infants with CM.

### Classification of congenital anomalies:

Classification of congenital anomalies is made on the basis of ICD 10 classification of congenital anomalies in which sections Q00 to Q89 are reserved for congenital anomalies [9] the commonest system involved was gastrointestinal (24%) followed by musculoskeletal [20], central nervous system [16]. Other systems involved were anorectal, genitourinary, ear and cardiovascular.

Among the individual anomalies, the commonest congenital anomaly detected overall was neural tube defects. these included Arnold Chiari malformations, meningomyelocele with hydrocephalus and congenital hydrocephalus with meningomyelocele. The diagnosis was made with the help of ultrasonographic and MRI findings. There are wide variations in the system-wise distribution of congenital anomalies in various studies.

This was comparable with a study conducted by Taksande et al [10]. which shows the cardiovascular system (23%), musculoskeletal system (21.9%), gastrointestinal tract (14%), genitourinary (18.9%), and central nervous system (9.1%). Central nervous system malformations were predominantly seen in the study by Sugunabai et al [19] and Malla et al (12) (44% and 40%, respectively); gastrointestinal system malformations are predominantly seen in the study by Desai and Desai et al [20].

Congenital anomalies can also be divided into major and minor anomalies. In the present 66% (14) of the total anomalies were major and 44% [11] of the total anomalies were minor.

**Outcome:** In the present study death occurred in 2 patients with congenital anomalies. one was an infant having multiple anomalies and another was an infant having MMC with hydrocephalus. The other 3 neonates with central nervous system involvement had increased morbidity in the form of no movements in the lower limb. The other patients were referred for intervention. In a study done by Patel et al [6], the majority of the babies with malformations were discharged (78.9% of the total malformed babies) only 11.8% of babies were expired and 2.6% of babies left against medical advice.

Thus, Congenital anomalies are an important cause of infant and childhood deaths, chronic illness, and disability. Strategies have to be developed to diagnose, treat, rehabilitate, and prevent birth defects.

In preparation for this and effective planning, crucial measures include obtaining data on prevalence, nature of birth defects, genetic contributions, morbidity, and mortality. The community-based study like the present one should be ideal for true estimation of the incidence of congenital anomalies in a population.

**Limitations:** The present study shows the incidence of congenital anomalies over a short period of 6 months. Also, it does not mention the follow up of these patients post their discharge.

## Conclusion

The overall incidence of congenital anomalies among live birth was 6.1%. the incidence in full-term was and incidence in preterms was 5.1% and 33% respectively. Male gender, multiparity, consanguinity were identified as risk factors. Gastroesophageal anomalies were most common followed by musculoskeletal, neurological and cardiac.

## What does the study add to the existing knowledge?

This study has studied in detail the various risk factors particularly maternal age, parity, consanguinity. Also, the outcome of the neonates aids in their further management and counseling of the parents.

## Author's contributions

**Dr. Madhura Prakash Fadnis:** Data collection, interpretation and manuscript writing

**Dr. Pratibha Shamkuwar:** Manuscript writing and data interpretation

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