

## Pattern of congenital abnormalities in a tertiary hospital and its impact on neonatal mortality

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### ABSTRACT

**Background:** Congenital abnormalities are major contributors of neonatal mortality and stillbirths. However, there is not sufficient data in our country on the prevalence of various congenital malformations and their impact on neonatal mortality. **Objectives:** To study the prevalence and pattern of congenital anomalies among neonates delivered in a tertiary hospital setting in 3 years and its impact on perinatal and neonatal mortality. **Materials and Methods:** This hospital based prospective descriptive study was undertaken at tertiary care hospital in Kerala. All babies born in the hospital from January 2013 to December 2015 (3 years) were included in the study. The baby was examined by a pediatrician during the first 24 h to identify any birth defects. A detailed history including familial and gestational factors was taken in babies with birth defects. Photographs, radiographs, ultrasound examination, echocardiography, and chromosomal studies were undertaken as required. The details were entered in a pro forma. The anomalies are classified as per ICD-10 criteria. Results were analyzed by simple statistical techniques recording number and percentage of cases. **Results:** The prevalence of birth defects in live born newborn was 1.9% whereas, in stillbirths, it was 15.3%. Congenital anomalies also contributed a major risk factor for neonatal death as 22% of the newborns, died in the immediate neonatal period, had some form of congenital anomaly. The major maternal risk factor found to be associated with congenital anomalies was gestational diabetes (21.3%). The patterns of congenital anomalies were musculoskeletal anomalies (25%), central nervous system (18%), genitourinary system (14%), congenital diaphragmatic hernia (12%), cardiovascular system (10%), gastrointestinal (7%), syndromes (6%), non-immune hydrops (5%), and others (3%). **Conclusion:** Prevalence of birth defects in this birth cohort was 1.9% comparable to other Indian data. In Kerala, one of the major causes of perinatal and neonatal mortality is congenital malformations.

**Key words:** Birth defects, Congenital abnormalities, Neonatal mortality, Stillbirths

Congenital abnormalities are structural abnormalities of prenatal origin that results from defective embryogenesis or deviation from normal development [1]. Birth defects, congenital anomalies, and congenital abnormalities were interchangeably used to describe developmental abnormalities present at birth. The pattern and frequency of various congenital abnormalities vary from country to country and in different parts of India due to variations in ethnic, socioeconomic and geographical factors of the population studied. The frequency of congenital malformation varies from 1.07% in Japan to 3% in Taiwan. The variation in frequency is also contributed by the methodology used for the study [2]. There is no community-based data available in India. Some hospital based studies show that the prevalence varies from 1.9% to 2.25% [3,4].

Congenital abnormalities are major contributors to neonatal mortality and stillbirths. According to a joint report by the World Health Organization (WHO) and March of dimes meeting report, birth defects account for 7% of the all neonatal mortality and 3.3 million under-five deaths [5]. In India, congenital

abnormalities account for 8-15% of perinatal deaths and 13-16% of neonatal deaths [6,7]. There is a significant reduction in the infant mortality rate (IMR) in India; especially in state of Kerala, where IMR is slowly approaching to a single digit. When IMR is getting reduced significantly, the contribution by the congenital abnormalities for the mortality will be more. Hence for a further reduction of IMR, prevention and treatment of congenital abnormalities should be the focus.

In these circumstances, we should have a concrete data on the prevalence and pattern of congenital abnormalities in Kerala. Since there is no community-based data available and is also difficult to study, we have done a prospective hospital based study in the tertiary care hospital to identify the prevalence and pattern of congenital abnormalities and their contribution to perinatal and neonatal death.

### MATERIALS AND METHODS

This prospective study was undertaken at a tertiary care hospital in Kerala, which caters to the population of south Kerala and

adjoining districts of Tamil Nadu. All babies born in the Hospital from January 2013 to December 2015 (3 years) were included in the study. The Institutional Ethical Committee clearance was obtained before the study. All parents were informed regarding the study, and their informed consent was taken for inclusion in the study. The babies delivered after 20 weeks of gestation (viability period) and parents given informed consent were included in the study. The baby was examined by a pediatrician during the first 24 h to identify any congenital abnormality. A major congenital anomaly was defined as an anomaly which will cause impairment of the function of that organ. A detailed list of anomalies to be included was provided along with the pro forma which was formulated from ICD 10 criteria.

Detailed histories including familial and gestational factors were taken in babies with birth defects. A neonate identified with significant abnormality was admitted to Neonatal Intensive Care Unit for evaluation and appropriate management. Photographs, radiographs, ultrasound examination, echocardiography, and chromosomal studies were undertaken as required. The details were entered in a predesigned pro forma. Cases with multiple malformations were diagnosed by reviewing databases such as Online Mendelian Inheritance in Man and London Medical Database [8]. The anomalies were classified as per ICD-10 criteria [9].

Data were analyzed by simple statistical techniques recording number and percentage of cases. The percentage of congenital anomalies in live births and stillbirths was calculated. The pattern of congenital anomalies involving various systems was also calculated as percentages compared with studies from other parts of India. Cause-specific mortality due to congenital malformations contributing to neonatal/perinatal mortality also calculated.

## RESULTS

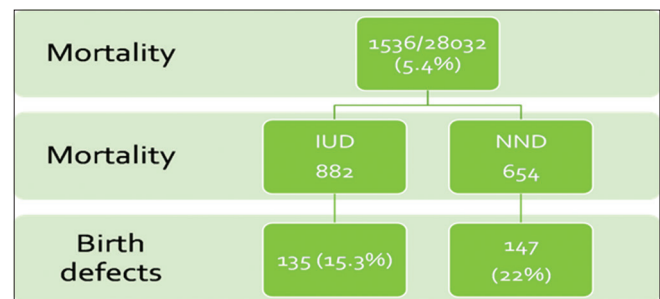
During the 3 years study, there were 28,032 deliveries out of which 27,150 were live births. Among these live-born babies, 519 had congenital abnormalities accounting for an incidence of 1.9% (19 per 1000 live births). In the study period, 882 cases were classified as stillbirths/fresh intrauterine death where 135 cases (15.3%) had one or multiple congenital abnormalities. Among the live born babies, 654 neonates died in the neonatal period, and 147 babies had malformations accounting to 22.0% cause-specific mortality (Fig. 1).

Table 1 summarizes the pattern of congenital abnormalities seen in neonates. The patterns of congenital anomalies were musculoskeletal anomalies (25%), central nervous system (CNS) (18%), genitourinary system (14%), congenital diaphragmatic hernia (12%), cardiovascular system (10%), gastrointestinal (7%), and syndromes such as Down syndrome, Cornelia De Lange syndrome (6%), non-immune hydrops (5%), and others (3%). Among musculoskeletal system, congenital talipes equinovarus was the most common anomaly whereas, among CNS anomalies, neural tube defect was the most common. In this study, 70.5% of the cases with major anomalies could not be picked up sufficiently early during the antenatal period so that any intervention could be

**Table 1: Pattern of congenital abnormalities seen in neonates with birth defects**

System	n (%)
Musculoskeletal system	138 (25.4)
CNS	96 (17.6)
Genitourinary system	75 (13.8)
Congenital diaphragmatic hernia	63 (11.6)
Cardiovascular system	54 (9.9)
Gastrointestinal system	36 (6.6)
Syndrome (Down syndrome, Cornelia De Lange syndrome, etc.)	33 (6.0)
Hydrops fetalis	30 (5.5)
Miscellaneous	18 (3.3)
Total	543 (100)

CNS: Central nervous system



**Figure 1: Cause-specific mortality due to congenital abnormality in stillbirths/fresh intrauterine devices and neonatal mortality**

done early. Of the total 519 cases with birth defects, 306 cases were having one, or multiple risk factors studied, which is 59% of cases with anomalies.

The major risk factors associated with birth defects were gestational diabetes mellitus (GDM) and amniotic fluid abnormality. The prevalence of GDM was 21.3% in neonates with congenital abnormalities whereas the prevalence was 16.5% among the total deliveries. In contrast, 12% of antenatal mothers had pregnancy induced hypertension (PIH) whereas the frequency of PIH in antenatal mothers with congenital abnormality is only 5%. Among the cases with congenital abnormalities, 3.8% of cases were associated with consanguineous marriage. Other maternal risk factors associated with congenital abnormalities were maternal hypothyroidism, and amniotic fluid abnormality - 13% cases had oligohydramnios, and 7% cases had polyhydramnios. 76% of the newborns with congenital anomalies were more than 28 weeks gestation whereas 24% were <28 weeks gestation.

## DISCUSSION

The prevalence of congenital malformations at birth varies in different studies because of complex interaction between the unknown genetic factors and environmental factors such as sociocultural factors and ethnic variables. This may also depend on the methodology used in the study. The overall prevalence of congenital anomalies in this study was 1.9%, which was

**Table 2: Pattern of congenital abnormalities in this study in comparison with other studies**

System	This study n (%)	El Koumi et al [10]	Sarkar et al [3]
Musculoskeletal system	138 (25.4)	23	33.2
CNS	96 (17.6)	20.3	11.2
Genitourinary system	75 (13.8)	13.5	10.5
Congenital diaphragmatic hernia	63 (11.6)	-	-
Cardiovascular system	54 (9.9)	9.5	9.1
Gastrointestinal system	36 (6.6)	16.2	15
Syndrome	33 (6.0)	6.8	8.7
Hydrops fetalis	30 (5.5)	-	-
Miscellaneous	18 (3.3)	10.8	
Total	543 (100)		

CNS: Central nervous system

comparable to other studies. A similar prospective study from Egypt showed 2.5% [10], and frequency in a birth cohort of UAE population was 1.05% [11]. The variation may be due to the different ethnic background of the population and methodology used in the different studies. In Indian population, a hospital based study from eastern India showed a prevalence of 2.22% [4] whereas another hospital based study from central India showed a prevalence of 1.91% [3]. In India, we are not having any population-based data. In a recent study from Northeastern India showed increase malformations in stillbirths [12]. A study conducted in Shimla also showed an increased incidence of congenital malformations in stillbirths [13]. All hospital based data are showing an increased prevalence since they are mainly catering high-risk pregnancies and referral cases leading to a minimal increase in the prevalence of congenital anomalies.

With regard to pattern of congenital anomalies in this study, the most common system involved was musculoskeletal system (25.4%), followed by CNS (17.6%), genitourinary (13.8%), congenital diaphragmatic hernia (11.6%) cardiovascular system (9.9%), and gastrointestinal tract (GIT) (6.6%). This was comparable with studies conducted by others [14-19]. Table 2 summarizes the pattern of congenital anomalies in this study compared with other two studies. However, some studies recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system [20,21], whereas Suguna Bai et al. reported GI malformations as the most common one [22]. With improved control of infections and nutritional deficiency disorders, congenital anomalies have become important causes of prenatal mortality in our country. In the cause-specific analysis of mortality, in our hospital, 15.3% of cases with stillbirths had some congenital malformations whereas 22.0% of cases with neonatal death had congenital anomalies. This is comparable to the existing literature data by the WHO [5]. Our cause-specific mortality due to congenital anomalies is higher than another part of India. Since our state, Kerala is having the lowest IMR of 12 and rapidly approaching to single digit IMR, this epidemiological change in mortality is an expected one. For

further reduction of infant mortality, genetic services need to be strengthened. Strengthening the antenatal screening system with newer advances in the field of obstetric imaging and better compliance of patients with routine antenatal checkup will help in early detection and intervention [23,24]. Early detection can help in making appropriate decision regarding continuation or termination of pregnancy.

According to March of Dimes (MOD) and WHO report 70% of the birth defects are preventable if the evidence-based community genetics services are used. Community genetics services include a number of activities for the diagnosis, care and prevention of genetic diseases at the community level [25,26]. Community genetic services have been given low priorities in India because of the paucity of resources, inadequacy of data on the burden of birth defects and an insufficient number of trained health personnel. Many intervention strategies are available for the prevention of birth defects such as preconception counseling, periconceptional folic acid, and control maternal risk factors like diabetes mellitus. All these strategies demand reproductive and child care before or early in conception till the diagnosis of birth defect, or even later [27].

There are certain limitations were present in this study. As it is a hospital based prospective study, the frequency of congenital abnormalities is more than in a community-based study. This data may not be extrapolated to the general population. More extensive community-based population studies or registry data need to evaluate the burden in the community level. Second, since the anomalies identified at birth were included in the study, some of the internal anomalies may be missed. Moreover, the major anomalies identified by antenatal ultrasonography and terminated before 20 weeks also is not reflected in this study.

## CONCLUSIONS

Prevalence of birth defects in this birth cohort is 1.9% comparable to other Indian data. In Kerala, one of the major causes of perinatal and neonatal mortality is congenital malformations. With improved control of infections and nutritional deficiency disorders, congenital anomalies have become important causes of prenatal mortality in our country. The control of risk factors such as GDM, folic acid supplementation during preconception period and quality, and timely antenatal ultrasound will help to reduce this incidence of congenital anomalies.

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