

## Original Article

### Prevalence of Congenital Malformations in Neonates- A Prospective Study

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#### ABSTRACT

**Background:** Congenital anomalies constitute the fifth largest cause of neonatal mortality in the country, but national estimates of the prevalence of these conditions are lacking. The objective of the study was to derive an estimate of the birth prevalence of congenital anomalies in India. **Material and Methods:** The retrospective study of live neonates from newborn to 1 month of age both inborn and outborn admitted to the hospital irrespective of their general condition with CMs comprised the study population. Details of investigations like ultrasonography, radiology, echocardiography, laboratory studies have done were noted from the case record. Their outcome in the form of morbidity, hospital stay, and mortality was analyzed. **Result:** Three thousand four hundred and fifty newborn babies of consecutive deliveries were examined at birth for the presence of congenital malformations. The overall prevalence of malformations was 3.18%. Neural tube defects were commonly found. The incidence of congenital malformations was higher in still born, low birth weight, male and preterm babies. **Conclusion:** CMs 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.

**Keywords:** Birth defects, Congenital anomalies, Congenital malformations, Birth prevalence, India.

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#### INTRODUCTION:

A quarter of global neonatal deaths occur in India. In 2015, the country reported a neonatal mortality rate of 23 per 1000 live births, responsible for 613,000 neonatal deaths.<sup>1</sup> While the highest contributors to neonatal deaths were preterm births (31%), intrapartum complications (19%), pneumonia (15%) and neonatal sepsis (15%), congenital anomalies constituted the fifth largest cause, being responsible for an estimated 9% of neonatal deaths in the year 2010.<sup>2</sup> There is evidence of transition in causes of infant and child mortality in low and middle-income countries, including India.<sup>3</sup> With a decrease in infectious causes of infant deaths, especially in urban areas in India, the proportion of mortality due to congenital anomalies is likely to increase.<sup>4</sup>

Global estimates suggest that congenital anomalies affect 2–3% of births. CAs contribute to a significant proportion of fetal and infant mortality.<sup>5</sup> Various sources estimate the

prevalence of CAs to be in the range of 1%–3% of all live born infants and the estimates are considerably higher for the infants that are stillborn or spontaneously aborted.<sup>6</sup> Older women, women with medical conditions such as hypothyroidism, uncontrolled diabetes, placental insufficiency, multiple pregnancy, and oligo hydramnios have a higher risk of major CAs than that of the general population. Major malformation has a significant effect on function or on social acceptability, for example, ventricular septal defect and cleft lip.<sup>7</sup> Dysmorphology is the study of abnormalities of the human form and mechanism that causes these abnormalities. About 30% of infant deaths and 30–40% post-neonatal deaths are due to CM. 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. This is the period where preventive intervention strategy will reduce the incidence of

developing CMs.<sup>8</sup> Anomalies are more common among spontaneous abortions. Many anomalies are severe and cause abortion. Congenital anomalies represent defective morphogenesis during early fetal life. A broader definition includes metabolic or microscopic defects at a cellular level. Major anomalies have serious medical, surgical and cosmetic consequences. In this study we have calculated overall prevalence of congenital anomalies both in live born and stillborn babies.

**MATERIAL AND METHODS:**

This community based prospective study was conducted in Department of Pediatrics, Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, India. The study was conducted comprises of 145 villages having total population of 186,567. They were examined soon after birth for major and/or minor congenital malformations.

Baby's gestational age, birth weight, sex and symptoms in postnatal period were noted. The detailed general and systemic examinations of the babies were carried out. As per the proforma made, complete medical, family, antenatal and personal history has taken. Thorough physical examinations of newborn babies were done. High risk newborns were examined in detail within 12 hours of birth. Details of investigations like ultrasonography, radiology, echocardiography, laboratory studies have done were noted from the case record. Immediate outcome of all malformed babies were recorded during the period of the mother's hospital stay.

Studies were eligible to be included in the review if they fulfilled the following inclusion criteria: reported data on the number of anomaly, affected babies or anomalies identified at birth among either live born and/or stillborn babies and were conducted in India. Exclusion criteria include case reports and papers focusing on etiology,

diagnosis or clinical management were excluded and studies that reported prevalence data of only a single anomaly or system were not included in the analysis as these represented non-random, selected cases, and would therefore distort prevalence estimates.

Birth prevalence of congenital anomalies was calculated as the total number of babies (both live born and stillborn) with anomalies per 10,000 births. The live birth prevalence was determined from the number of anomaly affected live births per 10,000 live births.

**RESULTS:**

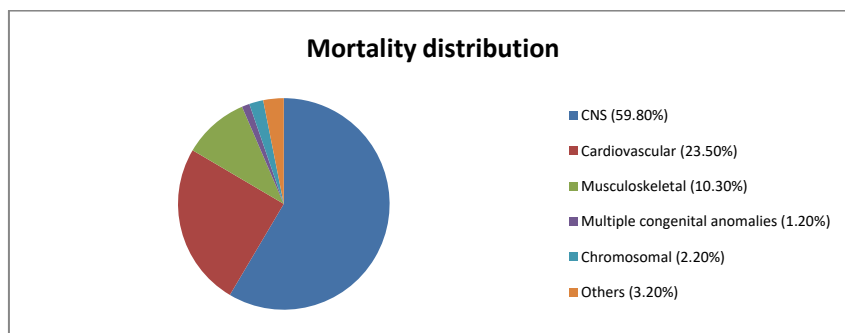
A total of 3450 consecutive births were studied for congenital malformation. There were 106 malformed babies found. The overall prevalence of congenital malformations was found to be 3.18%. Thirty women had abortions and 40 others delivered stillborn babies. Abortions and stillbirths could not be further investigated to rule out presence/absence of CAs because of limited resources and facilities to conduct such investigations in community based settings.

The age range of the study population was 18–45 years with mean age ±SD as 24 ± 6 years. About 7.9% of women, were aged <20 years or above 34 years. Out of 1230 live births, 58.6% were males and 41.4% were females. The overall incidence of CAs in males was slightly higher than in females, i.e., 28.1/1000 live births versus 22.3/1000 live births. 34 newborns had single CAs while 6 newborns were having multiple CAs.

Congenital malformations of the central nervous system were the highest followed by musculoskeletal system, gastrointestinal system, cardiovascular system, Genitourinary system, respiratory system, chromosomal and ear.

Table 1: Prevalence of anomalies

Total No. of deliveries	3450
Total No. of twin deliveries	8
Total No. of triplet deliveries	1
Total No. of babies born	3420
Total No. of malformed babies	40
Total No. of abortions	30



**Graph 1:** Pie chart showing mortality distribution

## DISCUSSION:

Congenital anomalies are not prioritized as public health problems in low income countries as they are considered to be rare conditions that are self-limiting due to the high mortality of affected infants.<sup>9</sup> Another reason for under-prioritization of these conditions is the understanding that most birth defects are not preventable through low-cost primary care strategies, the major approach of public health services of low income countries.

Neural tube defects (NTDs) like anencephaly are potentially preventable through a low cost primary prevention method of preconception folic acid supplementation, but there are as yet no national guidelines on folic acid fortification/ supplementation in India.<sup>10,11</sup> Combined with preconception iron supplementation, this primary care intervention could not only reduce the number of NTDs in the country, but also reduce anemia, a persistent maternal health challenge in low income countries.<sup>12</sup>

Community-based studies reported a higher prevalence of musculoskeletal anomalies, with talipes, a potentially treatable anomaly, being reported as the most common congenital anomaly among live births. Thus, in addition to determining the large numbers of affected births, this review identified that the two most commonly reported congenital anomalies were preventable/treatable through low cost methods. For example, the management of talipes through casting is relatively inexpensive, is widely available, and with proper compliance will prevent disability.

In present study, attempts have been made to find out the total and individual prevalence of anomalies in hospital deliveries. The overall incidence of congenital malformations was 3.18% in present study. Compares well with the observations of Marden et al(1964) 2-4%, Goravalingappa &Nashi(1979) 3.13%, Ghose et al(1985) 1.5%,Graham(1988) 2%,Mishra PC & Baveja R(1989) 1.46%,Mohanty et al(1989) 1.61%,Verma IC et al(1991) 3.6% and Guha AK (1995) 2%.<sup>10-16</sup>

The relative difference in the occurrence of various malformations might be due to geographic and racial differences. The true incidence of congenital anomalies depend upon several factors and therefore two studies are never strictly comparable. In present study Congenital malformations of the central nervous system were the higher followed by malformations of the cardiovascular system malformations of the musculoskeletal system which is similar to study by Goravalingappa &Nashi(1979) and Guha AK(1995) who found high incidence of central nervous system malformations.<sup>10-12</sup> While Mishra PC & Baveja R(1989) found high incidence of multiple congenital anomalies. Ghose et al (1985) and Mohanty et al(1989) found higher incidence of musculoskeletal system malformations.

The incidence of malformation was higher in mother aged of 21–30 years, and 9.2% in mother >31 that is high on comparing with a study by Taksande *et al.* and Saiyad and

Jadav (incidence of malformation 36% and 20% live births, respectively). Taksande *et al.* reported a higher incidence of malformations among the multiparas (19.5%).

Birth defects data from studies conducted during the earlier period could be influenced by the high number of home births, and this could also be a limitation in the estimates. Most of the studies were hospital based. Community based studies were few, and none of the studies mentioned data on home births. For hospital based studies, the catchment areas of hospitals are undetermined due to high patient mobility. Furthermore, the studies included data from large public hospitals which frequently serve as referral centers for high risk mothers and complicated cases. Such methodological issues could be one of the reasons for the different rates observed for anencephaly versus spina bifida, as the latter is the more common condition. Another factor influencing the estimates was that majority of the studies used only clinical assessment for case ascertainment. Incomplete ascertainment may therefore contribute to under-estimation of some anomalies. For example, the low prevalence of congenital heart defects as compared to available registry data could be ascribed to use of only physical examination at the time of birth.

Similarly, Down syndrome which is one of the most common birth defects, was not reported in most of the included studies. This discrepancy could be because our meta-analysis included studies that reported birth defects detected in the first week of life, while Down syndrome may be diagnosed after discharge. Another very important source of under-estimation would be the lack of data on termination of pregnancies due to fetal anomaly, as none of the studies reported this data. Despite these limitations, this review is important, as it is the first to report the magnitude of birth defects in India, and the need to establish a systematic method of surveillance for these conditions. The first point arising from the study is to determine whether surveillance for birth defects in India should be hospital or population based.

## CONCLUSION:

Increasing awareness about maternal risk factors during pregnancy and educational programs on CMs needs to be highlighted to decrease the incidence of congenital anomalies and their co morbidities. CMs 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.

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