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Assessment of factors responsible for congenital anomalies in newborn

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Abstract
Birth prevalence of congenital anomalies in developing countries is actually underestimated due to poor registry, lack of diagnostic techniques and their reliability. Like other low and middle income countries, still, congenital anomalies are not considered as a major problem in India. But literature search reveals that India has the highest number of children with birth defects. Hence, based on the above literature findings, present study was planned to generate a systematic data on the magnitude of congenital anomalies, their pattern of prevalence, healthcare impact and impact on neonatal health. Prevalence studies give an idea about the pattern of occurrence of anomalies in different places, changes over a period of time and also give some clues to identify the aetiology.
The study was planned in Department of Paediatrics, Darbhanga Medical College and Hospital, Laheriasarai. 100 neonates identified with birth defects from May 2017 to Feb 2018 were enrolled in the present study. Approval of the institutional ethical committee was taken prior to conduct of the study and all participants were duly informed and consent taken. The aim and the objective of the present study was conveyed to parents of the neonates.
From present study it has been concluded that congenital anomalies in new-borns were significantly associated with fetal factors like still birth, prematurity and low birth weight. Their incidence can be reduced by following prevention strategies including primary, secondary and tertiary levels of prevention. These strategies can be applied at various stages of pregnancy that is Preconception, Antenatal and Postnatal period.

Keywords: congenital, anomalies, abnormalities, neonates, India

Introduction
Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.
A birth defect, also known as a congenital disorder, is a condition present at birth regardless of its cause. Birth defects may result in disabilities that may be physical, intellectual, or developmental. The disabilities can range from mild to severe. Birth defects are divided into two main types: structural disorders in which there are problems with the shape of a body part and functional disorders in which there are problems with how a body part works. Functional disorders include metabolic and degenerative disorders. Some birth defects include both structural and functional disorders [1].
Birth defects may result from genetic or chromosomal disorders, exposure to certain medications or chemicals, or certain infections during pregnancy. Risk factors include folate deficiency, drinking alcohol or smoking during pregnancy, poorly controlled diabetes, and a mother over the age of 35 years old. Many are believed to involve multiple factors. Birth defects may be visible at birth or diagnosed by screening tests. A number of defects can be detected before birth by different prenatal tests [2].
Congenital anomalies are the major cause of new born deaths within four weeks of birth and can result in long-term disability with a significant impact on individuals, families, societies and health-care systems. In nearly 50% of cases, the exact cause of congenital anomaly could not be identified, although there are some known risk factors which can be linked with the causation of malformation. Congenital anomalies can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens (an agent, which can cause a birth defect) and micronutrient deficiencies.
According to the World Health Organization (WHO) in 2010, an estimated 270 000 deaths during the first 28 days of life were reported due to congenital anomalies globally. According to March of Dimes (MOD) global report on birth defects 7.9 million births (6% of total births) occur annually worldwide with serious birth defects and 94% of these births occur in the middle and low income countries. According to joint WHO and MOD meeting report, birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths. The prevalence of birth defects in India is 6-7% which

translates to around 1.7 million birth defects annually. The common birth defects include congenital heart disease (8-10 per 1000 live births), congenital deafness (5.6-10 per 1000 live births), and neural tube defects (4-11.4 per 1000 live births) (March of Dimes report, 2006) [3].

Some birth defects are clinically apparent at birth; others may only be diagnosed later in life. The structural defect such as spina bifida is obvious at birth whereas haemophilia a functional defect (a bleeding disorder) is not usually obvious until infancy or childhood. The Ministry of Health and Family Welfare, Government of India has addressed the problem with the implementation of various national health programmes. In 2013, National Child Health Screening and Early Intervention Services covered 30 health conditions of the children aged 0-18 years through various approaches.

India Newborn Action Plan (INAP) formulated in September 2014, has integrated the approaches for the prevention and care of newborn with birth defects into primary health care, with an emphasis on maternal and child health. INAP is India's committed response to the Global Every Newborn Action Plan (ENAP) by WHO with a vision to eliminate preventable newborn deaths and stillbirths [4].

Neonatal screening including physical examination of all neonates and screening for functional disorders such as congenital hypothyroidism, phenylketonuria, sickle-cell disease and glucose-6-phosphate dehydrogenase deficiency by trained primary health care providers can be performed. Neonates with birth defects may be further referred to appropriate level of medical/surgical facilities. Effective life-saving medical treatment is available for several birth defects with functional disorders such as thalassaemia (inherited recessive blood disorders), sickle cell disorders and congenital hypothyroidism [5].

Many structural congenital anomalies (about 50%) can be corrected with paediatric surgery in early life, such as simple congenital heart defects, cleft lip and palate, club foot, congenital cataracts, and gastrointestinal and urogenital abnormalities. Simple, cost-effective, and non-invasive treatment also exists for certain conditions such as clubfoot. Appropriate treatment is also needed for congenital disorders manifesting themselves after the neonatal period. This includes the early detection and treatment with rehabilitation services. Ministry of Health and Family Welfare (MoHFW), Government of India has made a provision for prevention, early diagnosis and management of birth defects under India Newborn Action Plan (INAP), 2014 along with basic mother and child care [6].

Birth prevalence of congenital anomalies in the developing countries is actually underestimated due to poor registry, lack of diagnostic techniques and their reliability. Like other low and middle income countries, still, congenital anomalies are not considered as a major problem in India. But literature search reveals that India has the highest number of children with birth defects [7].

Hence, based on the above literature findings, present study was planned to generate a systematic data on the magnitude of congenital anomalies, their pattern of prevalence, healthcare impact and impact on neonatal health. Prevalence studies give an idea about the pattern of occurrence of anomalies in different places, changes over a period of time and also give some clues to identify the aetiology.

Methodology

The study was planned in Department of Paediatrics, Darbhanga Medical College and Hospital, Laheriasarai. 100 neonates identified with birth defects from May 2017 to Feb 2018 were enrolled in the present study. Approval of the institutional ethical committee was taken prior to conduct of the study. The aim and the objective of the present study was conveyed to parents of the neonates.

Detailed general and systemic examinations of the babies were carried out within 24 hours of birth and only visible anomalies were noted in stillborn by two paediatricians separately for each baby. All live born babies were further followed for 72 hours. The new-born babies who required intensive care were shifted to NICU of same institute.

Ultrasound abdomen, 2D-Echo, neuro sonogram and X-rays were done to detect internal anomalies. CT/ MRI brain were done whenever required. Anomalies were divided into central nervous system (CNS), musculoskeletal, gastrointestinal, genitourinary, cardiovascular system (CVS), and miscellaneous disorders.

Following was the inclusion criteria of the present study:

1. Antenatal women registered in /referred to our outpatient department who were detected to have structural anomalies in foetus after gestational age of 24 weeks (calculated by LMP and /or first trimester USG).
2. All women who gave birth to babies with structural defect after 24 weeks of gestational age.

Results & Discussion

Data from 100 neonates were collected and is presented as below. Based on the findings, the data is further discussed.

Table 1: Demographic & General Variables

Variable	Number of Cases
No. of Cases	100
Mother age at birth	
Below 20 years	65
Above 22 years	35
Mother BMI	
Non Underweight	79
Underweight	21
No. of Children's	
1-2	86
More than 3	14
Gender	
Male	69
Female	31
Birth Weight	
Less than 2 kg	95
More than 2 kg	5

Table 2: Distribution of Congenital Malformations in Newborns

Congenital Malformations	Number of Cases
Musculoskeletal	38
Central nervous System	23
Cardiovascular	19
Genitourinary tract	11
Gastrointestinal	5
Miscellaneous	3
Multiple	1
Total	100

The true incidence of congenital malformations depends upon several factors and no two studies are strictly comparable. It depends upon ethnic background, population sample (hospital or community based, live birth or total birth), nature of study (prospective or retrospective), age at the time of diagnosis ,duration of follow up , autopsy rate, diagnostic facility available and enthusiasm and acuteness of physician. In the present series, low incidence in comparison to other studies is possibly because of malformations only present at birth were included. All those malformations recognized as result of autopsy study or which were diagnosed later on was excluded. The World Health Assembly at their 2010 meeting passed a resolution urging member states to raise awareness about the importance of birth defects as the cause of child morbidity and mortality [8].

The annual report of Indian Council of Medical Research says that the commonest congenital anomalies are cardiac in nature (0.57%) [9]. Cardiovascular anomalies were less in our study which could be due to lack of autopsies in stillborn born babies. The relative difference in the occurrence of various anomalies might be due to geographic and racial difference. True incidence of CA depends on several factors and therefore two studies are never strictly comparable.

Goravalingappa & Nashi [10] and Guha AK [11] also found high incidence of central nervous system malformations. While Mishra PC & Baveja R [12] found high incidence of multiple congenital anomalies. Ghose *et al.* [13] and Mohanty *et al.* [14] found higher incidence of musculoskeletal system malformations.

The prevalence of anomalies in this study is similar to other studies done in different parts of the country. A study on congenital anomalies in a referral center in Maharashtra showed an incidence of 1.9% [15]. Another study showed overall incidence of malformations at birth as 3.7%, with 3.2% among live births and 15.7% among stillbirths [16]. The trends over the past 10 years have not showed much variation in the prevalence, which indicates that knowledge and detection have remained more or less the same among the rural population.

The etiology of congenital birth defects remains mostly unknown. There are many causes implicated like chromosomal abnormalities, genetic defects and some environmental factors. There have been studies to look at the proportion of cases actually attributable to known risk factors. They looked at NTDs and suspected risk factors and concluded that known risk factors contribute to <50% of NTD cases [17].

One of the limitations of this study is that there is a well-known association between folic acid deficiency and neural tube defects. Serum and blood folate levels could not be determined due to their high cost. Also, a definitive diagnosis of chromosomal abnormalities could not be made because of the lack of availability of these appropriate tests. Since this study was a cross-sectional descriptive study, the findings may not be projected to the entire population. Nevertheless, these results emphasize an important public health issue and present a baseline for other well-designed studies.

Congenital anomalies are important causes of fetal deaths; thus, it becomes mandatory to determine the incidence and prevalence of congenital abnormalities in society. The present study demonstrated a high frequency of congenital malformations in the young age group and especially among primigravida women. The most frequently reported risk factor was consanguineous marriage. CNS malformations were the most prevalent anomaly detected. Early prenatal diagnosis is therefore very helpful in decreasing perinatal mortality by allowing for the option of early termination of pregnancy. Further studies are required to evaluate interventions that may be oriented to eliminate risk factors and reduce the incidence of congenital anomalies.

Conclusion

From present study, it can be concluded that congenital anomalies in new-borns are significantly associated with fetal factors like still birth, prematurity and low birth weight. The incidence of congenital anomalies can be reduced by following prevention strategies including primary, secondary and tertiary levels of prevention. These strategies are applicable at various stages of pregnancy that is preconception, antenatal and postnatal period.

References

1. What are the types of birth defects?. www. Nichd.nih.gov. Retrieved, 2017.
2. How do health care providers diagnose birth defects?. Www.nichd.nih.gov. Retrieved 8 December 2017.
3. http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf
4. https://www.nhp.gov.in/disease/gynaecology-and-obstetrics/congenital-anomalies-birth-defects
5. http://nrhm.gov.in/images/pdf/programmes/inap-final.pdf
6. https://www.who.int/en/news-room/fact-sheets/detail/congenital-anomalies
7. Kar A. Preventing Birth Defects in India. Econo Politic Week. 2011; 46:48.
8. WHO. Birth Defects. Sixty-Third World Health Assembly. Agenda item 11.7. [Internet]. 2012 [cited 2012 Feb]. Available from: http://apps.who.int/gb/ebwha/ pdf_files/WHA63/ A63_R17-en.pdf
9. ICMR, New Delhi: Reproductive health; Annual report, 2002-03, 91.
10. Goravalingappa JP, Nashi HK. Congenital Anomalies in a study of 2398 consecutive births. Indian Journal of medical research. 1979; 69:140.
11. Guha DK. Neonatology First Edition, 1995, 466.
12. Mishra PC, Baveja R. Congenital anomalies in New borns A Prospective study .Indian Pediatrics, 1989, 26.
13. Ghosh S, Bhargava SK, Bhtani R. Congenital anomalies in longitudinally studied birth cohort in a urban

community. Indian j. Med. Res. Nov. 1985; 82:427.
14. Mohanty C, *et al*. Congenital Anomalies in new born A study of 10,874 consecutive birth.J. Anatomical Society of India. !989; 38:2.
15. Taksande A, Vilhekar K, Chaturvedi P, *et al*. Congenital malformations at birth in Central India: a rural medical college hospital based data. Indian J Hum Genet. 2010; 16:159.
16. Agopian AJ, Tinker SC, Lupo PJ, *et al*. National Birth Defects Prevention Study. Proportion of neural tube defects attributable to known risk factors. Birth Defects Res A Clin Mol Teratol. 2013; 97:42-6.
17. Bhat BV, Babu L. Congenital malformations at birth–a prospective study from south India. Indian J Pediatr. 1998; 65:873-81.