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Prevalence and pattern of birth defects in a cross-sectional study at a teaching hospital in Eastern India

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Abstract

Objective: Birth defects or congenital anomalies are defined as abnormalities of body structure or function which are present at birth and are of prenatal origin. The aim of this study is to estimate the magnitude of birth defects and to analyze system-wise pattern and associated epidemiological factors.

Methods: This is a hospital based cross-sectional study, conducted in the department of obstetrics and gynaecology in Rajendra Institute of Medical Sciences, Ranchi over a period of March 2018 to August 2019. During the study period, all structural birth defects identified at birth either in a liveborn or stillborn or in an abortus were included.

Results: Out of total 15402 consecutive births during the study period, 109 fetuses and newborns had structural birth defect, giving the prevalence of 7.1 per 1000. 37.6% defects were not diagnosed antenatally. Central nervous system (CNS) defect was most common anomaly (56%), followed by musculoskeletal defect (23%). Anencephaly was commonest among CNS defect and cleft lip with cleft palate was commonest among musculoskeletal defect. CNS defect was most common anomaly observed among stillborn whereas musculoskeletal defect was most common anomaly seen among liveborn. Majority (71.6%) delivered vaginally. Male:female sex ratio was 1.33:1. Among 109 mothers, 29.3% had not taken folic acid in antenatal period.

Conclusion: Birth defects contribute significantly to perinatal mortality and morbidity. There is need to increase awareness about preventable measures in pre-conceptional period to decrease the perinatal morbidity and mortality, thus to decrease the burden on the family and health care system.

Keywords: Birth defects, congenital anomalies, central nervous system defect, musculoskeletal defect, prevalence

Introduction

Birth defects are structural or functional anomalies that occur during intrauterine life, identified prenatally, at birth or detected in later period. The term congenital defect, congenital anomalies and congenital malformation are synonyms. Birth defects are worrisome for the parents, contribute to long term disability and have a significant impact on family, society and health care system. March of Dimes report shows, about 7.9 million children (6% of total birth) are delivered with serious birth defects worldwide every year and 94% of these occur in middle and low economic countries. Birth defects account for 7% of all neonatal mortality and 3.3 million under 5 death in world.[1,2] In India, the prevalence of birth defects varies from 61-69.9/1000 live birth and accounts for 8-15% perinatal death and 13-16% neonatal death.[1,2] Birth defects registry in India (BDRI) has also analyzed over 0.7 million births with defects and neural tube defect is the most common among all. These defects can occur for many reasons including inherited and environmental conditions and in 50%-60% cases, for unknown reasons. Thus, the causes are complex and appears to be multifactorial involving genetic factors, environmental factors or their interactions.[1] Resource limited countries are highly burdened with a high fertility rate, nutritional deficiencies, exposure to teratogen and several congenital infections.[1] Advanced maternal and paternal age, consanguinity, exposure to teratogens, substance abuse,

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nutritional deficiencies, numerous infections, systemic diseases like diabetes, family history of birth defects, radiation exposure are some of the risk factors. Many of these factors are preventable. A wide range of preventive and intervention approaches include Rubella vaccination, folic acid administration from pre-conceptional period, micronutrient fortification of staple food(iodine), prevention and management of syphilis, adequate antenatal care and pre-conceptional counselling and care for various genetic and systemic diseases. Most of the low-middle income countries like India, lack accurate birth defect surveillance and therefore does not have a reliable epidemiological data. There is paucity of data regarding the prevalence of birth defects in Jharkhand state. So, this study was designed to estimate the magnitude of congenital anomalies, to identify the organ system involved among the fetuses born with defects in our institution and to analyze various epidemiological factors and risk factors that could have association with birth defects. Such information is important to understand the public health burden and design prevention and management programmes in the country.

Methods

Study design and setting

This is a cross-sectional hospital-based study conducted in the department of Obstetrics and Gynaecology in Rajendra Institute of Medical Sciences, Ranchi, Jharkhand over duration of 18 months, from March 2018 to August 2019. The Institutional Ethics Committee approval [No. 71, dated 20/02/2018]. During the study period all birth defects identified at birth either in a liveborn or stillborn or in an abortus were included. Birth defects identified antenatally by ultrasonography who had delivered or undergone medical termination of pregnancy at our institution were also included. Those who delivered outside our institution were excluded from study.

Data collection

Mothers who delivered babies with birth defects were interviewed face to face according to pre-structured proforma including age, parity, gestational age, consanguinity, past obstetrical history, family history, significant antenatal history like intake of folic acid, iron, calcium, antenatal ultrasonography, history of maternal illness, infections and systemic diseases, details of ingestion of any drug, exposure to teratogen or radiation to explore possible risk factors related to birth defects. Case ascertainment or diagnosis was based on ultrasound findings (wherever available) and clinical examination of abortus and neonate from head to toe soon after delivery by attending obstetrician and paediatrician. Live born with defects that were compatible with life were sent to paediatric surgeon for further management. For sonographically diagnosed minor anomalies, opinion from paediatric surgeon was taken during the antenatal period. Informed consent obtained from mother to take pictures of the newborn. Anomalies were classified based on the International Classification of Disease (ICD-10) system.

Statistical analysis

Data was entered in a predesigned MS Excel sheet and analyzed by SPSS software (version 20). The frequency distribution, tables and graph were prepared for the variables. The prevalence of congenital anomalies was calculated in percentage and as the total number of babies (live or stillborn) with anomalies per 1000 births. Categorical variables are expressed as number and percentage. Continuous variables are expressed as mean and SD.

Results

Total number of consecutive hospital births during study period was 15,402, out of which 109 fetuses and newborns were found to have structural birth defects. Thus, the burden of the problem comes out to be 7.1 per thousand births or 0.71%.

Table 1 shows that the majority (66.1%) of the mothers belonged to of 20-30years age group. 59(54.1%) mothers were primigravida and a decreasing trend in prevalence was observed with the increase in parity. 72.5% birth defects were observed in low socio-economic status. The sex ratio of foetus was found to be 1.33:1, while one each (0.9%) had ambiguous genitalia and absent genitalia. About half of the babies with defect born at term and approx three-fourth of foetuses delivered vaginally. As depicted in Table 2, although antenatal ultrasonography was done in 86 (78.9%) cases, birth defects could not be detected in 18 cases. Antenatal ultrasonography was not done in 23(21.1%) cases.

Table 1. Demographic characteristics (n=109)

Variables	Number (%)
Maternal age	
<20yrs	30 (27.5)
21-30yrs	72 (66.1)
>30yrs	07 (6.4)
Parity	
Primigravida	59 (54.1)
Multigravida	50 (45.9)
Socioeconomic status	
Low	79 (72.5)
Middle	30 (27.6)
Gestational age	

<20wks	8 (7.3)
>20-<37wks	45 (41.3)
>37wks	56 (51.4)
Sex of newborn	
Male	61 (56)
Female	46 (42.2)
Ambiguous genitalia	01 (0.9)
Absent genitalia	01 (0.9)
Mode of delivery	
Vaginal	78 (71.6)
Caesarean	31 (28.4)

Table 2. Ultrasonography for the diagnosis of birth defects in antenatal period (n=109)

Diagnosis by Ultrasonography	Number (%)
Antenatal Ultrasonography done	86 (78.9)
Birth defects detected	68 (62.4)
Birth defects not detected	18 (16.1)
Antenatal Ultrasonography not done	23 (21.1)

Table 3 shows possible risk factors which could be associated with the occurrence of congenital anomalies. The most common factor was nutritional deficiency with lack of folic acid intake in the first trimester in 32(29.3%) cases, and all of them were related to Central nervous system (CNS) and neural tube defects (NTD). Risk factors could not be found in 32(29.3%) cases. Table 4 depicts the system-wise pattern of birth defects. 88% of babies had isolated birth defect involving single system while 12% had involvement of multiple system. The most common structural defect noted in the present study was related to central nervous system, accounting for 56% of total cases followed by musculoskeletal system (23%). Among central nervous system defects, anencephaly was the most common (18.3% of total) . As depicted in table 5, central nervous system defect was most common anomaly observed among stillborn,44(75.8%), while musculoskeletal system was most common anomaly observed among live-born, 21(48.8%).

Table 3. Risk Factors related to birth defects(n=109)

Risk Factors	Number(%)
Consanguinity	02(1.8)
Family History	07(6.4)
Lack of folic acid intake in first pregnancy	32(29.3)
Teratogen exposure	06(5.5)
History of infection	02(1.8)
History of diabetes mellitus	02(1.8)
Oligohydramnios in Ultrasonography	07(6.4)
Polyhydramnios in Ultrasonography	19(17.4)
Unknown	32(29.3)

Table 4. System-wise prevalence and distribution of birth defects (n=109)

Type of Birth defect (ICD* 10)	Number (%)	Prevalence (per/1000 births)
ISOLATED DEFECT (Single system defect)	96 (88)	
Central nervous system	61 (56)	3.9
Anencephaly (Q00.0)	20 (18.3)	
Hydrocephalus (Q03.9)	19 (17.4)	
Anencephaly+ spina bifida (Q00.0)	06 (5.5)	
Hydrocephalus+ spina bifida (Q03.9)	07 (6.4)	
Meningocele/ meningomyelocele (Q05)	05 (4.6)	
Encephalocele (Q01.9)	03 (2.7)	
Microcephaly (Q02)	01 (0.9)	
Musculo-skeletal system	25 (23)	1.6
Cleft lip+ Cleft palate (Q37.0-Q37.99)	14 (12.8)	
Talipes (Q66.0)	06 (5.5)	
Amelia, meromelia, micromelia (Q71.0-Q73.8)	04 (3.7)	
Sacrococcygeal teratoma (Q82.6)	01 (0.9)	
Gastro-intestinal system	05 (4.5)	0.32
Gastrochisis (Q79.3)	03 (2.7)	
Omphalocele (Q79.2)	01 (0.9)	
Tracheo-esophageal fistula (Q39.2)	01 (0.9)	
Cardio-vascular system	03 (2.7)	0.19
Cyanotic heart disease (Q24.9)	02 (1.8)	
Cystic hygroma (D18.1)	01 (0.9)	
Urinary system	02 (1.8)	0.13
Multicystic dysplastic kidney (Q61.02)	01 (0.9)	
Posterior urethral valve and hydronephrosis (Q62.1)	01 (0.9)	
MULTIPLE DEFECTS (Multiple system)	13 (12)	

*ICD- International Classification of diseases

Table 5. Fetal outcome among birth defects (n=109)

System affected	Liveborn (n=43) n(%)	Stillborn (n=58) n(%)	Abortus (n=8) n(%)
Central nervous system	15 (34.8)	44 (75.8)	02 (25)
Musculoskeletal	21 (48.8)	04 (6.9)	00
Gastrointestinal	02 (4.6)	01 (1.7)	02 (25)
Cardio-vascular system	01 (2.3)	01 (1.7)	01 (12.5)
Urinary system	02 (4.6)	00	00
Multiple system	02 (4.6)	08 (13.8)	03 (37.5)

Discussion

Congenital anomalies prevalence varies from country to country and within the country due to racial, social, ethnic and environmental factors. It is the leading cause of death among under five children in developed countries and becoming a major cause in developing countries owing to the fact that the prevalence of infections and malnutrition are decreasing due to better perinatal and neonatal care.

The magnitude of the structural birth defect ranges from 0.84% to 2.6% in different parts of India.[3-7] The magnitude of the problem in our study is 0.71% or 7.1 per 1000 births, which is comparatively less than previous studies. A meta-analysis of 52 hospital-based studies and 3 community-based studies conducted in India from 1960 to 2015 concluded the prevalence of congenital anomaly to be 184.48/10,000 births.[8]

The maximum cases with birth defects, 59(54.1%) was observed among primigravida which was in contrast to the other studies.[1,5,9] The maximum frequency of birth defect was observed among mothers of 21-30 years age group (66.1%), which is similar to the other studies.[1,5,9] This might be due to the fact that most of the deliveries occur in this age group, which is again a consequence of early marriage, early childbirth and high fertility rate in this age group.

41.3% deliveries with birth defect occurred at term gestation, 51.4% between 20-37 weeks. Only 8(7.3%) cases came before 20 weeks with a documented ultrasonography report and were offered medical termination of pregnancy. A study by Jayasree S, et al reported maximum deliveries at term among 911 anomalous babies in their study.[5] Other studies reported maximum presentation in preterm period.[6] 58 out of 109 (53.2%) were stillborn in our study which is consistent with findings of other study.[1] In contrast, other studies found most of the babies born with birth defects were alive.[5] This disparity may be due to the different time of presentation of mothers to hospital, type of anomaly compatible with life and quality of antenatal care given to the mothers.

69.2% mothers belonged to lower socio-economic group which is in harmony with other study.[3] This can be related to nutritional deficiency and poor antenatal care. Most of the admissions in the department belonged to the low-income group, so its correlation could be satisfactorily explained. In 41/109(37.6%) pregnant women, structural anomalies could not be detected either due to lack of routine antenatal ultrasonography in 23 women or birth defects could not be diagnosed despite routine antenatal ultrasound in 18 cases. The prenatal detection rate of birth defect by ultrasound in our study was 79.1%(68/86). This finding emphasizes the need for high quality ultrasonography by trained sonologists.

In the present work, 78 (71.6%) babies delivered vaginally, but 28.4% underwent cesarean section. Similar results was reported in a study done in Uganda [1]. Almost all cesarean sections were done for some obstetric reasons (previous cesarean with failed induction, oligohydramnios, contracted pelvis, or as life-saving intervention in hypertensive disorders, antepartum hemorrhage. The defect was unknown before delivery in some of these cases.

Our study supports male predominance with the sex ratio (male:female) of 1.33:1. It might be due to the coincidence of overall more male deliveries during the study period. The sex ratio observed in other studies were, 2:1, 1.6:1, 1.9:1 and 1.4:1 respectively.[1,5,6,9]

The pattern of structural anomalies observed in the present study is similar to the trend observed in the study by P.Bhinde et al.[8] Central nervous system defect being the commonest followed by musculoskeletal defect, gastrointestinal defect, cardiovascular system and urinary tract defect. A meta-analysis in 2015, reported 4.5/1000 births with neural tube defect, which is close to that of our study (3.9/1000).[10] Neural tube defect was also the most common anomaly observed in Kishimba R S,et al.[11] In contrast, some studies have reported musculoskeletal defect as the most common birth defect.[4,9]

Overall, anencephaly(20)and hydrocephalus(19)were most commonly observed CNS defects in our study. This finding is in coherence with that reported in other studies.[5,12] Talipes was the most common musculoskeletal defect observed in different studies,[1,9,13] as opposed to cleft lip and cleft palate being the most common in the present study. Gastrochisis and cyanotic heart disease are most common among GIT and CVS system, respectively, which is similar to other studies.[5,12] Hypospadias was observed as the most common anomaly in genitourinary system in a study[1] and few showed, pelvicalyceal dilata-

tion, PUJ obstruction and hydronephrosis as more common [5,12] while only 2 cases with urinary defect observed in our study. Isolated birth defects were more common than multiple defects, as seen in a prospective study in Nigeria.[14]

Among the associative factors, 32(29.3%) mothers had not taken folic acid tablets in the antenatal period and was associated with CNS defect (26 cases among isolated CNS defect and 6 cases among multiple defects). Evidence shows that pre-conceptional folic acid supplementation can reduce the primary incidence of neural tube defect by 62% and its recurrence by 70%.[15] No risk factors could be identified in another 32 (29.3%) cases. It can be due to recall bias or genetic causes. Six mothers gave history of intake of teratogenic dugs in 1st trimester -3 had taken antiepileptic valproic acid who delivered babies with CNS defect, 1 had taken misoprostol for abortion but later deliver a stillborn preterm baby with amelia, meromelia. Rest 2 had taken some ayurvedic medicine found to have a multiple system defect. Two (1.8%) mothers who had a history of fever in the 1st trimester and positive TORCH profile- gave birth to a baby with microcephaly and another with hydrocephalus. Two (1.8%) mothers had diabetes mellitus where one delivered the baby with atrial septal defect (ASD) and other with sacro-coccygeal teratoma. Many evidence-based studies show that diabetes in mothers found to be associated with CNS and CVS defects.

CNS defect was most common anomaly observed among stillborn while musculoskeletal system was most common anomaly observed among liveborn. This observation is similar to other studies.[8,11]

The major strength of this study is prospective nature, where all the births were thoroughly evaluated, bedside maternal interviews, examined by qualified health care providers, and real time entry of data. Retrospective studies depend on data records which may underestimate the burden of structural defects. However, our study has the limitation of hospital-based study which cannot be generalized to the population, hence more community-based and long- term studies are needed. Moreover, this study did not include genetic and metabolic causes of birth defects.

Conclusion

The magnitude of structural birth defects is 7.1 per thousand births or 0.71%. Most common anomaly observed is CNS defect, followed by musculoskeletal defect. CNS defect was most common anomaly observed among stillborn while musculoskeletal system was most common anomaly observed among liveborn. CNS defects was significantly associated with nutritional deficiency and polyhydramnios while musculoskeletal defects were associated with oligohydramnios and teratogen exposure. Although birth defects are not completely preventable, but its incidence can be reduced by a range of preventive and intervention approaches, including Rubella vaccination, folic acid administration, micronutrient fortification of staple food(iodine), prevention and management of syphilis, timely identification of a family risk of inherited disease and carrier screening with genetic counselling. There is a basic need to educate people about the importance of an early antenatal booking visit, first trimester NT scan, target scans, fetal echocardiography and biochemical tests - dual marker, quadruple markers. Anomaly scan by qualified sonologist would reduce missing out early diagnosis of birth defects. Mandatory pre-conceptional folic acid supplementation can significantly reduce the incidence of CNS or neural tube defects. Other preventive public measures could be implemented at community level like-educating adolescent girls regarding the effects of consanguineous marriage, familial diseases, substance abuse, ensuring rubella vaccination. There is a need for more surveillance at community level and proper registry system to understand the pattern and etiologies related to birth defects in India.

References

1. Mumpe-Mwanja D., et al. A hospital- based birth defects surveillance system in Kampala, Uganda. BMC Pregnancy and Childbirth 2019;19:372:1-9. [PubMed][CrossRef]
2. Christianson A, Howson CP, Modell B. The hidden toll of dying and disabled children. March of Dimes global report on birth defects;2006.
3. Shrestha S, et al. Prevalence of congenital malformations among babies delivered at a tertiary care hospital. Journal of the Nepal Medical Association 2020;58(225):310-13. [PubMed][CrossRef]
4. Baruah J, Kurse G, Bora R. Pattern of gross congenital malformations in a tertiary referral hospital in Northeast India. Indian J Pediatr.2015. [PubMed][CrossRef]
5. Jayasree S, D'Couth S. Prevalence of congenital anomalies in a tertiary care centre in North Kerala, India. Int J Reprod Contracept Obstet Gynecol 2018;7:864-9. [CrossRef]
6. Gandhi Md.K, et al. A study on incidence of congenital anomalies in newborns and their association with fetal factors: A prospective study. Int J of Res in Medical Sciences 2016;4:1200-03. [CrossRef]
7. Taksande A.et al. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian J Hum Genet.2010 Sep;16(3):159-63. [PubMed] [CrossRef]

8. Bhinde P. et al. A national estimate of the birth prevalence of congenital anomalies in India: Systemic review and Meta-analysis. BMC Pediatrics J (2018) 18:175. [PubMed] [CrossRef]

9. Sarkar S. et al. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India. J Clin Neonatol. 2013;2:131-134. [PubMed][CrossRef]

10. Allagh KP, Shananna BR, et al. Birth prevalence of neural tube defects and orofacial clefts in India: a systemic review and meta-analysis. PloS One. 2015;10(3):e0118961. [PubMed][CrossRef]

11. Kishimba R.S, et al. Birth prevalence of selected external structural birth defects at four hospitals in Dar es Salaam, Tanzania, 2011-12. Journal of Global health 2015;5(2):020411 [PubMed][CrossRef]

12. Patil SS. et al. Congenital anomalies in fetus during pregnancy- predisposing factor and management in rural set up. Int J Reprod Obstet Gynecol 2018;7:3173-8. [CrossRef]

13. Agrawal D, Mohanty BB, et al. Study of incidence and prevalence of musculoskeletal anomalies in a tertiary care hospital of eastern India. J Clin Diagn Res.2014;8(5):AC04-6. [PubMed][CrossRef]

14. Adeleye AO, Olowookere KG. Central nervous system congenital anomalies: a prospective neurological observational study from Nigeria. Congenit Anom (Kyoto) 2009;49:258-61. [PubMed][CrossRef]

15. Cunningham FG, Leveno KJ, Bloom SL, Dashe JS, Hoffmann BL, Casey BM, Spong CY, editors. Preconceptional care. In: Williams Textbook of Obstetrics, 25th edition, New York: McGraw-Hill, 146-54.