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Original Research Article

A study of congenital anomalies in a tertiary care hospital in North East region, India

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ABSTRACT

Background: Congenital anomalies are important cause of morbidity and mortality in newborns and are defined as structural and functional abnormalities including metabolic disorders present at birth. These defects are of prenatal origin resulting from defective embryogenesis or intrinsic abnormalities in the process of development and are associated with various risk factors.
Methods: Our study is a cross-sectional study done at Regional Institute of Medical Sciences, Imphal over period of one and half years from May 2016 to October 2017. Aim of study was to find out incidence of congenital anomalies and proportions of different types of congenital anomalies. Outcome was studied in relation to maternal age, religion, parity, gestational age, sex of baby, outcome and birth weight of baby.
Results: Total numbers of congenital anomalies were 91 out of 13658 births. Incidence of congenital anomalies was 0.66%. Most common congenital anomaly was cleft lip (17.6%) followed by CTEV (13.2%) and was more common among male term babies. These were most common in 18-24 years of maternal age group (34.1%) followed by 30-34 years (26.4%) and among women of parity P0-P2.
Conclusions: Congenital malformations are a major cause of still births and infant mortality. A level II targeted scan should be done at 18-20 weeks to find out anomalies and reduce the prevalence. There should be widespread education in the community regarding the common congenital malformations, their outcomes and possible available modes of treatment.

Keywords: Anomalies, Newborn, Targeted scan

INTRODUCTION

Congenital anomalies are defined as structural or functional anomalies that occur during intrauterine life. These anomalies result from defective embryogenesis or intrinsic abnormalities in process of development and are prenatal in origin.

Birth defects can be isolated abnormalities or part of a syndrome and are one of the most important cause of neonatal morbidity and mortality both in developed and developing countries.[1] In developing country like India due to the high incidence of infectious diseases, nutritional disorders and social stress, the development defects are often over shadowed, but the present scenario

is changing rapidly. Congenital anomalies accounts for 8-15% of perinatal deaths and13-16% of neonatal deaths in India.[2] There are several known factors that are associated with birth defect such as maternal TORCH infection, genetic factors, drugs, maternal age, consanguinity etc.

Due to congenital anomalies 3,03,000 newborns die within 4 weeks worldwide every year and Contribute to significant morbidity and mortality in newborns.[3] Most common congenital anomalies are cardiovascular diseases, neural tube defects and Down syndrome.

Major anomalies have serious medical, surgical and cosmetic consequences. It contributes to long term disability, which may have significant impacts on individuals, families, health-care systems and societies.

The aim of study was to find out incidence of congenital anomalies and proportion of different types of congenital anomalies among abortus, still born and live born fetuses.

METHODS

A descriptive cross-sectional study was done in Department of Obstetrics and Gynaecology, RIMS, Imphal, Manipur. This study included abortions, still births and newborns delivered at RIMS, Imphal. Data was collected from May 2016 to October 2017 using a structured form containing details of maternal age, gestation at delivery, mode of delivery, sex, birth weight and outcome of baby, parity of mother, history of congenital malformations in previous pregnancies, consanguinity. They were examined soon after birth for major and or minor congenital defect.

Diagnosis of congenital anomalies was based on clinical evaluation of new born babies by the paediatrician and consultant neonatologist. A detailed general physical and systemic examination of the babies was carried out. Antenatal ultrasonography findings were noted. Immediate outcome of all malformed babies were recorded during the period of the mothers hospital stay. No autopsy examinations were carried out.

Out of 13685 births 91 were congenital anomalies. Congenital anomalies were divided into craniospinal, vascular, musculoskeletal, gastrointestinal, genitourinary, cardiovascular and chromosomal. Data analysis was done using simple descriptive statistics.

RESULTS

In this study 91 babies out of total 13,658 births had congenital anomalies. Incidence being 0.66%, few babies had more than one congenital anomalies. Congenital anomalies were most commonly found in maternal age group of 21-30 years (48.3%) (Table 1) followed by 31-40 years (37.5%) and also more common among Hindu (71.4%) (Table 2) followed by Muslim.

Table 1: Age distribution of congenital anomalies.

Age group	No. of pregnant women	Percentage
≤ 20 years	12	13.2
21-30 years	44	48.3
31-40 years	35	37.5

Table 2: Distribution of congenital anomalies according to religion.

Religion	No. of pregnant women	Percentage
Hindu	65	71.4
Muslim	18	19.8
Christian	8	8.8

Among the study groups 50.5% cases were among primigravidas followed by primiparas which were 27.5% (Table 4).

Congenital anomalies were found to be most common among term babies with gestational age >37-40 weeks (53.8%) (Table 3) followed by the babies with gestational age 28-37 weeks (31.9%), <28 weeks (6.6%) and all were singleton births. Most of them were unbooked cases with infrequent antenatal check up without any anomaly scan. 65% of the mothers have history of previous spontaneous abortion. Among the post dated babies congenital anomalies were found to be 7.7%.

Table 3: Distribution of congenital anomalies according to gestational age.

Gestational age	No. of patients	Percentage
< 28 weeks	6	6.6
28-37 weeks	29	31.9
> 37-40 weeks	49	53.8
>40 weeks	7	7.7

Table 4: Distribution of congenital anomalies and parity.

Parity	No. of patients	Percentage
P0	46	50.5
P1	25	27.5
P2	13	14.3
≥P3	7	7.7

Distribution of congenital anomalies

In the study, male babies (57.1%) were more commonly affected than female babies (38.5%). Out of 91 congenital anomalies 4 babies have ambiguous genitalia and most of the babies (64.8%) delivered or expelled vaginally, LSCS was done in 28.6% and 6.6% were aborted. It was found that 50.5% babies were born with birth weight >2500-4000 grams followed by 29.9% of babies with birth weight >1500-2500gms, 18.7% of babies with <1500gms. 1.1% of babies have birth weigth >4000gms.

Congenital anomalies were seen more in live births, 69.2% in compared to still births which are 13.2%.

The most common congenital anomalies in the study were musculoskeletal which 50.5% is but most of them are nonfatal but are main factor contributing for perinatal morbidity. Second most common anomaly is craniospinal 19.7%. Cardiovascular anomalies were found to be least detected in this study group which is 1.09% (Table 5).

Table 5: Craniospinal anomalies.

Congenital anomalies	No. of babies
Anencephaly	8
Encephalocele	2
Meningocele	2
Hydrocephalus	3
Holoprosencephaly	1
Dandy walker malformation	1
Microcephaly	1

Table 6: Vascular anomalies.

Vascular anomalies	No. of babies
Capillary hemangioma	1

Table 7: Cardiovascular.

Cardiovascular	No. of babies
Left hypoplastic ventricle	1

Table 8: Musculoskeletal anomalies.

Congenital anomalies	No. of babies
Cleft lip	4
Cleft lip and Cleft palate	12
Cleft palate	1
CTEV	12
Polydactyly	4
Syndactyly	1
Phocomelia	1
Syrenomelia	1
Limb deformities	3
Mandibular hypoplasia	1
Flat nasal bridge	1
Absent philtrum	1
Alveolar cyst	1
Periauricular sinus	1
Anotia	1
Sacrococcygeal teratoma	1

It was found that among the musculoskeletal anomalies cleft lip (17.6%) and CTEV (13.4%) were the most common while anencephaly (8.7%) was most common among craniospinal anomalies. Chromosomal anomalies, DOWN syndrome were detected in 7.6% of the cases. Incidence of gastrointestinal and genitourinary anomalies was more or less similar in this study group accounting for 9.8%.

Table 9: Gastrointestinal system.

Congenital anomalies	No. of babies
Omphalocele	3
Gastroschisis	4
Imperforate anus	1
Diaphragmatic hernia	1

Table 10: Genitourinary system.

Congenital anomalies	No. of babies
Micropenis	2
Undescended testis	1
Bifid scrotum	1
Paraphimosis	1
Ambiguous genitilia	4

Table 11: Chromosomal anomalies.

Congenital anomalies	No. of babies
Down's Syndrome	**7**

About 80% of the babies were compatible with life 20% were non compatible. The increase in perinatal mortality was mainly due to associated preterm labour, prematurity and IUGR. The incidence of CNS anomalies was higher in the stillborn. History of consanguinity was seen in 3 out of 91 patients.

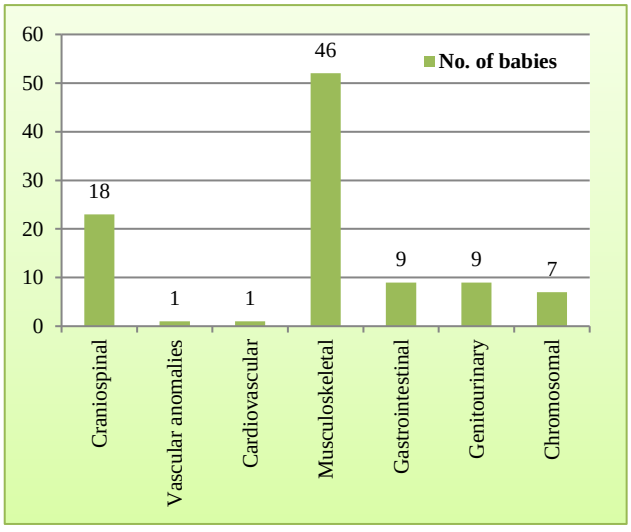

Figure 1: Gross distribution of congenital anomalies.

Table 12: Sex wise distribution of congenital anomalies.

Sex of babies	No. of babies	percentage
Male	52	57.1
Female	35	38.5
Ambiguous	4	4.4

Figure 2: Anencephaly and gastroschisis.

Social awareness about consanguinity, preconceptional counselling, supplementation of folic acid, targeted level II scan can be done to reduce the incidence of congenital anomalies.

Table 13: Distribution of congenital anomalies and birth weight of babies.

Birth weight (grams)	No. of babies	Percentage
≤ 1500	17	18.7
> 1500-2500	27	29.7
>2500-4000	46	50.5
> 4000	1	1.1

Table 14: Mode of delivery among anomalous babies.

Mode of delivery	No. of patients	Percentage
Vaginal delivery/ expulsion	59	64.8
LSCS	26	28.6
Abortion	6	6.6

Table 15: Outcome of anomalous babies.

Outcome	No. of patients	Percentage
Alive	63	69.2
Intrauterine death	7	7.6
Perinatal death	9	9.9
Still birth	12	13.2

DISCUSSION

Congenital anomalies are important causes of still births and infant mortality, and are contributors to childhood morbidity. The pattern and prevalence of congenital anomalies may vary from time to time or with geographical location or racial differences.[4] With improved infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India.

Incidence of congenital anomalies in our study is 0.66% which is more or less comparable to other studies in different parts of the country like Kokate P et al, Rani MS et al and Chowdhary P et al where the incidences were 0.9%, 0.9% and 1.06% respectively but it contradicts to Pabbati J et al where it is 4.08%.[5-8] Most common congenital anomalies in this study are musculoskeletal followed by craniospinal, gastrointestinal, genitourinary etc. which is also comparable to Pabbati J et al study. But it contradicts many studies (Kokate P et al, Rani MS et al and Chowdhary P et al). The less number of neural tube defects can be explained by widespread antenatal iron folic acid prophylaxis. Male babies are more commonly affected than females and it coincides with other studies from India as well as outside. This can be explained by more lethal malformation in female babies.

In our study most of congenital anomalies (80%) were compatible to life, which is also comparable to many other studies.

In this study congenital anomalies are most common among maternal age group of 21-30 years (48.3%) in comparison to most of other studies which are more common in maternal ages of >35 years, this may be explained because of the increase in the number of early marriage among the study group.[9] Previous studies have reported that significantly higher incidence of malformation among the mother of gravid 4 or more but our results contradict this as it is more common in primigravidas. The incidence of congenital anomalies was significantly higher in term babies compared to preterm babies which are not in accordance with many previous studies reported from this country. This different in pattern and prevalence may be explained by geographical location, environmental and genetic factors, socio cultural, racial and ethnic variables. Consanguineous marriages are reported to play a major role in the occurrence of congenital anomalies.[10] In the present study also 3 out of 91anomalies are detected in consanguineous marriage.

Incidence of congenital malformed babies appears more nowadays as compared to past because of advanced diagnostic facilities and availability of neonatal intensive care unit which lead to increase chances of survival of malformed babies. Increasing awareness about prenatal care is the need of the hour.

CONCLUSION

As congenital anomalies are the major cause of still births and infant mortality the importance of regular ANCs should be understood by pregnant ladies, and prenatal screening for congenital anomalies should be done. If

possible high risk pregnancies should be identified. A targeted level II scan should be done at 18-20 weeks to exclude anomalies.

Once anomaly is detected, discussions of various management options have to be done with parents, neonatologist, paediatric surgeon and neurosurgeon when necessary. Termination of pregnancy is a better option in case of lethal congenital anomalies.

Routine screening should be done even in low risk women since a cost of routine screening is not more than burden of a severely morbid and disabled child on family and society.

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