International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Praneshwari RK et al. Int J Reprod Contracept Obstet Gynecol. 2019 Aug;8(8):3295-3299
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: http://dx.doi.org/10.18203/2320-1770.ijrcog20193554

Original Research Article

Congenital anomalies in a tertiary care hospital in North East region, India

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Received: 01 March 2019
Revised: 15 June 2019
Accepted: 06 July 2019

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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 3 years from January 2015 to December 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, and gestational age, sex of the baby, outcome and sex of the baby.
Results: Total numbers of congenital anomalies were 257 babies out of 29879 births giving the incidence of 0.86%. Most common congenital anomalies in this study are musculoskeletal followed by craniospinal, genitourinary, cardiovascular and gastrointestinal. It was more common in preterm babies and parity 1-3, more common in 21-30 years of maternal age. Consanguinity was seen in 7 out of 257 patients.
Conclusions: Congenital malformations are a major cause of still births and infant mortality. Targeted scan should be done at 18-20 week 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 mode of treatment.

Keywords: Musculoskeletal, Congenital anomalies, Consanguinity, Craniospinal

INTRODUCTION

Congenital anomalies are defined as structural or functional anomalies that occur during intrauterine life. These anomalies results from defective embryogenesis or intrinsic abnormalities in the process of development and are prenatal in origin.[1] According to WHO an estimated 3,03,000 members die within 4 weeks of birth every year worldwide due to congenital anomalies. Congenital anomalies can contribute to long term disability, which may have significant impacts on individuals, families, health care systems and societies. The most common severe congenital anomalies are heart defects, neural tube defects and down syndrome.[1] Although congenital anomalies may be the result of one or more genetic, infections, nutritional or environmental factors, it is often

difficult to identify the exact causes. Some congenital anomalies can be prevented. Vaccination, adequate intake of folic acid or iodine through fortifications of staple foods or supplementation and adequate antenatal care are just 3 examples of prevention methods. Approximately 50% of all congenital anomalies cannot be looked to a specific cause, there are some genetic, environmental and other causes or risk factors.

METHODS

This is a cross-sectional study done in the Department of Obstetrics and Gynaecology, RIMS, Imphal, Manipur during three years i.e. January 2015 to December 2017. Data are collected using a structured form containing details of material age, gestation at delivery, mode of delivery, sex, birth weight and outcome of baby, parity of mother, and history of congenital malformations in previous pregnancies history of irradiations or teratogenic, drug exposure or consanguinity. Abortions, stillbirths and newborns were included in study. Data was analysed using simple descriptive statistics. They were examined soon after birth or major and minor congenital defect.

Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the pediatrician and consultant neonatologist. A detailed general physical and systemic examination of the babies was carried out. The ultrasound findings were noted. Outcome of all malformed babies were recorded during the period of the mothers hospital stay. No autopsy examinations were carried out.

RESULTS

In the present study, 257 0ut of 29879 babies born had congenital anomalies. Incidence thus comes to 0.86%.

Regarding the age distribution of the mothers with anomalous babies, women in the age group of 21-30 years had the highest number of congenital defects (61.9%) followed by those more than 30 years of age (29.2%) (Table 1).

Table 1: Distribution of congenital anomalies according to age.

Age of group	No. of pregnant women	Percentage
≤ 20 years	23	8.9
21-30 years	159	61.9
> 30 years	75	29.2

Congenital anomalies were more common (51.8%) in the women with para 1, para 2 and para 3 as a whole (P1-3), followed by the nulliparous women, that is P0 (36.6%) followed by those with parity more than 3 (11.6%) (Table 2). Preterm babies were having the maximum number of congenital anomalies (49.8%) followed by term babies (46.67%) while abortions and post-term babies constituted 2.35% and 1.18% cases respectively (Table 3).

Table 2: Distribution of congenital anomalies and parity.

Parity	No. of patients	Percentage
P_0	94	36.6
P_{1-3}	133	51.8
$>P_3$	30	11.6

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

Gestational age	No. of patients	Percentage
Abortion	6	2.35
Preterm	127	49.8
Term	119	46.67
Post- term	5	1.18

Distribution of various congenital anomalies

Most common congenital anomalies were musculoskeletal (139/257) is the highest followed by craniospinal anomalies (57/257) and genitourinary system (20/257).

Table 4: Distribution of musculoskeletal anomalies.

Congenital anomalies	No. of babies	Percentage
CTEV	37	26.62
Cleft lip	33	23.74
Polydactyly	23	16.55
Cleft palate	16	11.51
Cleft lip and Cleft palate	13	9.35
Limb deformities	6	4.31
Phocomelia	1	0.72
Syrenomelia	1	0.72
Mandibular hypoplasia	1	0.72
Flat nasal bridge	1	0.72
Absent philtrum	1	0.72
Alveolar cyst	1	0.72
Periauricular sinus	1	0.72
Anotia	1	0.72
Sacrococcygealteratoma	1	0.72
Hemimelia	1	0.72
Arthrogyposiscongenita	1	0.72
Total	**139**	**100**

Among the musculoskeletal anomalies CTEV (26.62%) and cleft lip (23.74%) were the most common followed by polydactyly (16.55%), cleft lip ad cleft palate together (9.35%), limb deformitied (4.31%) and one case (0.72%) each of phocomelia, sirenomelia, mandibular hypoplasia, flat nasal bridge, absent philtrum, alveolar cyst, periauricular sinus, anotis, sacrococcygealteratoma,

hemimelia, arthrogryposis congenital have been noted (Table 4).

Table 5: Distribution of craniospinal anomalies.

Congenital anomalies	No. of babies	Percentage
Hydrocephalus	28	49.12
Meningocele	10	17.54
Anenceohaly	8	14.04
Encephalocele	7	12.29
Microcephaly	2	3.51
Holoprosencephaly	1	1.75
Dandy walker malformation	1	1.75
Total	**57**	**100**

Craniospinal anomalies constituted 22.17% of the congenital anomalies. The most common was found to be hydrocephalus 49.12% followed by meningocele (17.54%), anencephaly (14.04%), encephaocele 7 (12.29%). Microcephaly (3.51%), and one case (1.75%) each of holoprosencephaly and dandy walker malformation were found (Table 5).

Table 6: Distribution of dermatological anomalies.

Congenital anomalies	No. of babies	Percentage
Capillary hemangioma	5	83.33
Accessory nipple	1	16.67
Total	**6**	**100**

Dermatological anomalies constituted 23% of all congenital anomalies. Among all the dermatological anomalies capillary haemangioma is the commonest constituting 83% followed by accessory nipple (15%) (Table 6).

Table 7: Distribution of cardiovascular anomalies.

Congenital anomalies	No. of babies	Percentage (%)
Cyanotic heart disease	9	81.82
Left hypoplastic ventricle	1	9.09
Acyanotic heart disease	1	9.09
Total	**11**	**100**

Cardiovascular anomalies constituted 4.28% of all cases most common being cyanotic heart disease (81.81%) each of left hypoplastiv ventrivle and acyanotic heart disease constituted 9.09% of the cardiovascular anomalies (Table 7).

Genitourinary anomalies accounted for 3.5% of all congenital anomalies most commonly hypospadias (55%), followed by micropenis (25%) and undescended testis (10%). Each of bifid scrotum and paraphimosis accounted for 5% of the cases (Table 8).

Table 8: Distribution of genitourinary anomalies.

Congenital anomalies	No. of babies	Percentage
Hypospadias	11	55
Micropenis	5	25
Undescended testis	2	10
Bifid scrotum	1	5
Paraphimosis	1	5
Total	**20**	**100**

Respiratory anomalies accounted for 0.77% of all the congenital anomalies. Diaphragmatic hernia and Tracheo-esophageal atresia constituted 50% each for the respiratory anomalies (Table 9).

Table 9: Distribution of respiratory anomalies.

Congenital anomalies	No. of babies	Percentage
Diaphragmatic hernia	1	50
Tracheo-esophageal atresia	1	50
Total	**2**	**100**

Opthalmological anomalies also accounted for 0.77% of all anomalies. Anopthalamia and congenital cataract occupy 50% each of the opthalmological anomalies (Table 10).

Table 10: Distribution of opthalmological anomalies.

Congenital anomalies	No. of babies	Percentage
Anopthalamia	1	50
Congenital cataract	1	50
Total	**2**	**100**

It was observed that gastrointestinal anomalies were seen in 3.89% cases. Gastrochiasis was found to be the most common gastrointestinal anomaly (60%). Omphalocele constituted 30% cases and imperforate anus constituted 10% of gastrointestinal anomalies (Table 11).

Table 11: Distribution of gastrointestinal system.

Congenital anomalies	No. of babies	Percentage
Gastroschisis	6	60
Omphalocele	3	30
Imperforate anus	1	10
Total	**10**	**100**

Chromosomal anomalies were seen in 3.89% of all cases of congenital defect. Out of total chromosomal anomalies Drown syndrome constituted 90% of the chromosomal anomalies followed by Turner syndrome (Table 12).

Congenital anomalies were most commonly found in males (61.5%) followed by female (35.8%) and lastly by ambiguous sex (2.7%) (Table 13).

Table 12: Distribution of chromosomal anomalies.

Congenital anomalies	No. of babies	Percentage
Down syndrome	9	90
Turner syndrome	1	10
Total	**10**	**100**

Table 13: Sex wise distribution of congenital anomalies.

Sex of babies	No. of babies	Percentage
Male	158	61.5
Female	92	35.8
Ambiguous	7	2.7

It was observed that 35.3% of the anomalous babies were born with birth weight >2500 gram, 32.7% with birth weight <1500 gram and 32% with birth weight between 1500 and 2500 grams (Table 14).

Table 14: Distribution of congenital anomalies and birth weight of babies.

Birth weight (grams)	No. of babies	Percentage
>2500	91	35.3
<1500	84	32.7
1500-2500	82	32.0

Most of the congenital defects are preterm diagnosed by ultrasonography. So the maximum mode of delivery is vaginally accounting to 74.7%. But 25.3% has to undergo LSCS as indicated by obstetric indication (Table 15).

Table 15: Mode of delivery among anomalies babies.

Mode of delivery	No. of patients	Percentage
Vaginal delivery/ expulsion	192	74.7
LSCS	65	25.3

Regarding the outcome of anomalies babies 220 are alive at birth i.e. 85.6%. Stillbirth accounts for 7.8% followed by perinatal death (3.5%) and intrauterine death (3.1%) (Table 16).

Table 16: Outcome of anomalies babies.

Outcome	No. of patients	Percentage
Alive	220	85.6
Still birth	20	7.8
Perinatal death	9	3.5
Intrauterine death	8	3.1

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.[2] Birth defects may result from genetic or chromosomal disorders, exposure to certain medications or chemicals or certain infections during pregnancy.[3]

Risk factors include folate deficiency drinking alcohol or smoking during pregnancy, poorly control diabetes and a mother over the age of 35 years old.[4] With improved infective and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India. Birth defects may be visible at birth or diagnose by screening test.[5]

Incidence of congenital anomalies in our study is 0.86% which is more or less comparable to other studies in different part of country. Kokate P et al, Rani MS et al, and Chowdhary P et al, had incidences of 0.9%, 0.9% and 1.06% respectively in contrast to Pabbati J et al, where it is 4.08% and 3% in United State.[6-11] Although we got nearly the same result as reported in other studies, the prevalence of congenital anomaly have been more than the present rate if it was a community based study and not nearly in a tertiary care setup. Most common congenital anomalies in this study are musculoskeletal followed by craniospinal, genitourinary, cardiovascular and gastrointestinal which is comparable to Pabbati J et al study.[10] Most of congenital anomalies (69.2%) are compatible to life, which is comparable to many studies.

In this study congenital anomalies are most common among maternal age group of 21-30 years (61.9%) 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.[12] Previous studies have reported that significantly higher incidence of malformation among the mother of gravid 4 or more but our result contradict this as it is more common in primigravidae. The incidence of congenital anomalies was significantly higher in term babies which are not in accordance with many previous studies reported in our country. This is may be explained by geographical location, environmental and genetic factors, socio cultural, racial and ethnic variables. Consanguinity is seen in 7 out of 2-57 mothers. 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.

CONCLUSION

Higher risk pregnancies should be identified in order to have conventional prenatal screening. Pregnant mother should be counselled to know the importance of regular ANCs. A targeted level (II) scan should be done at 18-20 weeks. 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 conditions. 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.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: The study was approved by the Institutional Ethics Committee

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Cite this article as: Praneshwari RK, Singh NN, Devi AT, Priya J, Singh LR. Congenital anomalies in a tertiary care hospital in north east region, India. Int J Reprod Contracept Obstet Gynecol. 2019;8:3295-9.