



CONGENITAL ANOMALIES IN NEONATES AND ASSOCIATED RISK FACTORS IN A TERTIARY CARE HOSPITAL: A SINGLE CENTER STUDY FROM INDIA

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ABSTRACT **Background:** 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. The objective of this study was to determine the proportion and pattern of congenital anomalies in live newborns in our area and to study the associated maternal and perinatal risk factors.

Materials and Methods: This was a prospective observational study and was carried out in the neonatal care unit of a large tertiary care Hospital in Bhubaneswar, Odisha, India from August 2015 to July 2016. All the babies who were born with congenital anomalies during this period in our hospital (inborn) and those entire babies who were brought to outdoor or emergency (outborn) were included. The patients having congenital anomalies were examined with detail by consultant paediatrician and neonatologist before documentation. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the paediatrician and consultant neonatologist. Data were entered into excel data sheet and the prevalence rate was estimated as a per cent of the total number of babies admitted in the unit within the period of the study. Data was analyzed using SPSS 20.

Results: The total number of neonates born and came to the hospital in the study time was 11867. Out of them 319 (2.9%) neonates were having some structural congenital anomaly. Male, multigravida, consanguineous, LSCS patients were having high incidence. Musculoskeletal defects were most common.

Conclusion: Public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended.

KEYWORDS : congenital anomaly, risk factors, CTEV

Introduction:

An estimated 303000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies^[1]. According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.^[1] However, as per the more recent WHO fact-sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.^[2] Structural malformations with other Congenital anomalies are one of the most important cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of Perinatal deaths and 13-16% of neonatal deaths in India.^[2,3] 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. Although congenital anomalies may be the result of one or more genetic, infectious, 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 fortification of staple foods or supplementation, and adequate antenatal care are just 3 examples of prevention methods^[1].

The objective of this study was to determine the proportion and pattern of congenital anomalies in live newborns in our area and to study the associated maternal and perinatal risk factors.

Materials and Methods:

This was a prospective observational study and was carried out in the neonatal care unit of a large tertiary care Hospital in Bhubaneswar, Odisha, India from August 2015 to July 2016. All the babies who were born with congenital anomalies during this period in our hospital (inborn) and those entire babies who were brought to outdoor or emergency (outborn) were included. All still borns were excluded from this study. All the newborns were looked for congenital malformations soon after birth and everyday during routine ward rounds. Relevant information along with antenatal, natal and postnatal history including maternal age, gestational age, sex, community, birth

weight, birth order and consanguinity was documented. The patients having congenital anomalies were examined with detail by consultant paediatrician and neonatologist before documentation. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the paediatrician and consultant neonatologist. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Antenatal ultrasonography (USG) findings were noted. Relevant radiological, histo-hematological and genetic tests were carried out. Appropriate investigations such as blood and serum analysis, radiography, ultrasonography, echocardiography and chromosomal analysis etc., System wise distribution of the anomalies was performed.

Consanguineous marriage is defined when that is found to have occurred between a male and a female who are blood-related, e.g., between brother and sister, between 1st cousins etc., According to WHO standards birth weights >2.5 kg were considered to be normal; whereas, birth weights <2.5 kg, <1.5 kg and <1kg were termed as low birth weight (LBW), very low birth weight (VLBW) and extremely low birth weight (ELBW) respectively. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as premature.

Data was entered into excel data sheet and the prevalence rate was estimated as a per cent of the total number of babies admitted in the unit within the period of the study (Number of babies with congenital abnormalities/total number of babies admitted in the hospital for the duration of study). Data was analyzed using SPSS 20. Rates and proportions were calculated with 95% confidence intervals. The proportions were compared using students *T*-test. Level of significance was set at $P < 0.05$. Ethical approval for the study, and consent to publish the clinical data obtained in the study, have been sought for from the Ethics and Research Committee of the Kalinga institute of medical science, Bhubaneswar, Odisha, India.

Results:

The total number of neonates born and came to the hospital in the study time was 11867. Out of them 319 (2.9%) neonates were having some

structural congenital anomaly. Multiple pregnancies were observed in 87 patients. Out of them 85 cases gave birth to twin babies whereas 2 mothers gave birth to triplets. From the 87 multiple pregnancies 23 were known case of induced ovulation. The incidence of anomalies in male sex was higher. (Table:1)

TABLE: 1

Variables		yes	no	total	% having cong anomaly	P value
Gender	Male	219	7807	8026	2.7%	
	Female	100	5060	5160	1.9%	
Parity of mother	Primigravida	201	7713	7924	2.7%	
	Multigravida	118	4154	4262	2.9%	
Age of mother	<20 yrs	81	2819	2900	2.8%	
	20-30 yrs	83	3981	4064	2.0%	
	30-40 yrs	109	3736	3845	2.8%	
	>40 yrs	46	1331	1377	3.4%	
consanguinity	Present	41	943	984	4.2%	
	Absent	278	11548	11826	2.3%	
Mode of delivery	NVD	234	10354	10588	2.2%	
	LSCS	85	1132	1227	6.9%	
Antenatal checkups commencement	First trimester	139	5288	5427	2.5%	
	Second trimester	132	3987	4119	3.2%	
	Third trimester	48	2273	2321	2.0%	
Family history of any deformity	present	31	1738	1769	1.7%	
	absent	288	9810	10098	2.8%	
Birth weight	>2.5 kg	57	4802	4859	1.1%	
	1.5-2.5 kg	41	2887	2928	1.4%	
	1-1.5 kg	206	2531	2737	7.5%	
	<1 kg	15	1328	1343	1.1%	
gestation	Term	81	7743	7824	1%	
	preterm	238	3805	4043	5.8%	

Among all the neonates the musculoskeletal abnormalities were most common followed by gastrointestinal problems. (Table:2)

Table: 2

Systems	Disease	numbers
Nervous systems		55
	Meningomyelocele	21
	encephalocele	06
	hydrocephalus	11
	anencephaly	2
	holoprosencephaly	2
	microcephaly	4
	Spina bifida	2
	others	3
	multiple	4
Musculoskeletal system		101
	CTEV	55
	calcanovalgus	7
	syndactyly	3
	polydactyly	10
	Absent depressor angulioris	3
	Absent pectoralis major	4
	Pterygym	3
	Osteogenic imperfecta	2
	phocomelia	2
Multiple defects	12	
Gastrointestinal System		68
	Cleft lip	21
	Cleft palate	12
	Tongue tie	4

	Imperforate anus	3
	TEF	5
	Ranula	3
	Gastrochisis	4
	omphalocele	3
	Duodenal atresia	2
	Malrotation of gut	1
	others	2
	Multiple anomalies	8
Cardiovascular system		27
	Acyanotic	18
	cyanotic	9
Respiratory system		24
	Diaphragmatic hernia	8
	Eventration of diaphragm	4
	Choanal atresia	4
	Multiple system	8
Genitourinary		21
	Hydronephrosis	8
	Ambiguous genitalia	2
	Posterior urethral valve	2
	Polycystic kidney	2
	hypospadias	1
	epispadias	2
	Bladder exstrophy	1
	others	1
	multiple	2
Skin		30
	hemangioma	18
	Skin tag	5
	others	4
	multiple	3
Syndromes		12
	Down	4
	Holt oram	1
	Edward	2
	Patau	1
	Others	4
Multiple system		33

Discussions:

The pattern and prevalence of congenital anomalies may vary over time or with geographical location. It depends upon the environmental and genetic factors including socio-cultural, racial and ethnic variables.^[4] With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India.^[5]

In the present study, the prevalence of congenital malformations in the newborns were 2.9%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.^[6,7] There are other reports from different parts of the world representing different frequency of congenital malformations.^[8,9] Although we got nearly the same result as reported in other studies, ^[6,7,8,9,10] the prevalence of congenital anomaly would have been more than the present rate. This study does not reflect the picture in the general population as this was purely a hospital based study with no attempt whatsoever to obtain a sample that would be representative of the general population. Be that as it may, it is possible that a community based study or one taking into account all deliveries occurring in the larger society may yield a higher prevalence. In our part of the world, for instance, some babies with congenital abnormalities brought to teaching or specialist hospitals do not present to the neonatology unit but are seen at other specialist units such as paediatric surgery unit or neuro-surgery unit etc. and a study conducted at the neonatology unit per se as is the case in this work may not be able to “capture” these other babies. If we could have included the abortions and stillbirths the actual incidence would have been quantified. Tertiary care hospital usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal for true estimation of incidence of congenital anomalies in a population. In the present study most common system involved was musculoskeletal system (31.6%), followed by gastro-intestinal tract (GIT) (21.3%), CNS

(17.2%), genitourinary (6.5%) etc., This was comparable with studies conducted by others.[11,12] In some studies CNS malformations like meningomyelocele and encephalocele are much more common [13] whereas Suguna Bai *et al.*[14] reported GI malformations as the most common one. The less number of neural tube defect can be explained by the universal antenatal iron folic acid prophylaxis. Male babies were more affected than female babies. It coincides with other studies from India as well as outside [5,6,7,8]. It can be explained by more lethal malformations in female.

Association of LBW with increased risk of congenital malformations is very well- documented.[5,6] Our finding is in accordance with that. The incidence of congenital anomalies was significantly higher in preterm babies as compared with the full term babies, which is in accordance with previous studies reported from this country. Mode of delivery also showed a significant association with congenital anomalies in this study with cesarean section being more commonly associated than normal delivery. Suguna Bai *et al.*[14] reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta *et al.*[15] documented statistically insignificant association of increased maternal age and congenital anomalies. The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were born of mothers aged 30-40 years; though, it was statistically insignificant. Our study reveals high incidence of congenital anomalies in multipara patients which was also apparent in previous studies.[5] Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations.[16,17,18] In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages.

Despite the high risk of recurrence of congenital malformations, there are no well-accepted preventive measures in developing countries like India. It indicates that strong preventive measures for congenital anomalies in this region are needed. Increasing awareness about familial prenatal care is the need of the hour. Need of a high sensitive screening test is well felt to decrease the malformations and to reduce the after effects of these anomalies.

Conclusion:

This study has highlighted the prevalence, types as well as several risk factors of congenital anomalies of children born in this area. As we belong to a developing country mass health education, regular antenatal visits by health workers and a highly sensitive prenatal screening method are much needed for prevention and early intervention for better future of the indexed pregnancy.

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