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Study of incidence and spectrum of congenital anomalies in newborn babies: A tertiary care hospital based study

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Abstract
Background: In a vast country like India, equitable child health, early detection and treatment of the childhood problems is of paramount importance for ensuring a healthy and dynamic future. The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes. Intrauterine diagnosis and fetal therapy has become a burning topic nowadays. Present study was conducted to determine the spectrum of congenital anomalies is our institute.
Aims and objectives: The study was carried out with the objectives to find out the incidence and patterns of congenital anomalies and to evaluate any maternal and environmental risk factor.
Materials and Methods: Hospital based observational study conducted over a period of 2 years (October 2015 to October 2017) in pediatrics and O & G dept. of SCB Medical College, Cuttack, India. The selected newborns were examined in detail and also investigations were done for the presence of congenital anomalies. Various data obtained were displayed in tables and charts. Statistical analysis of the observations were done using percentages, mean, S.D, P-value and chi-square test.
Results: Out of 1305 cases, 39 were found to have some form of congenital anomaly with an incidence of 2.98%. It was related to musculoskeletal system (33.3%) mostly followed by gastro-intestinal (23.1%). In our study males (71.8%) showed more anomalies than female babies. Mean age of the mothers was 20-30 years. Parents belonging to low socio-economic status had more chance of babies with anomaly (61.5%)
Conclusion: Proper prenatal counseling and education is required for the females. Avoidance of teratogens will help to some extent. Proper nutrition and care of pregnant women should be taken to prevent prematurity and LBW babies. Antenatal visits should be proper, screening and interventions should be early, diagnosis and surgical correction of the malformed babies offer the best chance of survival.

Keywords: congenital anomaly, new born, tertiary care hospital

Introduction: Background

Congenital anomalies can be defined as structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth or sometimes may only be detected later in infancy. In simple terms congenital refers to the existence at of before birth [1].

The etiology of congenital anomaly may be genetic or environmental. Among genetic causes, chromosomal abnormality makes up about 6%, single gene disorders about 25% and multifactorial factors 20-30%. In about 50% of cases, the cause is not known [2]. Genes play an important role in many congenital anomalies. This might be through inherited genes that code for an anomaly or resulting from sudden changes in genes known as mutations.

Consanguinity, socio-economic factors, maternal age environmental factors, infections and maternal nutrition also play a role. Early intrauterine period (between 3rd to 8th weeks of gestation) is the vital period of life for the normal development of organs. Any insult within that period may result in congenital abnormalities. It can further be argued that interventions within this period targeted at preventing insults (or removing the effects of insults) to the developing fetus will reduce the likelihood of an abnormality developing later in life.

In a vast country like India the need for ensuring a healthy and dynamic future for a large population and creating a developed society, agile and able to compete with the rest of the world, stands as of paramount importance. The dream of such healthy and developed society can be achieved through concerted efforts and initiatives undertaken in a systematic manner at all levels. Equitable child health, care and early detection and treatment can be the most pragmatic initiative or rather solution at this juncture.

The proportion of perinatal deaths due to congenital

malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centres providing good neonatal care.

Aims and Objectives

The present study was carried out with the objectives to find out the proportion and pattern of congenital anomalies in live newborns, to evaluate maternal and environmental risk factor and to study the associated perinatal risk factors.

Material and Methods

This study was a tertiary care hospital based observational study conducted over a period of 2 years (October 2015 to October 2017) at OPD and IPD of pediatrics and postnatal ward of O & G department of SCB Medical College, Cuttack, India. Institutional ethics committee of the hospital approved the study.

The newborns in OPD and IPD having visible external congenital anomalies, those delivered in OBG dept. having anomalies and all IPD newborns who on investigation showed internal anomaly were included in the study. The very sick newborns and where parents refused giving consent were excluded from the study. The newborns were examined and investigated as appropriate for the detection of any anomaly. For each case, a detailed antenatal and maternal history was obtained also. Various data obtained were displayed in tables and charts, statistical analysis of the observations were done age of percentages, mean, standard deviation, chi-square test P value less than 0.05 was considered statistically significant.

Results

During the study period total 1305 newborns were included in the study out of which 39 were found to have (2.98%) some form of congenital anomaly. Most common was CTEV followed by cleft palate (Table-1). The commonest system showing congenital anomaly was musculoskeletal followed by G.I (33.3%), 23.1%) (Table-2). Out of 32 patients who underwent imaging 23 had X-ray abnormality, 6 had abnormal neuroimaging (Table-3). Mean age at admission for patients with congenital anomaly was 3.54 ± 2.90 days and mean birth weight being 2.40kgs (Table-4). Male babies outnumbered the females in having anomaly (Table-5). Mean gestational age of mothers of babies having congenital anomaly was 36.69 ± 2.67 weeks which was lower in mothers of normal babies (Table-6). More proportion of anomalies were seen in 20-30 years age group of mothers (Table-7). Primi mothers had more newborns having anomaly and history of abortion was also significantly associated (Table-8), 25.6% of congenital anomaly babies were born to mothers having hypertension (Table-9) babies with family history of congenital disease (28.2%) and belonging to low socio-economic status (61.5%) had more chance of having congenital anomaly (Table-10). Antenatal factors like oligohydramnios and breech presentation was associated with more occurrence of anomaly (Table-11) C.Section as a mode of delivery was more found in patients (38.5%) with congenital malformation (Table-12).

Table 1: Type of congenital anomaly found in the study population

Type of congenital anomaly	Number	Percentages
Ambiguous Genitalia	2	5.1
CAHD	1	2.6
CCHD	2	5.1
Cleft Lip	4	10.3
Cleft Palate	3	7.7
CTEV	7	17.9
Diaphragmatic Hernia	1	2.6
Down Syndrome	3	7.7
Encephalocele	1	2.6
Haemangioma	3	7.7
Hydrocephalus	1	2.6
Hypospadias	1	2.6
Meningomyelocele	1	2.6
Polydactyly	2	5.1
Syndactyly	1	2.6
Skin tag	2	5.1
TEF	3	7.7
Vertebral Anomaly	1	2.6

Table 2: Distribution of system involved in patient with congenital malformation

System involved	Number	Percentages
Musculoskeletal	13	33.3
Gastro-intestinal	9	23.1
CVS	3	7.7
CNS	6	15.4
Genitourinary	3	7.7
Respiratory	6	15.4
Skin & Syndrome	7	17.9

Table 3: Imaging finding of the patients with congenital

Imaging finding	Abnormal finding (n)	Percentages
Neuroimaging (n=32)	6	18.75
ECG (n=7)	5	71.42
Abdominal USG (n=11)	6	54.54
X ray (n=32)	23	71.87

Table 4: Association of age at admission and birth weight of patients with congenital malformation

	Congenital Malformation		P value
	Present (Mean ± SD)	Absent (Mean ± SD)	
Age at admission	3.54 ± 2.90	8.02 ± 4.24	< 0.001
Birth weight	2.40 ± 0.38	2.43 ± 2.43	0.640

Table 5: Association gender and development of congenital malformation

Gender	Congenital Malformation		P value
	Present N (%)	Absent N (%)	
Male	28 (71.8)	623 (49.2)	0.005
Female	11 (28.2)	643 (50.8)	

Table 6: Association of age at admission and birth weight of patients with congenital malformation

	Congenital Malformation		P value
	Present (Mean ± SD)	Absent (Mean ± SD)	
Gestational age	36.69 ± 2.67	36.08 ± 1.53	0.018
Maternal age	30.23 ± 3.56	28.87 ± 3.38	0.013

Table 7: Maternal age group and its association with congenital malformation

Age	Congenital Malformation		P value
	Present N (%)	Absent N (%)	
20 – 30 years	25 (64.1)	1084 (85.6)	0.007
> 30 years	11 (35.9)	182 (14.4)	

Table 8: Association of obstetric history with congenital malformation

Gender	Congenital Malformation		P value
	Present N (%)	Absent N (%)	
Gravid Primi	23 (58.9)	329 (26.0)	0.001
G2 – G3	13 (33.3)	618 (48.8)	
>G3	3 (7.8)	319 (25.2)	
Parity Primi	23 (58.9)	278 (29.7)	<0.001
Multipara	16 (41.1)	659 (70.3)	
Live birth Present	26. (66.7)	739 (62.2)	0.881
Absent	13 (33.3)	449 (37.8)	
Abortion Absent	23 (58.9)	1102 (77.1)	<0.001
Present	16 (41.1)	164 (12.9)	

Table 9: Association of maternal co-morbidity with congenital malformation

Co-morbidity		Congenital Malformation		P value
		Present N (%)	Absent N (%)	
Diabetes	Present	10 (25.6)	189 (14.9)	0.049
	Absent	29 (74.4)	1077 (85.1)	
Hypertension	Present	0 (0)	13 (1.0)	0.463
	Absent	52 (100)	1253 (99.4)	
Graves' Disease	Present	0 (0)	7 (0.6)	0.591
	Absent	52 (100)	1259 (99.4)	

Table 10: Association of maternal addiction and other environmental factors

History		Congenital Malformation		P value
		Present N (%)	Absent N (%)	
Smoking	Present	1 (2.6)	103 (8.1)	0.206
	Absent	38 (97.4)	1163 (91.9)	
Alcohol	Present	2 (5.1)	51 (4.0)	0.732
	Absent	37 (94.9)	1215 (96.0)	
H/o drugs	Present	2 (5.1)	100 (7.9)	0.525
	Absent	37 (94.9)	1166 (92.1)	
Fever with rash	Present	3 (7.7)	127 (10.0)	0.631
	Absent	36 (92.3)	1139 (90.0)	
Family h/o of cong. Disease	Present	11 (28.2)	92 (7.3)	<0.001
	Absent	28 (71.8)	1174 (92.7)	
SES	Upper	1 (2.6)	426 (33.6)	<0.001
	Middle	17 (43.6)	409 (32.4)	
	Lower	21 (53.8)	463 (35.1)	

Table 11: Association of different antenatal factor with congenital malformation

Antenatal factors		Congenital Malformation		P value
		Present N (%)	Absent N (%)	
Reproductive assistance	Present	2 (5.1)	75 (5.9)	0.853
	Absent	37 (94.9)	1191 (94.1)	
Foetal movement	Present	37 (94.9)	1241 (98.0)	0.173
	Absent	2 (5.1)	25 (2.0)	
Oligohydramnios	Present	12 (30.8)	204 (16.1)	0.015
	Absent	27 (69.2)	1062 (83.9)	
Foetal presentation	Vertex	11 (28.2)	1004 (79.7)	<0.001
	Breech	28 (71.8)	255 (20.3)	

Table 12: Association of mode of delivery with congenital malformation

Antenatal factors		Congenital Malformation		P value
		Present N (%)	Absent N (%)	
Mode of delivery	NVD	24 (61.5)	870 (68.7)	0.011
	CS	15 (38.5)	272 (21.5)	
	Assisted delivery	0 (0)	124 (9.8)	

Discussion

The pattern and incidence of congenital anomalies generally varies over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables [3]. But with better control of infections and nutritional deficiency diseases, congenital anomalies have become important cause of perinatal mortality in developing countries like India [4].

In our study, the incidence of congenital anomalies is 2.98% which is higher than other studies about congenital anomalies from other parts of India and around the world, 2.2%, 2.3%, 2.9%, 2.8% [5, 6, 7, 8]. We may get higher incidence after including the abortions and stillbirths. Tertiary care hospitals usually do not have definite catchment area and serious cases are more commonly encountered. Hence, incidence 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. Nevertheless this is the only referral centre of Odisha for newborn babies so surprisingly we got a higher incidence of congenital anomalies. Still we have missed some cases because instead of coming to pediatric medicine these cases go to other departments like neuro and pediatric surgery.

Now coming to the pattern of congenital anomalies, musculoskeletal system (38.5%), followed by Gastrointestinal (19.2%), CNS (17.3%), Respiratory (15.4%) and Skin & syndrome (15.4%) were the systems most commonly involved as shown in table 2. Other systems involved were CVS and Genitourinary system, all comparable to other studies [5, 7, 9]. In some studies CNS malformations like meningomyelocele and encephalocele are much more common. Whereas Suguna Bai *et al.* [10].

Most common congenital anomaly was congenital talipes equinovarus (CTEV) i.e. 23.1%, followed by Cleft lip, Cleft palate, Down syndrome and Polydactyly and each had a incidence of 7.7% as shown in table 1. Other congenital anomalies seen in our study were Haemangioma (5.8%), CAHD, CCHD, Ambiguous genitalia, hydrocephalus, Diaphragmatic hernia, and Encephalocele.

As this is a tertiary centre and highest centre for referral in Odisha most of the newborns having congenital anomalies got referred. Age at admission for the patient with congenital anomalies was 3.56 ± 2.81 days which was significantly lower than patients without congenital anomalies (P value < 0.001) as depicted in table and figure 4..But still referral of newborn having congenital anomalies on day 4 of life is pretty late causing increased perinatal mortality rate compared to other parts of India.

As shown in other studies, [6, 7, 8, 9] the mean birth weight was lower in congenital anomalies group (2.38 kgs) compared to patients without congenital anomalies (2.43 kgs) but this

difference was not statistically significant (P value = 0.312 as shown in table and figure 4. This is a contrasting data in comparison to other reference studies mostly reason being most of the newborns are of generally LBW in this part of INDIA because of poverty and poor maternal health though in low birth weight babies congenital anomalies more common but statistical significance could not be established between weight and congenital anomalies.

More male babies with congenital anomalies than females were noted in the present study.

Male preponderance was similar to the other studies. Association of gender with development of congenital anomalies revealed that males were more prone to congenital anomalies (63.5%) compared to females (36.5%). This association was statistically significant (P value=0.044) as shown in table 5 and figure 6 it may be because of the fact that the females were afflicted with more lethal congenital anomalies and could not survive to be born with signs of life, results were similar to [5, 7, 9] described in these studies.

In our study we found that there was association of gestational age with congenital anomalies, it is more common in preterm babies. The incidence of congenital anomalies was significantly higher in preterm babies as compared with the full term babies, which is in conformity with the previous studies reported from this country. Mean gestational age of mother of babies with congenital anomalies was 35.44 ± 2.77 weeks compared to Mean gestational age of mother of babies without congenital anomalies (36.08 ± 1.53) and this difference was statistically significant (P value = 0.005). As shown in other studies [5, 7, 8, 9] (table-6).

Suguna Bai *et al.* [10] reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta *et al.* [14] documented statistically insignificant association of increased maternal age and congenital anomalies. While seeing for association with maternal age and when maternal age was divided into groups it was found that more proportion of congenital anomalies was seen in age group 20-30 years compared to normal baby's mother. This difference was statistically significant (P value = 0.007). (Table 7) and this result suggests that as maternal age is increasing chances of congenital anomalies is increasing.

In antenatal history there was association of congenital anomalies with history of abortion in previous birth suggesting lethal congenital anomalies, mostly chromosomal p value was less than 0.001 (table 7).

While seeing for association of maternal diseases with congenital anomalies, maternal morbidity like diabetes was significantly associated with congenital anomalies. 25.6% of babies with congenital anomalies had maternal diabetes while only 14.9% of normal babies had maternal diabetes and this difference of proportion was statistically significant (P value = 0.049). (Table 9) similar to Ordonez *et al.* [11].

In other antenatal history we did not get any association with hypertension and graves disease that was against previous study [11] (table 10)

There was no association of smoking, alcohol intake, with any congenital anomalies.

As opposite to study [12], the reason being most of the women here in this place of India are non-smokers and not addicted to any drugs and alcohol (table 9).

We did not get any significant association of history of fever during antenatal period with congenital anomalies contradicting to various studies mostly because of poor recall and poor educational and les antenatal checkups.

There was significant association of history of congenital anomalies in the family suggesting more of a genetic cause. It was positive in 26.9% of newborn having congenital anomalies. Patients with family history of congenital disease were more prone to develop malformation (26.9%) compared to patients without family history (7.3%). This difference in proportion was statistically significant (P value<0.001) as shown by other study [13] (table 9)

This difference in proportion was statistically significant (P value<0.001). Patients belonging to lower socio-economic status had more chances of developing congenital anomalies (61.5%) compared to upper class (3.8%) as can be explained by various nutrition deficiency poor antenatal checkups and environmental exposure. (Table 9).

In our study while looking for association of different antenatal factors with congenital anomaly there was no association with decreased fetal movements and reproductive assistance.

We got statistically significant association with breech delivery. Breech presentation was more seen in patients with congenital anomalies (67.3%) compared to normal babies (20.3%). This difference in proportion was statistically significant (P value <0.001) as shown in other study, it could be because of higher NTD as in meningococcal and other defects presentation will be mostly breech.

We found out association of oligohydramnios with congenital anomalies, it was present in 30.8% of mother suffering from Oligohydramnios compared to 16.1% in normal babies. This difference was statistically significant (P value = 0.015) (table 11) as shown in other study conducted in FRANCE.

On seeing the mode of delivery association we found that caesarean section was associated with congenital anomalies as shown in other studies. Caesarean section as a Mode of delivery was more found in patient with congenital anomalies (38.5%) as compared to normal babies (21.5%) and this difference was statistically significant (P value = 0.023). (Table 12).

There was no relation between abnormal colour of liquor, crying, sucking and NICU admission.

On investigation we found out that out of 32 subjects who underwent neuroimaging 6 patient (18.7%) had some abnormal imaging findings. Similarly, 5/7 subjects had ECG abnormality, 6/11 subjects had abnormal abdominal USG finding and 23/32 subjects had X-ray abnormality. Table 3 and figure 4 shows the imaging findings of study population with congenital anomaly as suggested by preponderance of musculoskeletal, NTD and CVS anomaly as we will get abnormal x-ray of foot in CTEV and abnormal x-ray chest in VSD and ASD etc. abnormal neuroimaging expected in NTDs.

Conclusion

This study has enlightened us about incidence, pattern and several risk factors of congenital anomalies of children seen in our area. It has also helped us to monitor the trend of congenital anomalies.

In this 21st century, while Medical Science has evolved in so many aspects along with development of a good number of effective health

Programmes by the Government, newborn care is still lagging behind in Odisha like in other parts of India.

The incidence rate obtained in this study, however, may not depict the real situation in the general population but it can give us some idea about existing problems and serve as an stimulus for further studies like this.

There are many programmes started by Government of India but they are not implemented at ground level.

More stress is to be laid upon girl's education and health so that proper prenatal counseling and education can be done.

Avoidance of known and probable teratogens should be given importance.

Proper nutrition, supplementation and care of a pregnant women should be done to prevent prematurity and low birth weight in newborns as from our study we concluded that congenital anomalies in newborns were associated with fetal factors lie prematurity and low birth weight.

Proper, regular and strict antenatal visits and sensitive prenatal screening need to be started for prevention, early intervention and even planned termination, when needed of the indexed fetus and proper diagnostic and management facilities should be given for that extensive team work is required between obstetrician and paediatrician.

By proper clinical examination, the life-threatening congenital malformation must be identified, as early diagnosis and surgical correction of the malformed babies offer the best chance for survival.

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