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BURDEN OF CONGENITAL ANOMALIES AT BIRTH AND THEIR ASSOCIATION WITH MATERNAL RISK FACTORS

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Abstract

Background: The World health organization (WHO) defines Congenital anomalies (CA) as structural or functional anomalies which are present at the time of birth. CA causes a significant proportion of infant mortality and morbidity and health care expenditure. The most common etiology is idiopathic (40-60%), followed by combination of hereditary and other factors (20%), single gene mutation (7%), chromosomal anomaly (6%) and 5% by maternal illness. It is estimated that 10–15% of CA are result of the adverse effect of environmental factors on prenatal development. This study was done to estimate incidence and spectrum of CA in various organ systems and their association with maternal risk factors. **Materials and Methods:** This prospective observational study was done for one year. All new born babies were attended by a paediatrician at birth and proper history and through examination were done and recorded in proforma. The data were entered in Microsoft excel sheet and analysed with statistical software SPSS version 20. **Result:** A total of 10299 babies were enrolled in the study. Incidence of CA was 8.8/1000 births. The mean age of mothers having babies with CA were 28.73 years. 83.5% of mothers were less than 35 years of age. 59% of the mother had male child whereas 41% had a female child. Among the study participants, 57.2% babies weighed more than 2500 grams and 42.8% weighed less than 2500 grams. The most common anomalies were GIT accounting for majority (44%), followed by CNS (34.1%) and CHD (13.2%). Among the maternal risk factors elicited for anomalous baby, 26 % had previous history of abortion (p-value <0.05), 15.5% had history of previous anomalous baby (p-value <0.05). **Conclusion:** Incidence of CA was 8.8/1000 births. GIT, CNS and CVS were most commonly affected. Dominant maternal risk factors for anomalous babies were previous history of abortion and previous anomalous baby.

INTRODUCTION

The World health organization (WHO) defines Congenital anomalies (CA) as structural or functional anomalies including metabolic disorders which are present at the time of birth.1 CA are also known as birth defects, congenital disorders or congenital malformations. CA are the major cause of new born deaths within four weeks of birth and can result in long- term disability with a significant impact on individuals, families, societies and health-care systems.2 Major congenital abnormality is defined as those defects which cause serious structural, cosmetic and functional disability requiring surgical or medical management. Minor congenital abnormality may be defined as unusual morphologic features that are of no serious medical or cosmetic consequences.3 CA is a common cause of morbidity and mortality not only in the new-born but also in childhood and beyond. Many CA result in social stigma and discrimination, which can lead to embarrassment, isolation and other reductions in community interaction. CA is due to defective organogenesis during the early foetal life and can affect any organ system of body. The most common

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etiology of CA is idiopathic (40-60%), followed by combination of hereditary and other factors (20%), single gene mutation (7%), chromosomal anomaly (6%) and 5% by maternal illness.4 The worldwide incidence of CA is estimated 3 to 7% but actual incidence varies between countries.5

Various studies done across the globe to find out the incidence and various risk factors associated with CA and got different results. In this study, we shall endeavor to estimate the overall burden and spectrum of CA both in live-born and stillborn and to evaluate the association of various maternal risk factors. It will also help to classify the etiology of malformations in neonates to allow proper counseling, initiation of early management to cure or halt its progression thus reducing disease burden, complication, physical disability, cost of treatment and stress on patient and family.

MATERIALS AND METHODS

This prospective study was conducted from January 2020 to December 2020 in department of Paediatrics, Jawahar Lal Nehru Medical College, Ajmer, Rajasthan, India. Ethical and scientific committee of our institute approved the study. Verbal consent was taken from each parent. We included all live born and stillborn babies with malformations in this study and all out born and intrauterine deaths were excluded.

Data collection - All mothers were interviewed before birth as per proforma which contain age of parents, last menstrual period (LMP), consanguinity, exposure to radiation, chemical that include teratogenic drugs and exposure to infection during first trimester of pregnancy. Detailed obstetric history with reference to previous abortions and still birth, oligo or polyhydromios and medical disease during pregnancy like diabetes, hypertension, eclampsia, anemia, rheumatic heart disease, liver or kidney disease were recorded. All routine and relevant investigations like CBC, urine analysis , Rh blood grouping and typing, HBsAg, VDRL and HIV were also recorded. Any foetal malformation reported in mother sonography was noted. All new born babies were attended by a paediatrician at birth and proper history, physical and systemic examinations were done as per proforma that includes birth weight, sex, lives born or still born, gestational age and details of congenital malformation. Detailed examination of umbilical cord and placenta were done for any structural defect. At the time of discharge every newborn was re-examined for CA. A gavage tube was used to check choanal atresia, oesophageal atresia and anorectal anomaly in suspected cases. All relevant investigations for baby were done whenever required. SET pulse oximetry was done for all newborn babies. ECG, X ray chest, USG abdomen and echocardiography were done when ever required.

Statistical analysis – The collected data were entered in Microsoft excel sheet and analyzed using statistical software SPSS version 20. Descriptive statistics given by mean, standard deviation, frequency and percentages. Chi-square test is used to find association between a categorical predictor and the outcome. P-value <0.05 is considered to be significant throughout the study.

RESULTS

A total of 10299 babies were present in this study. Out of these, 91 babies had one or the other CA. The incidence of CA in this study was 8.8/1000 live births. 59% of the mother had male child whereas 41% had a female child. Among the study participants, 57.2% babies weighed more than 2500 grams and 42.8% weighed less than 2500 grams and mean birth weight was 2.460 kg. Among mothers with congenital anomalous babies, 70.3% had delivered full-term and 29.7% had preterm babies. 57.1% mother's had vaginal delivery and 43.1 % had LSCS.

The mean age of mothers having babies with CA was 28.73 years. 83.5% of mothers were less than 35 years of age. In babies with congenital anomalies, 71.4% of their mothers had age more than or equal to 35 years. There is a significant association between Age and CA with a p-value<0.05.

Among the maternal risk factors 26.4% of the mothers had a previous history of abortion and 15.4% had previous history of congenital malformation. In babies with CA, 55.5 % of their mothers had a previous history of abortion (P value 0.0015) and 82.3% had previous congenital malformed babies (P value 0.001). These were two major maternal risk factors observed for CA.

In our study, the most common anomalies were GIT accounting for majority (44%), followed by CNS (34.1%) and congenital heart disease (13.2%). Among the 44% GIT anomalies, the four most reported anomalies were imperforate anus (9.9%), trachea-oesophageal fistula (9.9%), cleft lip and cleft palate (6.6%) and cleft palate (6.6%). Others were cleft lip (2.2%), duodenal atresia (2.2%), omphalocele (3.3%), and gastroschisis (1%). Among the CNS malformations, the most common were myelomeningocele (15.3%) and hydrocephalus (14%), encephalocele (3.3%) and anencephaly was 1%. Among the total anomalies 13.2% had congenital heart disease, with echocardiography done showed VSD (6.8%) and ASD (4.5%) being the two most common CHD. Other reported CHD include TOF (1%) and TGA (1%).

Limb anomalies were least in the present study, accounting for 8.7% of total anomalies. 3.4% having CTEV, absent left radius in 2.3%, polydactyly in 1% and syndactyly in 1% of study babies. Out of 91 babies 1.1% had congenital cystic adenomatous lung malformation and multicystic dysplastic kidney was

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present in 1.1% of the babies. In our study 2% of the study participants had multiple system anomalies.
In chest X ray findings most common reported anomaly was tracheooesophageal fistula (10.2%) followed by cardiomegaly (3.4%) and CDH (2.3%). In USG scan of abdomen 79% reported normal study, while 10% reported dilated bowel loops.

Table 1: Maternal risk factors associated with CA

Maternal risk factors (Babies with CA no=91)	Frequency	Percentage
Previous abortion	24	26.37
PreviousH/ocongenitalmal formation	14	15.38
Folicacidsupplementation	12	13.18
Polyhydramnios	6	6.5
Oligohydramnios	7	7.6
Pre-eclamptictoxaemia	5	5.4
Substanceabuse	3	3.2
Maternaldiabetes	2	2.2
Drug in take in the first trimester	2	2.2
Radiation exposure	0	0

Table 2: System wise distribution of CA

S No	System	CA (n = 91)	Percentage
1	GIT	40	43.93
2	CNS	31	34.1
3	Congenital heart disease	12	13.2
4	Limb anomalies	8	5.68
5	Urogenital anomalies	1	1.1
6	Lung malformation	1	1.1
7	Multiple malformation	2	2.2

Table 3: Maternal age association with CA

Maternal Age (groups)	Congenital anomalies		p-value	Odds ratio (95% CI)
	Yes	No		
< 35 years (n=248)	76 (30.6%)	172 (69.4%)	0.000145*	0.177 (0.066 – 0.473)
≥ 35 years (no=21)	15 (71.4%)	6 (28.6 %)		

*p-value < 0.05 – statistically significant

Table 4: Birth weight association with CA

Birth weight	Congenital anomalies		p-value	Odds ratio (95% CI)
	Yes	No		
< 2.5 kg (n=77)	39(50.65)	38(49.35%)	0.000222*	2.76 (1.59 – 4.78)
≥ 2.5 kg (n=192)	52(27.1%)	140(72.9%)		

*p-value < 0.05 – statistically significant

Table 5: Results of bi-variable analysis to find the associated maternal risk factors and congenital anomalies

Maternal risk factors	Congenital anomalies Yes (no=91)	No (no=178)	P value	Odds ratio (95% CI)
Previous abortion Yes (n=44) No (n=225)	24 (15.5%) 67 (29.7%)	20 (45.5%) 158 (70.2%)	0.0015*	2.83 (1.46 - 5.47)
Previous H/o congenital malformation Yes (no=17) No (no=252)	14 (82.3%) 77(30.8%)	3 (17.6%) 175(69.4%)	0.001*	10.61 (2.96 - 37.97)
Polyhydramnios Yes (no=36) No (no=233)	6(0.17%) 85(36.4%)	30(83.3%) 148(63.5%)	0.019*	0.35 (0.14 – 0.87)
Folic acid supplementation Yes (no=26) No (no=243)	12(46.2%) 79 (32.5%)	14(53.8%) 164(67.5%)	0.1622	1.78 (0.79 – 4.02)
Drug intake in the first trimester Yes (no=4) No (no=265)	2(50%) 89(33.5%)	2(50%) 176(66.5%)	0.491	1.98 (0.27 – 14.27)
Radiation Exposure Yes (no=1) No (no=268)	0(00%) 91(33.9%)	1(100%) 177(66%)	0.790	0
Maternal diabetes Yes (no=14) No (no=255)	2(14.2%) 89(34.9%)	12(85.7%) 166(65.5%)	0.112	0.31 (0.07 - 1.42)
Oligohydramnios Yes (no=29) No (no=240)	7 (24.1%) 84 (35%)	22 (75.9%) 156(60%)	0.304	0.59 (0.24 – 1.44)
Pre-eclampticToxaemia Yes (no=27) No (no=242)	5 (18.5%)	22(81.5%)	0.076	0.41 (0.15 – 1.13)

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	86 (35.5%)	156(64.5%)		
Substance abuse Yes(no=5) No (no=264)	3 (60%) 88 (33.3%)	2(40%) 176(66.6%)	0.212	3.00 (0.49 – 18.28)

*p-value < 0.05 – statistically significant

DISCUSSION

The incidence of CA in this study was 8.8/1000 live births. This is similar to studies conducted in which range was between 1.2 and 9.5%.6-11 The incidence of CA in India varies from 8.6 – 20.2 per 1000 births in different studies.2-3 The incidence of congenital malformations was 39.1/1000 births with a significantly higher incidence among the consanguineous group as against the non consanguineous group.12 A study carried out by the Indian council of Medical Research at various centres the incidence of major malformations was 18.9 in Chennai and in Hyderabad 14 per 1000 birth as comparable with the present study. The relative difference in the incidence of various malformations might be due to socio – economic, environmental and racial difference, in addition to the factors likely to be potentially teratogenic7.

In babies with CA, 71.4% of their mothers had age more than or equal to 35 years and 30.6% had age less than 35 years. In bi-variable analysis there was a significant association between age of mother and CA [p-value = 0.000149, odds ratio (95% CI) = 0.177 (0.066 – 0.473)]. In regard to maternal age and CA, a study conducted by Patel Z.M. et al the incidence of CA was higher in mothers > 35 years of age.8. A study done by Vikram Duta et al, does not found any correlation of congenital malformations in babies with maternal age.13

In this study babies with CA, most of them were term babies (70.3%). In a study by Gupta et al, it was found that 43.7% of mothers with congenital anomalous babies belong to gestational age > 36 weeks.14 Fatema et al identified majority of babies belonging to gestational age between 34 and 36 weeks.15

The most common mode of delivery in the current study was vaginal delivery (57%) and there was no significant association between mode of delivery and CA.

In the current study 59% of the mother had male child whereas 41% had a female child and there was no significant association between gender of baby.

Among the CA babies, 57.2% babies weighed more than 2500 grams and 42.8% weighed less than 2500 grams and there was a significant association between birth weight and CA (Chi – square = 13.635, p-value = 0.000222) .

In our study, GIT anomalies were most common accounting for majority (44%), followed by CNS anomalies (34.1%), and congenital heart disease (13.2%). Gupta et al reported most common congenital anomaly were of CNS (53.3%) followed by GIT (13.2%).14 Among the GIT anomalies, the three most reported anomalies were imperforate anus (9.9%), trachea-oesophageal fistula (9.9%) and cleft lip and cleft palate (6.6%). Our results were in par with similar studies conducted in India.15-16 Another two studies reported 11.76% and 20.83% of GIT anomalies respectively.16-17

Among the CNS malformations, the most common were myelomeningocele (15.3%) and hydrocephalus (14%). Gupta et al in their study also reported similar findings, but anencephaly was most prevalent in their study. Study done by Lavanaya et al reported 6.3/1000 incidence of neural tube defects (NTDs) and anencephaly (40%), spina bifida (46%), and encephalocele (6%) were the common ones.18 Verma et al found high incidence of neural tube defects in people of Punjab and Rajasthan.10 It is clear that prevalence of anencephaly and spina bifida is higher in Punjab, Rajasthan as compared to that in the southern and Eastern states of India. In fact incidence in these states is as high as Ireland, where the highest rate of neural tube defects had been recorded. The exact reason for such a difference in rate for neural tube defects in different parts of India is not clear; some of the likely genetic and environmental factors have been established. Among the total anomalies 13% had CHD in our study, with ECHO done showed VSD (6.8%) and ASD (4.5%) being the two most common CHD.

Limb anomalies were least reported in our study, accounting for 9% of total anomalies. 3.4% having CTEV and absent left radius in 2.3% of the babies. Gupta et al reported common musculoskeletal anomalies such as CTEV, achondroplasia, polydactyly, sacrococcygealteratoma, and skeletal dysplasia.14 Bhatt et al reported an incidence of musculoskeletal malformation in 9.69/1000 births as the commonest one in their study.6 Lucas et al reported 5.9% incidence of skeletal system anomalies.19

Urogenital anomalies accounted for only 1% in our study. But renal anomalies were a major contribution in other similar studies where ambiguous genitalia, hydronephrosis, hypospadias, cystic kidney disease, PUV were seen among the babies.14-15

Among multiple system anomalies, in our study 2% of the study participants were seen which is comparatively lower than other studies where as high as 10% - 37% were reported.8-9

In Chest x ray among the study participants, 82% reported normal study. Among abnormal findings, most common reported anomaly was TEF (10.2%) followed by cardiomegaly (3.4%) and Congenital Diaphragmatic Hernia (2.3%).

In USG scan 79% reported normal study, while 10% reported dilated bowel loops in our study. Other significant findings were duodenal atresia,

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congenital diaphragmatic hernia (CDH), and multi cystic dysplastic kidney. Gupta et al in their study reported polyhydramnios and oligohydramnios as a predominant finding in the mothers and when corelated with babies they were significantly associated with renal anomalies.14

CONCLUSION

The incidence of congenital malformations in the present study is 8.8 per 1000 births. The most common system involved in the study was GIT (43.95%) followed by CNS (34.1%). The incidence of malformations were higher in babies born to mothers over the age of 35 years or above. The most common maternal risk factor which were associated for CA were mothers having history of abortions in past (26.4%), followed by previous history of congenital baby (15.4%). CA diagnosis is not fruitful unless facility for correction, treatment and support to the patients and family at tertiary care level hospital is available.

Conflicts of Interest Nil

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Ethics approval Yes.

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