

## Original Research Article

# Spectrum of congenital malformations at birth among neonates in a private medical college in South Rajasthan

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**Received:** 18 May 2019

**Revised:** 30 June 2019

**Accepted:** 02 July 2019

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### ABSTRACT

**Background:** Congenital anomalies contribute to about 12% neonatal deaths annually. Neonates with multiple congenital malformations pose a very difficult management problem for the treating physician. This study was done to know the incidence, pattern of congenital anomalies and to study various maternal risk factors leading to congenital anomalies which may help us in devising strategies for better patient counseling and management.

**Methods:** Prospective cross sectional study carried out from 1st Jan 2014 to 31st December 2018 in a private medical college in India. Neonates (both live and still born) delivered in our hospital during this period formed the part of study group. All congenital anomalies present were documented and classified according to system involved.

**Results:** Total number of neonates with congenital anomalies were 90, out of which 73 were live births and 17 were still births. The overall incidence of congenital anomalies was 2.375%. The commonest system affected was musculoskeletal system (27.7%) followed by CNS (24.4%). Among the maternal risk factors studied, increased maternal age, consanguineous marriage, maternal gestational diabetes mellitus were all significant risk factors associated with congenital anomalies.

**Conclusion:** Congenital anomalies are a global health problem. In our study we have documented that multiparity, consanguinity, diabetes mellitus, Pregnancy induced Hypertension (PIH), maternal anemia, maternal malnutrition to be major contributing factors for congenital anomalies. Present study highlighted that musculoskeletal and CNS systems to be the most commonly affected by congenital malformations. Antenatal scans remain an important diagnostic tool in screening for congenital anomalies. A good clinical examination at birth could help in early detection of life threatening congenital malformation thereby improving chances of his or her survival.

**Keywords:** Congenital anomalies, Neonates

### INTRODUCTION

According to World Health Organization (WHO), congenital anomalies are defined as structural or functional anomalies including metabolic anomalies which are present at the time of birth.<sup>1,2</sup> An estimated 303,000 neonates die within 4 weeks of birth everywhere worldwide due to congenital anomalies. Congenital anomalies account for 8-15 % of perinatal deaths and 13-

16% of all neonatal deaths in our country.<sup>3</sup> As other causes of neonatal mortality like infections, nutritional deficiencies are being brought under control, congenital malformations are rapidly emerging as one of the most important causes of neonatal mortality.<sup>4</sup> Congenital anomalies can contribute to long term disability which may have significant impact on individuals, families and the society on the whole. Congenital anomalies are a group of diverse disorders of prenatal origin that can be

due to single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens, maternal infections, maternal illness etc. The most important maternal nutritional deficiencies seen in Indian population are that of Iron deficiency, Folic acid and Zinc deficiencies. Of which the later two have been associated with neural tube defects. Due to folic acid being prescribed regularly in antenatal checkup incidence of neural tube defects has decreased appreciably.<sup>2</sup> Congenital anomalies can be classified into major and minor.<sup>1</sup> Major defects are structural anomalies that have cosmetic or medical consequences which may require surgical intervention for correction. Minor anomalies are those with no medical or surgical significance but maybe useful in identifying specific syndromes of which they may be a part of. Both major and minor anomalies may present in various patterns such as a part of syndrome or as a part of developmental disruption, deformation. Neonates with congenital anomalies pose unique challenges to the treating physician as far as management is concerned.

This study was carried out to find the incidence and distribution of neonates with congenital anomalies.

**METHODS**

The study was a cross sectional study carried out during a period of 5 years from Jan 1, 2014- 31st Dec 2018 in the department of pediatrics in a private medical college in South Rajasthan. The hospital caters to both urban and rural population and is also a teaching hospital for undergraduate students. All live and still births which took place during this period in our hospital formed the study group. All neonates were examined for congenital malformations at birth and daily on rounds till discharge. Relevant information like maternal age, antenatal history, history of drugs taken by mother during pregnancy, maternal illness, history of consanguinity, exposure to any known teratogens, natal history, birth weight , sex, other relevant findings were recorded on a previously designed performa. Antenatal ultrasonography findings and anomaly scan findings were recorded. Relevant histological, hematological, radiological , genetic tests were carried out. USG was done as a routine to rule out any internal congenital malformations. 2 D echo was done in all cases where any congenital heart disease was suspected. All anomalies were classified according to system. The findings were noted on an excel worksheet and statistical analysis was carried out using SSPS software. Prior clearance from institutional ethics committee was sought before commencing the study.

**RESULTS**

During the study period there were a total of 3789 births out of which 3735 were live births and 54 were still births(Table 1).

**Table 1: Profile of Study Population.**

Details	Number	Percentage
Total Births	3789	
Live Births	3735	98.57%(3735/3789)
Still Births	54	1.43%(54/3789)
Neonates with Congenital malformations	90	2.37%(90/3789)
Live births with congenital malformations	73	1.95%(73/3735)
Still births with congenital malformations	17	31.48%(17/54)
Males with congenital malformation	36	40%
Females with congenital malformations	54	60%

**Table 2: Pattern of distribution of congenital anomalies according to system.**

System	Anomalies	Number	Percentage
CNS	Anencephaly	09	10%
	Meningocele	04	4.44%
	Encephalocele	01	1.11%
	Hydrocephalus	07	7.77%
	Corpus Callosum agenesis	01	1.11%
CVS	Congenital Heart Disease	03	3.33%
GIT	Cleft Lip and Palate	03	3.33%
	Tracheo oesophageal fistula	03	3.33%
	Gastrochisis	01	1.11%
	Omphalocele	01	1.11%
	Imperforate anus	04	4.44%
Respiratory	Diaphragmatic hernia	03	3.33%
Genito Urinary	Hypospadias	10	11.11%
	Posterior urethral valve	02	2.22%
	Polycystic kidney disease	01	1.11%
	B/L hydronephrosis	02	2.22%
Ear	Absent pinna	01	1.11%
	Pre auricular Sinus	06	6.66%
Eyes	Anophthalmia	01	1.11%
Musculo skeletal	CTEV	15	
	Sacrococcygeal teratoma	02	
	Polydactly	08	
Miscellaneous	Down Syndrome	02	

**Table 3: Maternal Risk factors in the study population.**

Maternal risk factors	No. of babies with anomalies	Percentage
Consanguinity	90	
Consanguineous	35 In 312 consanguineous parents	11.2%
Non Consanguineous	55 In 3477 non consanguineous parents	1.58%
Antenatal anomaly scan	Done only in mothers of 45 affected neonates	
Anomalies missed	13	28.8%
Anomalies detected	32	71.2%
Parity		
Primi	28	31.11%
Gravida 2	34	37.77%
Gravida 3 and above	28	31.11%
Antenatal problems in mothers		
GDM	20	22.22%
PIH	16	17.77%
Anemia complicating Pregnancy	06	6.66%
No antenatal problems	48	53.33%

**Table 4: Association between age of mother and congenital anomaly.**

Age of Mother in years (Range)	Neonates with congenital anomalies	Percentage
18-23	29	32.22%
24-29	21	23.33%
Above 30 years	40	44.44%
Total	90	100%

Df=1,  $\chi^2=0.6$ ;  $p < 0.05$  is significant

**Table 5: Association between Degree of Consanguinity and Congenital anomaly.**

Consanguinity	Neonates with Congenital anomalies	Percentage(%)
1 <sup>st</sup> Degree	27	77.14
2 <sup>nd</sup> Degree	04	11.4
3 <sup>rd</sup> Degree	03	8.57
Total	35	100

Df=1,  $\chi^2=0.5$ ;  $p < 0.05$ , significant

**Table 6: Distribution of cases according to fetal outcome (73 cases).**

Fetal outcome	No.	Percentage
Discharged	38	52.05%
Expired within 48 hrs of birth	20	27.39%
Operated	03	4.13%
Referred	12	16.43%
Total	73	100%

90 babies had congenital anomalies. Out of the 90 babies who had congenital anomalies 73 were live births and 17 were still births. The overall incidence of congenital anomalies was 2.375%. Incidence of congenital anomalies among live births was 1.955 whereas it was 31 % among still born babies. Still born babies had a significantly higher incidence of congenital malformation as compared to live born babies. Among the babies with congenital malformations there was a female preponderance with 54(60%) female babies with congenital anomalies as compared to 36 male babies(40%). 70 babies had one or more malformations. We had 10(11.1%) cases of multi system involvement. In our study the commonest system affected was the musculoskeletal system with 25 out of 90 babies(27.77%) having congenital malformation related to musculoskeletal system. CNS anomalies were the second commonest with 22 babies(24.44%) having CNS malformation (Table 2). Among the maternal risk factors studied, increased maternal age, consanguineous marriage, maternal gestational diabetes mellitus were all significant risk factors associated with congenital anomalies (Table 3). We found a higher incidence of congenital anomalies in mothers whose maternal age was more (Table 4). Consanguinity was also associated with higher incidence of congenital anomalies (Table 5). As majority of our patients are from rural background only in 50% of cases with congenital anomalies maternal antenatal scan was done, in 28.8% of cases anomalies were missed on antenatal anomaly scan. Out of the 90 babies with congenital anomalies 38(42.2%) were discharged from hospital as stable; 37(41%) expired, 3 were operated upon and 12 were referred to higher centres (Table-6).

**DISCUSSION**

With improvements in health care facilities in our nation we have been able to control infectious diseases and nutritional problems, which used to be a major cause of infant mortality till now. If the present trend of improvement in health care continues congenital malformations and death due to them would be one of the major causes of neonatal and infant mortality in our country similar to the prevailing trend in the western world.

The incidence of congenital anomalies in our study was 2.37% which is comparable to studies from other parts of

the country. Singh et.al reported an incidence of 1.5%, Basavanhappa reported an incidence of 3.083%.<sup>5,6</sup> Desai et.al from Bombay and Doddabasappa et.al from Bangalore have reported an incidence of 3.61% and 4 % respectively.<sup>7,8</sup> Vyas et.al from Kota and Gandhi et.al from Surat both have reported much lower incidence of congenital anomalies at birth i.e 1.23%.<sup>9,10</sup>

In our study the maximum number of congenital anomalies was from the musculo skeletal system followed by the central nervous system. Tenali et.al and Bhat et.al have also reported musculoskeletal congenital anomalies as the commonest.<sup>11,12</sup> Other studies have found anomalies of the Central nervous system to be the commonest followed by musculo skeletal system.<sup>13-18</sup> Central nervous system anomalies and anomalies of the musculoskeletal system appear to be the two most common systems affected by congenital malformation.

In our study CTEV was the commonest musculo skeletal anomaly followed by polydactyly. Neural tube defects contributed to 21% of total congenital anomalies, the high incidence of neural tube defects in our study could be attributed to lack of antenatal care and non-supplementation of folic acid as majority of ladies whose babies had these defects were from rural background. Uro genital anomalies were also not uncommon, hypospadias was the commonest uro genital anomaly and patients were managed well by the urologist. We had 2 cases of B/L hydronephrosis and 1 case of Posterior urethral valve ,all of them were diagnosed antenatally. The low incidence of CVS anomalies in our study can be explained by the fact that majority of CVS malformations do not manifest in 1st week of life, hence the low incidence in our study.

Association of maternal risk factors with congenital anomalies has been well established from studies across the world. In our study maternal diabetes was found to be a significant risk factor for occurrence of congenital anomalies. Other risk factors which were identified were maternal age, maternal anemia and maternal infections. Studies across the country have also found a similar co relation.<sup>11,13,14,17,19</sup>

Our study has also reinforced the fact that consanguinity is an important risk factor for occurrence of congenital malformations. The presence of congenital anomalies in neonates born to consanguineous parents was 11.2% as compared to 1.58% among neonates born to non consanguineous parents. Our study has also shown that statistically mothers above the age of 30 years are more likely to give birth to a congenitally malformed baby as compared to mothers below age of 30 years. This is in concurrence with other studies across India.

Out of the 73 live neonates with congenital anomalies 38 were discharged while 20 expired within 48 hours of birth. The highest death rate was found in neonates with multiple congenital anomalies. This could be explained

by the fact that multiple malformations prevent harmonious development of fetus leading to subsequent multi organ failure. This finding is similar to that reported by other authors like Charlotte et.al, Sarkar et.al, tenali et.al.<sup>11,20,21</sup>

The only drawback of our study is the sample size is small, inspite of it being a five year study. It can be explained by the fact that since it is a private medical college the number of deliveries are less.

## CONCLUSION

Congenital anomalies are an important cause of neonatal morbidity and mortality across the world. In our study we have documented that multiparity, consanguinity, diabetes mellitus, Pregnancy induced Hypertension(PIH), maternal anemia, maternal malnutrition to be major contributing factors for congenital anomalies. Present study highlighted that musculoskeletal and CNS systems to be the most commonly affected by congenital malformations.

Large multi centric studies are required to calculate the incidence of congenital malformations among neonates. Provision of good antenatal care, regular folic acid supplementation, anomaly scans, educating women of reproductive age group about pit falls of consanguineous marriage would help a great deal in reducing the incidence of congenital anomalies.

*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:** Bhatia R, Bhatia G. Spectrum of congenital malformations at birth among neonates in a private medical college in South Rajasthan. *Int J Contemp Pediatr* 2019;6:2053-7.