

## Original Research Article

# Burden and pattern of congenital anomalies in newborns in a tertiary care hospital

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### ABSTRACT

**Background:** Anomaly is defined as a significant morphological or anatomic variation in the phenotype from the standard reference population. Objectives of current study was to investigate the incidence and pattern of congenital anomalies in the hospital delivered neonates, JNMCH, AMU, Aligarh.

**Methods:** All the newborns delivered during the study period from October 2019 to November 2021 at JNMC, AMU, Aligarh were thoroughly examined and assessed for the presence of any congenital anomalies. Neonatal and maternal parameters were recorded in a predesigned proforma. Relevant investigations in the form of X-ray, ultrasonography and echocardiography were carried out in case of any suspected anomaly.

**Results:** Out of the total 10932 neonates delivered during the study period, 197 babies had congenital malformations including 166 live births and 31 still births with an incidence of 1.8%. Musculoskeletal system was the most commonly system involved and congenital talipes equinovarus was the single most common malformation. Higher incidence of malformation was seen to be associated with still birth, prematurity, male gender and low birth weight babies.

**Conclusions:** Regular antenatal checkups and anomaly scans can detect congenital malformations early. Pre conceptional counselling about maternal age at conception and birth order may help in preventing malformations associated with syndromes such as Down's and Edward syndrome. Peri conceptional folate supplementation can prevent CNS malformations such as Neural tube defects significantly. Early diagnosis and treatment of maternal disease conditions like anemia, preeclampsia, hypertension and diabetes may affect occurrence of malformations.

**Keywords:** Congenital malformations, Neonates, Musculoskeletal system

### INTRODUCTION

Congenital anomalies are defined as any morphological or anatomic variation in the phenotype from the standard reference population. WHO defines birth defects as a spectrum of structural, functional and metabolic defects which can occur either singly or as a group, present usually at birth and having a prenatal origin.<sup>1</sup> The worldwide incidence of congenital anomalies is estimated as 3-7%, but it may vary between countries.<sup>2,3</sup> According to the WHO, congenital abnormalities caused an estimated 270,000 deaths worldwide during the first 28 days of life

in 2010. In India, birth malformations account for 6-7 percent of all births, resulting in around 1.7 million birth defects each year.<sup>4</sup> Pathogenesis of major congenital anomalies lies in the interference with normal physiological processes such as intracellular signalling pathways, apoptosis and neural crest derivatives. First trimester (1-13 weeks period of gestation) is undoubtedly the most critical period for organogenesis. Any adverse medications, drugs, environmental toxins and infectious agents can significantly interfere with the process of normal foetal development and can result in congenital malformations.

### Gaps in knowledge

There has been a number of studies on the incidence of congenital anomalies in newborns in various parts of India but a study regarding the subject is lacking in this part of the country. In a developing country like India, where around 1.7 million birth defects are reported annually a detailed knowledge about congenital malformations, systemic distribution, risk factors and early diagnosis and management are needed to reduce the morbidity and mortality associated with it.<sup>4</sup>

### Need for this study

The study provided information on baseline characteristics, incidence, systemic distribution, prenatal and neonatal risk factors. This would aid in the development and improvement of the current health-care system, allowing for early diagnosis, prompt treatment and appropriate referral to higher-level centers.

## METHODS

### Study design, location and duration

Current study was a prospective cohort study conducted at neonatology section of department of pediatrics in collaboration with department of gynaecology and obstetrics, JNMC, AMU, Aligarh from October 2019 to November 2021.

### Study subjects

All newborns (n=10932) delivered during the study period were included in the study. Newborns were examined for the presence of any congenital anomalies soon after birth by the attending paediatrician, during the regular ward rounds and before discharge. Among live births with malformations outcomes in terms of survival and mortality was noted. Mortality was classified according to the time duration whether it occurred during the immediate (<24 hours) or early neonatal period (<7 days).

Neonatal variables like gender, anthropometry, gestational age and diagnosis at admission were noted. Birth weight >2.5 kg was classified as normal, 2.5-1.5 kg as low birth weight (LBW), 1-1.5 kg as very low birth weight (VLBW) and <1 kg was considered as extremely low birth weight (ELBW). Gestational age assessment was done by using the new Ballard score and newborns were classified as term and preterm. Those born at <37 completed weeks of gestation were considered as preterm, between 37 0/7 and 36 6/7 weeks of gestation as Term and after 42 weeks were considered as post term. Maternal parameters such as maternal age, consanguinity, gravida, maternal illness, drug intake, history of infertility treatment, previous abortions or still births and antenatal ultrasonography (USG) findings were recorded in a predesigned proforma. Relevant investigations in the form of X-ray, ultrasonography and echocardiography

were carried out in case of any suspected anomaly and to find out the extent of involvement. Malformations were classified according to system involved, whether single or multiple.

### Inclusion criteria and exclusion criteria

Inclusion criteria for current study were all neonates with birth defects delivered at JNMC were included in the study. Out born babies with malformation were excluded.

### Ethics committee approval

Clearance was sought from institutional ethics committee, faculty of medicine, AMU, Aligarh.

## RESULTS

During the study period there were total 10932 deliveries out of which there were 10318 live births and 614 still births. Out of total deliveries there were 5 triplets and 203 twin deliveries. Incidence of malformation in our study was found out to be 1.6% in live newborns, whereas 5.04% in still births with an overall incidence of 1.8% as shown in the (Table 1).

**Table 1: Distribution of study subjects according to live/still birth; out of total 10932 newborns, 197 were with anomalies with an incidence of 1.8%.**

Variables	Total patients	Cases with anomalies	Incidence (%)
Still birth	614	31	5.04
Live birth	10318	166	1.60
<b>Total</b>	<b>10932</b>	<b>197</b>	<b>1.80</b>

During the study, significantly higher incidence of malformations was noted in still birth as compared to the live newborns ( $X^2=38.75$ ;  $p=0.00$ ). The ratio of incidence of malformations in stillbirth to livebirth was 3.15. Similar findings were noted in preterm babies which were more commonly affected than term babies. Distribution of newborns according the birth weight showed highest incidence of malformation in low birth weight babies (LBW). Sex wise distribution showed significant male preponderance ( $X^2=4.46$ ;  $p=0.034$ ) with a male to female ratio of 1.64 among malformed neonates. Maternal factors showed higher incidence of malformation among multigravida (3%) than primigravida (0.98%) indicating higher chances of malformations with increasing birth order ( $X^2=66.7$ ). A total of 197 couples were found to have consanguineous marriage out of which 10.2% (n=20) had malformed babies. Association between congenital anomalies and perinatal risk factors is shown below in the (Table 2).

Musculoskeletal system was the most commonly affected system amongst the malformed babies (n=68; 34.5%) with congenital talipes equinovarus (CTEV) being the single most common (n=37; 18.7%) deformity.

**Table 2: Association between congenital anomalies and maternal and perinatal risk factors.**

Variables	Groups	Congenital anomaly		Total	X <sup>2</sup> value; df; p value
		Yes, N (%)	No, N (%)		
Live/still birth	Still birth	31 (5.04)	583 (94.9)	614	38.75;df=1; p=0
	Live birth	166 (1.6)	10152 (98.4)	10318	
Term	Preterm	77 (3.5)	2124 (96.5)	2201	47.12; df=1; p=0
	Term	120 (1.34)	8808 (98.6)	8928	
Birth weight (g)	<1000	5 (0.6)	773 (99.3)	778	15; df=3; p=0.0016
	1000-1499	12 (0.95)	1247 (99)	1259	
	1500-2499	86 (2.25)	3778 (97.7)	3864	
	>2500	94 (1.8)	5134 (98.2)	5228	
Sex	Male	120 (1.9)	5960 (98)	6080	4.46; df=1; p=0.034
	Female	73 (1.4)	4968 (98.5)	5041	
Gravida	Primigravida	68 (0.98)	6882 (99)	6950	66.72; df=1;p=0
	Multigravida	129 (3)	4050 (96.9)	4179	
Mode of delivery	Normal	128 (1.55)	8112 (98.5)	8240	8.5; df=1; p=0.003
	C-section	69 (2.4)	2820 (97.6)	2889	
Consanguinity	Yes	20 (10.2)	175 (89.7)	195	82.20; df=1; p=0
	No	177 (1.6)	10757 (98.3)	10934	

**Table 3: Systemic distribution of congenital malformations and outcome.**

System involved	Malformed babies (n=197)	Still birth (n=31)	Live birth (n=166)	Expired	Survived
				N (%)	N (%)
Musculoskeletal	68	4	64	9 (14)	55 (85.9)
GIT	47	11	36	14 (38.9)	22 (61.1)
CVS	39	6	33	12 (36.3)	21 (63.6)
CNS	37	10	27	11 (40.7)	16 (59.2)
GU	34	5	29	8 (27.6)	21 (72.4)
RESP	19	5	14	12 (85.7)	2 (14.28)

Next commonly involved systems were GIT followed by CVS, CNS, genitourinary, respiratory system in that order. Miscellaneous malformations encountered during the study included babies with white forelock, accessory nipple, congenital toxoplasmosis, sublingual cyst, pinna deformity and anophthalmia. There were 5 cases of Down's syndrome and 3 cases of Pierre Robin syndrome; while 2 cases of Edward and 1 case of Patau syndrome were noted. Multiple maternal risk factors were encountered in the study. Severe anaemia was seen in 6 mothers whereas a total of 148 mothers were anaemic (including mild, moderate and severe anaemia). Hypothyroidism was present in 13 mothers (6.6%) and were receiving treatment in the form oral tablets. There were 19 mothers (9.6%) with Gestational diabetes mellitus who were on treatment receiving insulin (9), diabetic diet (9) and oral hypoglycaemic agent (1). Also, there were 4 (2%) couples who were taking treatment for infertility.

Preeclampsia was present in 11% mothers (N=22). Liquor abnormalities were also noted in which 17.8% mothers (N=35) had polyhydramnios whereas 17.3% mothers had oligohydramnios (N=34) and rest of the mothers had no liquor abnormality. However there was no history of malformations in previous pregnancies.

There was history of abortion in 26% mothers while history of intrauterine death and sibling loss was present in 3% and 0.5% mothers respectively. Around 39% of the malformations were detected antenatally and rest were detected postnatally; immediately after birth or during routine ward rounds. Certain malformations were obscured at birth and were detected with the help of imaging techniques such as echocardiography and ultrasonography as in case of congenital heart diseases and malformations involving GIT system. Majority of the malformations that were antenatally diagnosed were cardiovascular (N=23; 29.9%) followed by GIT (N=22; 28.6%) and CNS (N=20; 26%). Least common anomaly to be detected was musculoskeletal system and genitourinary system (each N=11; 14.3%). Frequency and percentage of anomalies diagnosed antenatally according to the system involved is shown below in the (Table 4).

## DISCUSSION

Our study found out overall incidence of malformations to be 1.8% which is in concordance with the previous studies by Taskande et al, Prajapati et al, Arjun Singh et al etc. as shown in table XVI.<sup>2-6,10,11</sup> Higher incidence of malformation was noted among still born (5.04%) as compared to the live neonates (1.6%) and the results were

statistically significant ( $X^2=38.75$ ;  $p=0.000$ ). A possible reason is that many malformations are severe and lethal resulting in fetal demise. Similar results have been

demonstrated in studies by Prajapati et al, Taskande et al, Arjun Grover and Neelam Grover.<sup>3,10,11</sup>

**Table 4: System wise distribution of congenital malformations.**

System	N	%	System	N	%
<b>Musculoskeletal system</b>			<b>CNS</b>		
CTEV	37	18.7	Congenital hydrocephalus	10	5.07
Rocker bottom foot	6	3.04	Spinal bifida	9	4.5
Polydactyly	6	3.04	Arnold chiari2	6	3.04
Syndactyly	5	2.53	Anencephaly	6	3.04
Arthrogryposis multiplex congenita	2	1.01	Holoprosencephaly	3	1.52
Calcaneovalgus	2	1.01	Dandy walker	3	1.52
Epigastric hernia	1	0.5	Encephalocele	2	1.01
Limb hypoplasia	1	0.5	Corpus callosal agenesis	1	0.5
Microtia	1	0.5	<b>Genito urinary</b>		
Achondroplasia	1	0.5	PUJ obstruction	8	4.06
Cystic hygroma	1	0.5	Ambiguous genitalia	4	2.03
Skeletal dysplasia	1	0.5	Hypospadias	4	2.03
Ectodermal dysplasia	1	0.5	Undescended testis	4	2.03
TAR syndrome	1	0.5	Micropenis	3	1.52
Umbilical hernia	1	0.5	Renal agenesis	3	1.52
<b>GIT</b>			Renal pyelectasis	3	1.52
Cleft lip and palate	15	7.6	Vescicorectal fistula	3	1.52
Anorectal malformation	9	4.5	Antenatal hydronephrosis	2	1.01
Tracheoesophageal fistula	5	2.53	Multi-cystic dysplastic kidney	1	0.5
Cleft palate	3	1.5	<b>Respiratory system</b>		
Congenital ascites	3	1.5	Lung hypoplasia	9	4.5
Gastroschisis	3	1.5	Congenital diaphragmatic hernia	5	2.5
Intestinal obstruction	2	1.01	Laryngomalacia	2	1
Omphalocele	2	1.01			
Duodenal atresia	2	1.01			
Ileal Atresia	2	1.01			
Hirschsprung disease	1	0.5			
<b>CVS</b>					
A cyanotic CHD	30	15.2			
Cyanotic CHD	4	2.03			
Dilated heart chamber	2	1.01			
Single umbilical artery	3	1.52			

Highest number of malformations involved musculoskeletal system and CTEV was the single most common anomaly encountered which was also seen in studies by Prajapati et al and Mathur et al whereas Suguna et al reported GIT as the most common malformation in their studies.<sup>3,12-16</sup> In our study we noticed GI malformations to be second highest after musculoskeletal system. Amongst GI anomalies, Cleft lip and palate was the most common anomaly ( $n=15$ ).<sup>17,18</sup> Other studies such as Taskande et al indicated CVS as the most the common system involved. Annual report of Indian council of medical research also mentions cardiovascular as the most common system involved (0.57%).<sup>19</sup> In these study, antenatal echocardiography was carried out if mothers had risk factors for fetal malformations whereas in our study echo was done

postnatally, only if babies had clinical signs and symptoms of congenital heart disease or if the diagnosis was made antenatally during routine antenatal checkups. Another possible explanation could be late diagnosis of congenital heart diseases after discharge from the hospital or during the decompensation states such as congestive cardiac failure, which usually presents late as in cases of A-cyanotic heart disease such as ventricular septal defect. During our study we encountered 30 cases of A-cyanotic heart disease maximum of which were ventricular septal defect ( $n=18$ ) followed by cyanotic heart disease ( $n=4$ ). Other defects included dilated heart chambers ( $n=2$ ) and single umbilical artery ( $n=3$ ). Highest incidence of CNS malformations was seen in study by Kalra et al.<sup>18</sup> In our study congenital hydrocephalus was the single most common ( $n=10$ ; 5.07%) CNS anomaly seen. Other

malformations noted were spinal bifida, Arnold Chiari II malformation, anencephaly, Holoprosencephaly, Dandy walker malformation and neural tube defects. With regular antenatal checkups and folate supplementation in pregnancy, incidence of neural tube defects has been declining in developing countries like India.<sup>20</sup>

**Table 5: Antenatal diagnosis of malformation.**

System involved	Malformed babies	Antenatally Diagnosed
Musculoskeletal	68	11 (14.3)
CNS	37	20 (26)
GIT	47	22 (28.6)
Genitourinary	34	11 (14.3)
CVS	39	23 (29.9)
Respiratory	19	14 (18.2)

**Table 6: Comparison of incidence of malformation in previous studies.**

Reference	Overall incidence	Incidence in live birth	Incidence in still birth
Present study	1.8	1.6	5.04
Taskande et al	1.9	1.84	4.68
Prajapati et al	1.69	1.52	6.27
Singh et al	1.5	1.39	4.46
Grover	1.78	1.3	15.1
Chaturvedi et al	2.72	-	-
Dutta et al	1.24	-	-

### Limitations

Antenatal USG was not done in some mothers as many of them were not registered antenatally. Specific investigations such as echocardiography, imaging and ultrasonography were done only in babies with clinical signs and symptoms so subclinical malformations might have remained undetected. The study being a hospital based prospective cohort study may have overestimated the incidence of malformations. Community based studies might depict actual burden of congenital anomalies. The study did not include malformations which were detected and terminated before 20 weeks. Periodic follow up might have detected some of the anomalies that were not obvious at the time of birth or during hospital stay but may have manifested later in life. The study was conducted during the period of COVID-19 pandemic which resulted in slightly less deliveries than usual due to frequent lockdowns.

### CONCLUSION

Incidence of malformations in our study was found out to be 1.8% overall and 1.6% and 5.04% in live and still birth

respectively. System wise distribution indicated musculoskeletal to be the most common system involved followed by gastrointestinal, cardiovascular, CNS, genitourinary and respiratory system in that order. Among neonatal risk factors low birth weight, Prematurity and male sex showed statistical significance. Maternal risk factors including birth order, consanguinity, mode of delivery and poor obstetric history were found to be statistically significant. Poor neonatal outcomes were seen in babies with respiratory, CNS and GIT system malformations.

### Recommendations

Regular antenatal checkups and anomaly scans can detect congenital malformations early. In our study only 40% of the malformations were antenatally detected whereas around 60% of the malformations were noticed postnatally, immediately after birth or during hospital stay. Robust antenatal screening programs and better adherence to ANC visits may aide in early detection which can help couples to take decision regarding continuation or termination of pregnancy. Pre conceptional counselling about maternal age at conception and birth order may help in preventing malformations associated with syndromes such as Down's and Edward syndrome. Peri conceptional folate supplementation can prevent CNS malformations such as Neural tube defects significantly. Early diagnosis and treatment of maternal disease conditions like anemia, preeclampsia, hypertension and diabetes may affect occurrence of malformations.

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