International Journal of Contemporary Pediatrics
Nikhurpa M et al. Int J Contemp Pediatr. 2020 Dec;7(12):2289-2294
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20204959

Original Research Article

Study of clinical profile and associated risk factors for congenital malformations in neonates: a hospital based prospective study

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Received: 25 September 2020
Revised: 22 October 2020
Accepted: 26 October 2020

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ABSTRACT

Background: Congenital malformations are major cause of neonatal morbidity and mortality and account for 8-10% of all peri-natal deaths especially in developing countries. Incidence of congenital malformation is increasing owing to reduction in other causes of neonatal deaths due to improvement in perinatal care worldwide. This study was aimed to evaluate the incidence, perinatal variables and contributing risk factors associated with birth defects that will help to plan future strategies for prevention, early diagnosis and timely management.
Methods: This is a hospital-based prospective observational study conducted in department of pediatrics of a tertiary care centre of western India over period of two year. All new-borns with anatomical congenital malformations detected antenatal or postnatal (prior to discharge) period were included in this study. Detailed and careful clinical examination was carried out for all new-born.
Results: The incidence of congenital malformation in the present study is 1.27%. Prematurity (76.1%), low birth weight (68%) and male neonates (67%) were associated with increased risk of congenital malformation. Most common system involved in this study was cranio-spinal system (47.3%). Overall, most common congenital anomaly was Hydrocephalus with meningomyelocele (8%). Among maternal variables, malnutrition (90%), consanguinity (40%) and abortions (40%) are strongly associated with malformations.
Conclusions: Congenital malformations are important cause of neonatal death. Strategies to diagnose, prevent, treat and rehabilitate the neonate are utmost necessary. Early screening to identify high risk pregnancies and timely management is strongly recommended.

Keywords: Congenital malformations, Neonatal mortality, Maternal risk factors

INTRODUCTION

Congenital malformation, also known as birth defects, includes structural or functional anomalies of prenatal origin, resulting from an abnormality or defect that occurs during the development process.[1] According to WHO, the term "congenital malformations" should be confined to structural defects present at birth. Congenital abnormalities are the most important cause of neonatal death in countries with low and very low U5MR.[2] Global estimates suggest that congenital anomalies affect 2-3% of births.[3] In 2015 worldwide out of 5.9 million, 2.7 million under- 5 deaths occurred in the neonatal period. In developing countries birth defects cause 5-7% of perinatal, neonatal and childhood mortality. India contributes to one-fifth of global live births. The current neonatal mortality rate is 28 per 1000 live births. With a decrease in infectious causes of neonatal deaths, the proportion of mortality due to congenital anomalies is likely to increase especially in urban areas in India.[4] Congenital malformations account for 8-10% of all peri-natal deaths and 13-16% of all neonatal deaths. In 2010,

the world health assembly (WHA) recognized the importance of birth defects as a cause of stillbirths and neonatal mortality, and that the attainment of millennium development goals 4 on reduction of child mortality will require accelerated progress in reducing neonatal mortality, including prevention and management of birth defects. As per march of dimes (MOD) estimates, every year 6% of children worldwide are born with a serious birth defect/congenital disorder due to genetic or environmental causes. Based on the annual births data of 2010 (163 million, as per the world health statistics, 2012 report) the estimate would be 9.78 million children. Globally, the most common serious birth defects of genetic or partially genetic origin are (Christianson, Howson and Modell, 2006): congenital heart defects (1040835 births), neural tube defects (323904 births), thalassemia and sickle cell disease (307897), down syndrome- trisomy 21 (217293 births), G6PD deficiency (177032 births). Combined, these five conditions account for about 25% of all birth defects the true magnitude of the number of births affected by congenital anomalies in India is unknown due to lack of national birth defects surveillance.[5] This study was conducted to evaluate the incidence of structural congenital anomalies and to predict the variables which contribute incidence as well as prevention of births defect. It also targets contributing risk factors for adverse pregnancy outcomes as some of these conditions can be prevented through primary care interventions targeted towards women in the preconception, intra-conception and antenatal periods.[6]

METHODS

This is a hospital-based prospective observational study conducted in Department of Pediatrics of tertiary care centre of western India over period of two years from 1st September 2016 to 31st August 2018. All newborn with anatomical congenital malformation detected antenatal or postnatal prior to discharge (i.e. less than or equal to 7 days) delivered during study period were included and newborn delivered outside hospital, still births and abortions were excluded from study. Detailed and careful clinical examination including congenital malformation was carried out of all newborns soon after birth. Patients were referred for super-specialty interventions whenever required and followed up for immediate outcome. Relevant information regarding maternal age, previous history of neonatal deaths, abortions, siblings with anomaly, smoking & alcohol habits in mother, gestational age, sex, community, birth weight, birth order and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labour, antenatal ultrasonography (USG) findings were noted. Relevant radiological and genetic tests were carried out. karyotyping and angiography were done as per requirement of malformation. Ultrasound was employed routinely to detect multiple congenital anomalies and to rule out majority of the internal congenital anomalies whenever indicated. 2D echocardiography was also used for all congenital heart diseases, along with the routine X-ray chest and electrocardiogram. Other investigations were MCU and DMSA. Computed tomography (CT) scan and magnetic resonance imaging (MRI) were advised only for certain special cases (when USG findings show significant abnormality). Malformations were divided into major and minor; major malformation interferes considerably with the function of all or part of the infant, minor malformation gives no serious medical or cosmetic consequences to the patients. The major malformations were divided into central nervous system (CNS), musculoskeletal, gastrointestinal, genitourinary, cardiovascular system (CVS), syndromes and miscellaneous disorders. Simple random sampling was done to include 150 newborns in study. Statistical analysis was done using chi-square test and p value. Data analysis was done using statistical software SPSS version 11.5 and descriptive and analytic (one-way analysis of variance (ANOVA), t-test) statistics and confidence interval 95%, p value<0.05 was considered significant.

RESULTS

The present study was a prospective observational study carried out on 150 newborns admitted to NICU of a tertiary care hospital, over period of two years. During the study period, 11792 newborns were delivered and after applying the exclusion criteria 150 patients were included in our study resulting incidence of 1.27% in our centre.

Out of 150 babies, 92 babies (61%) were male and 58 babies (39%) were female. In present study number of preterm newborns were 114 (76.1%) followed by term 32 (21.3%) and post-term 4 (2.6%). As per birth weight distribution 68% (102) were low birth weight (less than 2.5 kg) followed by 19% (29) normal weight (2.5-4 kg) and 13% (19) with birth weight more than 4 kg, this high incidence of low birth weight is probably due to congenital malformation predisposing to preterm birth which ultimately leads to low birth weight.

Among maternal risk factors, out of 150, 90% (135) mothers were having malnutrition, while 40% (60) mothers were having history of consanguinity, 40% (60) mothers were having history of abortion. Higher incidence 23% (35) of anomaly was found in third degree consanguinity in this study. Maternal diabetes was present in 10% (15) of cases. Infections, fever was present in 10% (15) of cases. History of maternal drugs intake was found in association with, valproate, leveteracetam, and eptoin in 16% (25) cases. History of polyhydramnios and oligohydramnios was present in 12% (18) and 11 % (16) respectively.

Among antenatal detection of anomalies, in 42 patients (28% of total cases) anomaly was detected antenatal by radiography. 1) In 1 patient, cardiac rhabdomyoma was diagnosed by fetal MRI 2) In craniospinal system 8 hydrocephalus (communicating and non-communicating),

3 meningomyelocele, 3 meningocele, 5 sacral agenesis, 2 encephalocele and 2 anencephaly were diagnosed antenatal by ultrasonography 3) In cardiovascular system 1 ebstein anomaly, 2 TAPVCs, 1 TGA were diagnosed in antenatal period by ultrasonography 4) In gastrointestinal system 3 omphalocele and 3 gastroschisis were diagnosed by antenatal USG. In renal system 5 renal agenesis were diagnosed by antenatal USG 5) In respiratory system 1 congenital diaphragmatic hernia and 1 congenital cystic adenomatous lung diagnosed by antenatal USG 6) 1 hydrops fetalis was diagnosed by antenatal USG.

Table 1: Distribution of anomalies (n=150).

Anomaly	N	Percentage (%)
Major (including lethal anomalies and syndromes	138	92%
Minor	12	8%

Most common craniospinal congenital anomaly was hydrocephalus with meningomyelocele (8%), followed by meningomyelocele (5.3%), sacrococcygeal teratoma (5.3%), encephalocele (5.3%). Higher incidence of craniospinal anomaly was because of higher referral rate to our tertiary care centre due to availability of super-speciality service. Most common cardiovascular congenital anomaly was VSD (3.3%) and ASD (3.3%). Most common gastrointestinal congenital anomaly was cleft lip & palate (5.3%), followed by omphalocele (4%) and tracheoesophageal fistula (3%). Renal anomalies were unilateral hydronephrosis (3.3%) and renal agenesis (3.3%). Anomalies of respiratory system were congenital diaphragmatic hernia (0.5%) and cystic adenomatous lung (0.5%). Most common lethal anomaly was encephalocele (5.3%) followed by gastroschisis (3.4%) and anencephaly (2.6%). Syndromes like down's syndromes (2%) and turner's syndrome (1.5%) were identified (Table 2).

Table 2: Major anomaly.

Major anomaly	N	Percentage (%)
Craniospinal system		
Anomaly	**(n=59)**	**(n=39.3)**
Communicating hydrocephalus with MMC	8	8
Non-communicating hydrocephalus with MMC	4	
Communicating hydrocephalus	4	4
Non-communicating hydrocephalus	2	
Meningomyelocele	8	5.3
Meningocele	6	4
Spina bifida occulta	5	3.3
Sacral agenesis	5	3.3
Holoprocencephaly	1	0.7
Dolicocephaly	8	5.3
Sacrococcygeal teratoma	8	5.3
Cardiovascular system		
Anomalies	**(n=25)**	**(n=17)**
Ventricular septal defect	5	3.3
Atrial septal defect	5	3.3
Double outlet right ventricle	2	1.3
Coarctation of aorta	2	1.3
Ebstein anomaly	2	1.3
Total anomalous venous connections	2	1.3
Transposition of the great arteries	2	1.3
Cardiac rhabdomyoma	2	1.3
Tetralogy of fallot	1	0.6
Other complex heart disease	2	1.3
Gastrointestinal tract system		
Anomalies	**(n=18)**	**(n=12.3)**
Omphalocele	6	4
Tracheoesophageal fistula	4	3
Cleft lip and palate	8	5.3
Renal system		
Anomalies	**(n=10)**	**(n=6.6)**
Unilateral hydronephrosis	5	3.3
Renal agenesis	5	3.3

Continued.

Major anomaly	N	Percentage (%)
Respiratory system		
Anomalies	**(n=2)**	**(n=1)**
Congenital diaphragmatic hernia	1	0.5
Cystic adenomatous lung	1	0.5
Lethal anomalies		
Anomalies	**(n=19)**	**(n=12.6)**
Encephalocele (craniospinal)	8	5.3
Anencephaly (craniospinal)	4	2.6
Gastroschisis (GIT)	5	3.4
Hydrops fetalis	2	1.3
Syndromes	**(n=5)**	**(n=3.3)**
Down's	3	2
Turner's	2	1.3

Table 3: Minor anomalies (n=12).

Anomalies	N	Percentage (7.9%)
Limb defects (musculoskeletal)	4	2.6
Polydactyly (musculoskeletal)	2	1.3
Clubfoot (musculoskeletal)	6	4

Clubfoot (4%), limb defects (2.6%) and polydactyly (1.3%) were minor anomalies identified (Table 3).

Among neonatal outcome overall 72% were discharged and 28% were expired (Table 4). Patients who were not operated and expired were because of poor general condition and poor prognosis. Encephalocele (5.3%) and anencephaly (2.6%) were two most common anomalies contributing to mortality.

Table 4: Neonatal outcome.

Outcome	N	Percentage (%)
Operated and discharged	49	33
Operated and expired	24	16
Not operated and discharged/LAMA	58	39
Not operated and expired	19	12

Table 5: Association of perinatal variable with congenital anomalies.

Variable	Group	Congenital abnormality				Total no.	$\chi 2$ p value
		Yes		No			
		No	%	No	%		
Maternal age	<20 years	42	1.1	3978	98.9	4020	2.992, p =0.23
	20-30 years	93	1.4	6598	98.6	6691	
	>30 years	15	1.4	1066	98.6	1081	
Parity	Primi	108	1.3	7972	98.7	8080	0.851, p =0.35
	Multi	42	1.2	3670	98.8	3712	
Consanguinity	Present	60	37	102	63	162	16951, p=0.00001*
	Absent	90	7.6	11690	92.4	11780	
History of abortion	Present	60	1.7	3360	98.3	3420	8.91, p=0.002*
	Absent	90	1	8282	99	8372	
Nutrition	Good	15	0.3	4702	99.7	4717	57.11, p=0.00001*
	Poor	135	1.9	6940	98.1	7075	

DISCUSSION

In India incidence of congenital malformation varies from 1.94% to 2.03% of birth in the present study incidence of congenital malformation in our centre was 1.27% as compare to Sarkar et al (2.22%) Taksande (1.9%) Singh (1.5%) Malla (0.36%).[7-11]

In the present study 76.1% of malformed babies were preterm and 21.3% babies were full-term and 2.6% were

post term which is comparable with Sarkar et al study in which 64% cases were pre term, 32% cases were term and 4% were post term.

In the present study 68 % of malformed babies had birth weight <2.5 kg that was similar with Sarkar et al study (77% of malformed babies). This difference is also highly statistically significant (p=0.00001). This is probably due to congenital anomaly predispose to preterm birth which ultimately leads to low birth weight.

In this study, male babies (67%) were more affected with malformations than female babies (39%). Study by Taksande and Dutta et al showing the similar results (61% male babies and 37.4% female babies, 64.7% male babies and 34% female babies respectively). Male preponderance may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life and also by fact that Autosomal recessive disorders are more common in males.[9,12]

Among maternal variables in present study 62% mothers were aged between 20 to 30 years, 28% were less than 20 years and 10% were more than 30 years, as compare to Sarkar et al. where 61 % mothers were aged between 20 to 30 years, 29 % mothers were less than 20 years and 10% were more than 30 years. Bai et al reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta et al documented statistically insignificant association of increased maternal age and congenital anomalies and Taksande reported higher incidence of malformations among the multiparas (19.5/1000 live births).[8,12,13] In the present study incidence was 22.59/1000 live births. Our result is consistent with this finding indicates a positive correlation between the birth order and the incidence of congenital anomalies in present study malnutrition (90%), consanguinity (40%) and abortions (40%) were three most important factors which were found to increase the risk of congenital anomalies comparable with Sarkar et al study which was 66%, 70%, 48% respectively.[8,9]

Most common system involved in this study was of craniospinal system (47.3%), followed by cardiovascular system (17%) and Gastrointestinal system (12.3%). This was comparable with study by Bai et al and Malla B where central nervous system malformation involvement 44% and 40% respectively.[12,13]

Cardiovascular system malformations were predominantly seen in study by Taksande (23%) Gastrointestinal system malformations are predominantly seen in study by Desai et al.[14] Differences between studies might be the effect of different racial, ethnic, and social factors in various parts of the world.

Overall, most common congenital anomaly was Hydrocephalus with meningomyelocele (8%), followed by meningomyelocele (5.3%), sacrococcygeal teratoma (5.3%), encephalocele (5.3%), cleft lip & palate (5.3%). Majority of babies with malformations discharged (72%) only 28% of babies expired and 39% of babies left against medical advice (LAMA). Encephalocele (5.3%) and anencephaly (2.6%) were two most common anomalies contributing to mortality.

Limitations

As it is a tertiary care hospital and referral institute, prevalence calculated may be higher than the general population. Hence, the data cannot be projected to the general population, for which population-based studies are necessary. Secondly, we could not include the abortions and stillborn, because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted and in most of the cases, parental consent is not available for pathological autopsy.

CONCLUSION

This study was aimed to evaluate the incidence, perinatal variables and contributing risk factors associated with birth defects that will help to plan future strategies for prevention, early diagnosis and timely management. As congenital malformation is becoming important cause of infant and childhood deaths, strategies to diagnose, prevent, treat and rehabilitate birth defects is necessary. Health education, nutrition, counselling about consanguinity before and after marriage, identifying high risk pregnancies like women >35 years of age, family history of birth defects, medical conditions, drug history need to be evaluated. Antenatal diagnosis is possible with maternal biochemical screening and ultrasonography. Extensive screening studies to determine the birth prevalence, types and distribution of congenital malformation are needful. Targeted and focussed approach should be stepped up to decrease incidence of congenital malformation.

ACKNOWLEDGEMENTS

Authors thank Dr Sumit Dabhi, Pediatrician, Baby-care children hospital, Junagadh (Gujarat) for his valuable support in data collection, compilation and clinical study to source.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Nikhurpa M, Munshi S, Shah D. Study of clinical profile and associated risk factors for congenital malformations in neonates: a hospital based prospective study. Int J Contemp Pediatr 2020;7:2289-94.