Annals of Neonatology

OPEN ACCESS
ISSN: 2636-3596

Original Article

Comprehensive Study of Congenital Malformations in Newborn Babies in A Tertiary Indian Care Hospital: A Prospective Observational Study

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DOI: 10.21608/ANJ.2024.314508.1101
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Abstract

Background: Congenital malformations are any irreversible condition existing in a child before birth, can be structural or functional anomalies and can be identified before birth, at birth or sometimes in later childhood. **Aim:** This study was carried out with aim to determine the incidence, pattern, various risk factors and outcome of congenital anomalies in neonates born in a tertiary care hospital and also emphasis the need of early detection and intervention, for decreasing associated morbidity and mortality. Early detection may give a chance to decide upon continuation of pregnancy and encourage preventive measures for prevention of its recurrence in subsequent pregnancy. **Methods:** This was an observational prospective study carried out in a tertiary care hospital in western Maharashtra, to study about the pattern of congenital malformation in this region. Data was analyzed using SPSS version 20. Results were presented in the tabular form and numbers of variables presented with help of frequency and percentages. **Results:** During the study period, 17000 babies were born, of which 300 had congenital malformations, making the incidence 17.6 per 1000 births of which 56% were male and 75% were less than 2.5kg. 46.7% babies were born to mothers >30yrs age. Out of the 300 cases, 20% died. The predominant system involved was central nervous system (31%). **Conclusion:** In India, prevalence of congenital anomalies remains high, third most common cause of perinatal mortality, hence emphasis should be given to early detection and intervention. Also increased awareness about preventable risk factors may help in reducing the burden of congenital anomalies on perinatal morbidity and mortality and improve the outcome. Preventive and interventional strategies will reduce the incidence of birth defects, decrease burden and also improve quality of living.

Key words: Congenital malformations, detection, risk factors , preventive measures

Introduction

Congenital malformations are any irreversible condition existing in a child before birth, can be structural or functional anomalies and can be identified before birth, at birth or sometimes in later childhood. Congenital malformations are multifactorial in origin, i.e. they are caused by the interaction of both genetic and environmental factors. The congenital anomalies account for 10%–15% of perinatal deaths and 13%–16% of neonatal deaths in India.[1] Preventive and interventional strategies will reduce the incidence of birth defects. Early detection and treatment with adequate health care facilities can be the done. The prevalence rate of congenital anomalies is increasing due to exposure of teratogens of various kinds and due to improving level of perinatal and neonatal care, hence India has reported congenital anomalies as, the third commonest cause of perinatal mortality.

Aim of the study: The objective of this study to assess the incidence, pattern, risk factors, outcome of congenital malformation and their correlation with various maternal risk factors.

Patients and Methods

This prospective observational study was conducted at a tertiary care center from January 2018 to January 2021. Approval from institutional ethical committee was obtained. Relevant history including familial and gestational factors were recorded in a proforma, these babies were examined and sex, birth weight and type of anomaly was recorded. System wise classification of anomalies were performed. Significant antenatal history like maternal illness, medication and antenatal ultrasonography findings were noted. A meticulous general and systemic examination was carried out by a consultant at the time of birth to detect any malformations. Ultrasound was employed routinely to detect multiple congenital anomalies and to rule out majority of the internal congenital

anomalies. 2D echocardiography was also used for all congenital heart diseases, along with the routine X-ray chest and electrocardiogram.

Ethical Considerations

The study was approved beforehand by the Research Ethics Committee of Dr. Vaishampayan Memorial Medical College and Hospital, Solapur and informed consent was provided by each participant's caregiver.

Data management and analysis

Statistical analysis was done using Z test and Chi-square test. Data was analyzed using SPSS version 20. Results were presented in the tabular form and numbers of variables presented with help of frequency and percentages.

Results

The incidence of congenital malformations was found to be 17.60 per 1000 live births, of which 56.3% male and 43.7% female newborns, 10.7% had a history of consanguineous marriage. Majority that is 56 .66% were low birth weight, followed by normal weight (27%), very low birth weight (11%) and extremely low birth weight (4%). 62.33% of newborn with congenital malformations were having gestational age of 37-42 weeks (full term) followed by 24.33% newborn with gestational age of 32-36 weeks (preterm) and 13.33% newborn were having gestational age of less than 32 weeks (very preterm). Proportion of full-term newborn were significantly higher than preterm newborn with congenital malformations. 46.66% mothers of newborn with congenital malformation were having age >30 years. Out of 300 newborns with congenital malformations 20% newborn died and 56.66% newborn were discharged, and 23.33% newborn were referred to higher centers.

We observed that 51% newborns had positive finding on ultrasonography during antenatal period. 30% newborns mothers had anemia and 13.7% newborns mother had history taking medications during antenatal period. 13% newborn, their mother had history of

abortion. It was observed that only 9.7% newborns mother had history eclampsia and 4% newborns mother had history of diabetes.

In this study out of total 300 newborn with congenital malformations 31% had CNS malformations, followed by 24% had skeletal, 14% had gastrointestinal, 13% had genitourinary, 10% cardiovascular, 3.33% & 2.33% each had facial and hematology malformations, respectively. In this study, only one (0.34%) newborn with cutaneous malformation was present and one (0.34%) newborn with ophthalmic malformation-Anophthalmia.

Out of these 300 newborns, 5 (1.66%) were having malformations with syndromic presentations. Two newborn each (0.66%) were having Down syndromes and Pierre Robin Syndrome. There was one case of Edward syndrome.

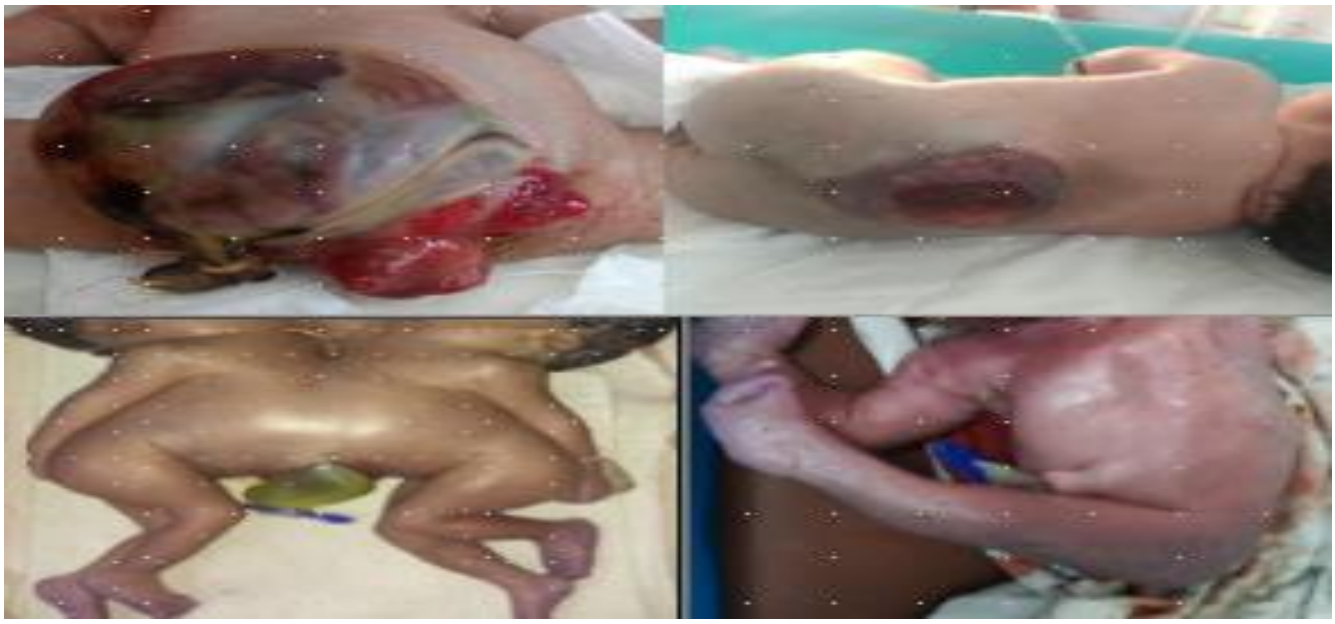

Figure1: Various congenital malformations.

Table 1: Demographic distribution and various presentation of congenital anomalies

Variables		**N =300**	**%(Percentage)**
Gender	Female	131	43.7
	Male	169	56.3
Birth weight(kg)	<2.5	215	71.6
	>2.5	85	28.3
Gestational age in weeks	≤32	40	13.3
	32-36	73	24.3
	37-42	187	62.3
Maternal age in years	<20	50	16.6
	21-30	110	36.6
	>30	140	46.66
Consanguinity	No	268	89.3
	Yes	32	10.7
Outcome	Death	60	20.00
	Discharge	170	56.66
	Referred	70	23.33
System involved	Cutaneous	1	0.34
	Ophthalmic	1	0.34
	Skeletal	72	24
	Genitourinary	39	13
	Central nervous	93	31
	Gastrointestinal	42	14
	Cardiovascular	30	10
	Syndromes	5	1.66
	Facial	10	3.33
	Hematology	7	2.33
Syndromes associated	Down Syndrome	2	0.66
	Edward Syndrome	1	0.33
	Pierre Robin Syndrome	2	0.66

Table 2: Correlation of various maternal risk factors with congenital anomalies

Variables		**N= 300**	**%**
Anemia	No	210	70
	Yes	90	30
Previous history of abortion	No	261	87.0
	Yes	39	13.0
History of medication	No	259	86.3
	Yes	41	13.7
History of eclampsia	No	271	90.3
	Yes	29	9.7
Presence of diabetes	No	288	96.00
	Yes	12	04.00
USG finding	Absent	147	49.0
	Present	153	51.0

Discussion

The incidence of congenital malformations was 17.60 per 1000 live births. Studies conducted by Ruth Wagathu et al [2] & Paramesh Pandalaet al [3] reported incidence 19.4 & 21.54 per 1000 live birth respectively. Proportion of male newborn with congenital malformations were significantly higher than female newborn similar was seen in Paramesh Pandala et al [3] study which reported 52.3% were males.

Proportion of newborn with low birth weight was significantly higher which was similar to Paramesh Pandala et al [3] i.e 61.6% were low birth weight whereas Studies conducted by Akinlabi E. Ajao et al 2019 [4] showed proportion of new born with normal birth weight were more as compared to low birth weight. Paramesh Pandala et al 2019 [3], Jayasree S. et al 2018 [5] reported, proportion of full-term births more compared to preterm births, similar to our study. Gholamreza Faalet al 2018 [6]

& Shantisena Mishra et al 2018 [7] reported similar finding where mothers with age greater than 30 years were 45.45% & 35.9% respectively. Percentage of consanguinity in the study conducted by Jayasree S. et al 2018 [5] was 12.4, Gholamreza Faal et al 2018[6] was 55.5%. Only one study, conducted by Shantisena Mishra et al 2018 [7] in which percentage of USG positive finding was 15.38% and this was contradictory to our study finding. Although routine screening for fetal abnormalities is very successful, there are limitations to the abilities of both the technique and the operators to detect every malformation. There are several reasons for this, not all malformations are evident at 20 weeks, when the routine ultrasound examination for malformations is performed; there is wide variation in both expertise of staff and quality of equipment. Vaishali J Prajapati et al 2015[8] reported percentage of anemic mothers was 18.58 and this was less than our study percentage. Our study findings were similar to findings of studies conducted by Maimoona Qadir et al 2017 [9] where 12.8% had previous abortions. Percentage of taking medication during antenatal period in the studies conducted by Maimoona Qadir et al 2017 [9] was 9.4% as compared to our study findings which reported 13.7%. Percentage of mothers with eclampsia was 5.3 and 10.62 in the studies conducted by Jayasree S. et al 2018 [5] & Vaishali J Prajapati et al 2015 [8] and this similar to our study findings. Prasannajeet Kokate et al 2017 [10] reported 6% & Mithlesh Dewangan et al 2016 [11] reported 3.5% mother with diabetes. Uchenna Ekwochi et al 2018 [12] reported 32.3% death & Ritu Vyaset al 2016 [13] reported 34% death. Out of total 300 newborn with congenital malformations 93(31%) were having CNS malformations, followed by 72(24%) newborn were having skeletal malformations. Study conducted by Paramesh Pandala et al 2019 [3] reported most common system involved was CNS

(22.32%) and Marwa Shawky et al 2019 [14] reported commonly affected system as gastrointestinal system (38%) followed by skeletal system. There were 5 cases of clinically identifiable syndrome. Two newborn each (0.67%) were having Down syndromes and Pierre Robin syndrome. There was one case of Edward syndrome. Percentage of downs syndrome in the studies conducted by Paramesh Pandala et al 2019 [3], Akinlabi E. Ajao et al 2019 [5] and Shantisena Mishra et al 2018 [7] was quite high as compared to our study findings.

Above discussion shows that most commonly affected system in various study was different from each other and this may be because of difference in hospital set up, difference in geographical location of respective population and socioeconomic factors, environmental & chemical factors, nutritional status and habits, high consanguineous marriages and also the study population, type of study design, inclusion criteria etc.

Conclusions

In India, prevalence of congenital anomalies remains high, third most common cause of perinatal mortality, hence emphasis should be given to early detection and intervention. Also increased awareness about preventable risk factors may help in reducing the burden of congenital anomalies on perinatal morbidity and mortality and improve the outcome. Preventive and interventional strategies will reduce the incidence of birth defects, decrease the burden, and improve quality of life.

Recommendation

Careful screening and premarital counseling for possible congenital malformations becomes mandatory to keep an account of incidence and prevalence of congenital anomalies in the society. Early detection can give a chance to families for deciding upon the continuation of pregnancy, especially in anomalies which are not compatible with

life where termination of pregnancy can be opted. Mothers with bad obstetric history and those with previous anomalous baby should be thoroughly screened antenatally as it has considerable repercussion on the mothers and the families. Finally increasing awareness about maternal care during pregnancy (for example supplementation of folic acid to high risk mothers can decrease the risk of CNS malformations), enlightening people with consequences of consanguineous marriages, early diagnosing congenital anomalies by prenatal diagnostic studies and by thorough clinical examination and diagnostic studies at birth can help in early intervention and better outcome and hence considerably decrease the perinatal morbidity and mortality.

Data Availability

The datasets used and/or analyzed during the current study available from the corresponding author on reasonable request.

Acknowledgements

The study group is grateful to all staff and members of NICU team who supported this work

Author's contributions

Siddhi Gawhale and Ellies Sawarbandhe helped in the study design, acquisition of data, and drafting the manuscript. Siddhi Gawhale, Ellies Sawarbandhe and Ashwin Vekhande were responsible for conception of the idea, study design, analysis of the data, and drafting of the first manuscript. Ellies Sawarbandhe is the senior author who was responsible for supervision of the whole research and revising the final manuscript. All authors approved the manuscript and agreed to be accountable for all aspects of the work

Conflict of interest

The authors declare that they have no known competing of interests

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Date received: 9th July 2024, accepted 17th September 2024

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Citation: Ellies D. Sawarbandhe; Ashwin Vekhande; Siddhi Gawhale. "Comprehensive Study of Congenital Malformations in Newborn Babies in A Tertiary Indian Care Hospital: A Prospective Observational Study". *Annals of Neonatology*, 2025, 7(1): 62-72 doi: 10.21608/anj.2024.314508.1101