International Journal of Contemporary Pediatrics
Kaur B et al. Int J Contemp Pediatr. 2022 Jul;9(7):674-679
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20221612

Original Research Article

Pattern of congenital malformations in new-borns: a hospital based retrospective study

Baljinder Kaur, Surinder Kaur*, Tanya Thakkar, Kirti Sood, Navreet, Avneet

Department of Pediatrics, Government Medical College, Patiala, Punjab, India

Received: 12 May 2022
Revised: 02 June 2022
Accepted: 09 June 2022

***Correspondence:**
Dr. Surinder Kaur,
E-mail: drsurinderkaur21@gmail.com

ABSTRACT

Background: Congenital malformation in new-borns is a major public health problem. Identification of the pattern of congenital malformations and hence control of birth defects is an urgent need.
Methods: This retrospective study was carried out in neonatal care unit (NICU) of GMC and RH Patiala, Punjab. All the deliveries conducted from January 2020 to December 2020 were included in the study. All the new born babies were thoroughly examined for the presence of congenital malformations. Detailed maternal history was recorded so as to evaluate association of various maternal risk factors with the congenital malformations. Collected data was analyzed on statistical package for the social sciences (SPSS) software, a p value of less than 0.05 was considered significant.
Results: A total of 3962 babies were delivered at GMC and Rajindra Hospital, Patiala during the study period. Out of this, 91 (2.29%) new-borns were identified with congenital malformations. The most common system involved was musculoskeletal 40 (43.9%) followed by central nervous system 24 (26.4%). Among maternal and fetal risk factors; parental consanguinity, maternal under nutrition/obesity, positive history of a congenital anomaly (CA) in the family, and still birth/intrauterine deaths and ambiguous sex of the newborn baby were significantly associated with higher frequency of CAs ($p<0.05$).
Conclusions: Congenital malformations in new-borns becoming the emerging cause of neonatal morbidity and mortality after neonatal infections. Institution of preventive measures with more focus on young mother's nutrition, provision of health education and early diagnosis of congenital malformations during antenatal period can help to curb the burden of this problem.

Keywords: Anomalies, Malformations, Neonatal, Antenatal, Congenital

INTRODUCTION

Congenital malformations or birth defects are the anomalies affecting body's structure, function or metabolism that are present at birth. Birth defects, congenital abnormalities and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects those are present at birth.[1] Birth defects are a diverse group of disorders of prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. Maternal infections such as rubella, maternal illnesses like diabetes mellitus (DM), iodine and folic acid deficiency, exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals, and doses of radiation are all other factors that cause birth defects.[2]

Birth defects can be classified according to their severity, system involved and whether they are involving a single system or multiple systems. It is a major public health problem globally. Annual estimates show the presence of serious birth defects in 7.9 million newborns.[2] Middle and low income countries account for more than 94% of births

with serious birth defects.[3] Genetics is the etiological factor in (30-40%), environmental in 5-10% and cause is unknown in nearly 50% of the congenital malformed babies.[4] Worldwide surveys have shown the prevalence of congenital anomalies between 1.07-4.3/100 births.[5] Data from India shows the prevalence between 2-3%.[6] Ecological, social, racial and economic factors play an important role in the geographical variation of birth defects. Considerable variation in frequency of CAs in different populations has been reported, from as low as 1.07% in Japan to as high as 3% in Taiwan. This wide variability could be due to the ecological variations of health facilities used for data collection in different studies.[7] The burden of this problem is still underestimated in the developing world due to the lack of healthcare diagnostic facilities in rural areas and lack of accuracy and adequacy of health care statistics.[8] The birth of a congenitally malformed neonate imparts enormous stress and burden to the affected families.[9] Identification of the various risk factors and creating public awareness can help to reduce the burden of this problem.[10] Early diagnosis of CAs by level 3 and level 4 antenatal ultrasounds in 1st and 2nd trimester of pregnancy are strong preventive measures.[11] Thus focus on the importance of institution of preventive and diagnostic measures as a tool for detection of congenital malformations in newborns can help to curb the problem.[12] The present study was planned to highlight the pattern of congenital anomalies and its incidence, in this population of North India. This study also included the assessment of various risk factors during antenatal and intranatal period, and their association with occurrence of congenital malformations in newborn babies, so that some preventive measures could be formulated. Further this study stressed on the importance of carrying out a thorough clinical examination of all newborn babies before discharge from hospital.

METHODS

A retrospective study was conducted at neonatal intensive care unit (NICU) of Government Medical College and Rajindra Hospital, Patiala, Punjab. The study period was from January 2020 to December 2020. All the babies born during this period were included in the study. Immediately after birth all the newborn babies were shifted to NICU and examined thoroughly by paediatrician on duty for the presence of any congenital malformations. All the babies were thoroughly examined at 24 hours and 48hours of life to confirm the presence of birth defect and appearance of any new signs and symptoms related to CAs. Investigations like X-ray, ultrasound and echocardiography were done later for establishment of appropriate diagnosis. Based on International Classification of Diseases-10 (ICD-10) classification, system wise categorization of all the CAs were done. Babies born before 37 weeks (less than 259 days) completed of gestation were considered as preterm, 37-41 completed weeks (259-293 days) as term NB and babies born after 42 weeks (after 294 days) of pregnancy were labelled as post term.[12] Depending on birth weight, neonate with birth weight less than 2.5 kg were classified as low birth weight (LBW), birth weight less than 1500 gm up to 1000 gram as very low birth weight (VLBW) and birth weight and less than 1000 gm as extremely low birth weight (ELBW) respectively.[12] Infants with birth weight <10th percentile of the expected weight for gestational age and sex of the baby were considered as SGA babies. Infants with birth weight >90th percentile of the expected weight for gestational age and sex of the newborn were considered as LGA babies. Infants with birth weight between 10th and 90th percentile were labelled as appropriate for gestational age (AGA) babies.[13,14] Neonate was considered as live birth when the product of conception, irrespective of weight or gestational age, and that, after separation from mother, shows signs of life such as breathing, heartbeat, pulsation of umbilical cord or definite voluntary muscles movements. When a product of conception that, after separation from mother, does not show any evidence of life is known as foetal death. A foetal death at a gestation of 22 weeks or more or weight 500 gram or more at birth is considered as stillbirths.[12] Detailed history was recorded from mother/attendant which included socio-demographic profile, age of both the parents, birth order, gestational age, mode of delivery, whether spontaneous conception or with treatment, family history of congenital malformation in siblings or in the family. Maternal history of fever with rash, exposure to drug/radiation, cigarette smoking and alcohol/substance abuse especially in 1st trimester of pregnancy was recorded. Details of medical/surgical ailments and any pregnancy related complications in mother were recorded. Details of antenatal visits and investigations done during this period with special attention to antenatal ultrasonographs for foetal well-being were recorded. Note was made of the any complication during intranatal period and delivery. History of consanguineous marriage was asked. Growth parameter record of the newborn included the weight, length and head circumference. Collected data was analysed using statistical package for the social sciences (SPSS) software. Frequencies and percentage of various factors were presented. Chi square and p value test were applied and a p value less than 0.05 were considered as statistically significant.

Inclusion criteria

All the babies delivered at the institute were included in the study.

Exclusion criteria

VLBW babies less than 1000 gm, babies delivered before 32 weeks of gestation (very preterm babies) and still born babies were excluded from the study.

Ethical issues

Before the initiation of this study, ethical clearance was sought from the institute's ethical committee.

RESULTS

During the study period, 3962 new born babies were shifted to NICU of Rajindra Hospital, Patiala. Congenital malformations were detected in 91 new born babies; hence an incidence of 2.29% was reported. Out of this, 84 (92.3%) new born babies were live births and 7 (7.6%) were Intrauterine deaths. Mode of delivery was normal in 58 (63.7%) and caesarean section in 33 (36.2%). Most of the deliveries were full term 52 (54.7%) and only 39 (41.1%) were pre term delivered between 32 to 36 weeks of gestation. Gender wise distribution of newborns with congenital malformations shows 50 (52.6%) were males, 40 (42.1%) were females and 1 newborn baby (1.1%) was having with ambiguous genitalia. Average weight of the NBs with CAs recorded was 2.72 kg (1.9-3.8 kg), average head circumference was 33.3 cm (30.0-36.0 cm) and average length was 48.5 cm (44-52 cm). Age distribution of mothers showed that maximum number 73 (80.2%) of mothers were in the age group of 21 to 30 years; followed by 31-40 years 11 (12.1%) and only 7 (7.7%) mothers were less than 20 years of age group.

Majority of neonates were 1st birth order 48 (52.7%) and 2nd in 37 (40.6%), and only 1 (1.09%) was 6th in birth order. Conception was spontaneous in all the mothers. History of pregnancy induced hypertension was recorded in 22 (24.1%). They were taking drugs for high blood pressure in last trimester only (Table 1).

Table 1: Characteristics of births with congenital malformations.

Characteristic	Congenital malformation mean (SD)
Mother's age	26.21 (3.59)
Birth order	2.11 (1.37)
Period of gestation	37.74 (2.13)
Weight of baby (kg)	2.72 (0.62)
Length (cm)	47.82 (2.63)
Head circumference (cm)	33.32 (1.48)

Antenatal ultrasound showed congenital anomalies in 46 (50.6%) mothers. Anomalies detected on antenatal ultrasonography were those pertaining to musculoskeletal, genitourinary and cardiovascular system. The most common system involved was musculoskeletal 40 (43.9%) followed by central nervous system; 24 (26.4%) and genitourinary system in 7 (7.6%) study population (Table 2).

There was history of siblings affected with congenital malformations in 12 (13.1%). Affected siblings had involvement of musculoskeletal system manifested as congenital talipes equinovarus. It is statistically significant. Congenital anomalies in new born with ambiguous genitalia were not compatible for life and caused intrauterine death of the baby. This was statistically significant (Table 3).

Table 2: Distribution of congenital malformations based on ICD-10 classification.

System/malformation	No. (%)
Musculoskeletal system	40 (43.9)
Congenital talipes equinovarus	
Right foot	10 (10.5)
Left foot	7 (7.4)
Both sides	14 (14.7)
Achondroplasia	2 (2.1)
Polydactyle	7 (7.4)
Nervous system	24 (26.37)
Hydrocephalous	11 (11.6)
Meningomyelocele + spina bifida + hydrocephalus	4 (4.3)
Spina bifida with chest wall deformity	1 (1.1)
Anencephaly	1 (1.1)
Anencephaly + encephalocoele + gastrochiasis	3 (3.2)
Corpus calosum absent	2 (2.1)
Microcephaly	2 (2.1)
Cardiovascular	2 (2.1)
Situs inversus with complex cardiac disease	1 (1.1)
Congenital acyanotic heart disease (ASD)	1 (1.1)
Genitourinary	7 (7.6)
Multicystic dysplastic kidney	5 (5.3)
Undescended testis	2 (2.1)
Other systems	16
Cleft lip	1 (1.1)
Cleft lip with cleft palate	4 (5.3)

Continued.

System/malformation	No. (%)
Skin tags	2 (2.1)
Low set ear	1 (1.1)
Omphalocoele	1 (1.1)
Single umbilical artery	5 (5.4)
Multiple anomalies	4 (4.2)
Total	91 (100)

Table 3: Congenital malformations in relation to maternal and fetal factors.

Factors	Total	Number of malformed neonates	%	P
Maternal age (years)				
Less than 20	305	7	2.29	>0.05
21 to 30	3178	73	2.29	
31-40	479	11	2.29	
Maternal nutrition				
Normal weight	3329	76	2.28	
Undernourished	554	13	2.3	<0.05
Obesity	79	2	2.5	
History of a congenital anomaly in the family				
Positive	304	12	3.94	<0.05
Negative	3658	79	2.15	
Sex of the new born baby				
Male	2054	50	2.43	
Female	1907	40	2.09	0.00
Ambiguous genitalia	1	1	100	
Birth weight (gm)				
1000-1500	341	4	1.17	
1500-2499	1168	28	2.39	>0.05
2500-3500	2334	56	2.39	
>3500	119	3	2.52	
Terms of pregnancy				
Pre-term neonates	1624	39	2.40	0.00
Term neonates	2167	52	2.39	
Outcome of pregnancy				
Live births	3754	84	2.23	0.00
IUD	208	7	3.36	

DISCUSSION

As improvement in antenatal, perinatal and neonatal care, lead to reduction in neonatal infections, preterm births and number of LBW babies. All these factors helped in reduction of neonatal morbidity and mortality due to neonatal sepsis and congenital pneumonias.[15] However, the proportion of neonatal deaths due to congenital malformations is increasing. Hence in coming period, congenital malformations will be the leading cause of neonatal morbidity and mortality.[16] Because present study was conducted at a tertiary care referral hospital, incidence of congenital malformations recorded in newborns was 2.29% which was comparatively less to the earlier study done in the same region in year 2012-13, which showed an incidence of congenital malformations in new born babies as 4.44%.[17] Congenital malformation were more in newborns with birth order of 4 or more, but in present study majority of neonates with CAs were in 1st 48 (52.7%), and 2nd 37 (40.6%) in birth order.[16]

In our study CAs were more in preterm 2.40% of total deliveries as compared to term babies (2.39%) which was comparable to earlier studies which showed higher incidence of congenital malformations in preterm babies.[16] In a study from Lebanon, family history of congenital malformations was present in 12 (13.1%) of study population, similar results were obtained in our study also.[17] There was a statistically significant association between maternal malnutrition (under nutrition/obesity), history of congenital malformations in family (3.94%) in present study. Luck in his study value of routine ultrasound scanning at 19 weeks found that some CAs were not compatible for life lead to intrauterine devices (IUDs) or still births.[21] Similar results were obtained from present study, neonate with ambiguous genitalia had 100% mortality (IUD).

Most studies have demonstrated detection rate of CAs on antenatal ultrasonography (USG), as low as 13-16% to as high as 85% but in present study it was 50.6%.[18-20]

System involved were musculoskeletal, genitourinary and cardiovascular. Similar anomalies were detected on antenatal ultrasound i.e. gastroschisis, extra renal dilated pelvis with mild calyceal separation, left multicystic dysplastic kidney and cardiomegaly.[18-21] A study by Bai in year 1982 reported that CAs were more in low birth weight infants and in neonates born to mothers with age above 35 years but opposite results were obtained in our study, majority of mothers of congenitally malformed babies were in age group of 21-30 years and no specific birth weight group predilection recorded.[20] This suggests focusing more on younger age group mothers in the form of preventive health care measures like compulsory institution of healthy diet rich in folic acid and other nutrients to women who are planning pregnancy and in first trimester of pregnancy. Antenatal ultrasound for detection of congenital malformations in early pregnancy should be available at a nearby health facility, so as to reduce the burden of problem. Hence health awareness among the women in reproductive age group should be provided at all levels of health care.

Limitations

The results of the present study couldn't be generalized as this study was from tertiary care institute which is a referral centre.

CONCLUSION

Congenital malformations in new-borns are becoming the emerging cause of neonatal morbidity and mortality after neonatal infections. The birth of a congenitally malformed baby imparts enormous stress to the affected families. Institution of preventive measures with more focus on young mother's nutrition, provision of health education and early diagnosis of congenital malformations during antenatal period can help to curb the burden of this problem.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: The study was approved by the Institutional Ethics Committee

REFERENCES

1. World Health Organization. Birth defects. Executive Board, 126th session, provisional agenda, December 2009. Available at: http://apps.who.int/gb/ebwha/pdf_files/EB1 26/B126_10-en.pdf. Accessed on 24 March 2022.
2. Lawn JE, Cousin S, Zupan J. Lancet neonatal Survival Steering team 4 million Neonatal deaths: When? Where? Why? Lancet. 2005;365:891-900.
3. Arnold S, Christopher PH, Bernadetti M. Global report on birth defects: New York: Birth defects foundation. 2006.
4. Rajangam S. Consanguinity and chromosomal abnormality in mental Retardation and or multiple congenital anomalies. J Anatomical Soc India. 2007;56:30-3.
5. Temtamy SA. A genetic epidemiological study of malformations at birth in Egypt. Eastern Mediterranean Health J. 1998;4:252-9.
6. Reich JD. Clinical screening for congenital heart diseases at birth –A long way to go. Indian Pediatrics. 2011;48:17-8.
7. Jahangir W, Ali F, Jahangir T, Masood MS. Prevalence of gross congenital malformations at birth in the neonates in a tertiary care hospital. APMC. 2009;3:47-50.
8. Marwah S, Sharma S, Kaur H, Gupta M, Goraya SPS. Surveillance of congenital Malformations and their possible risk factors in a teaching hospital in Punjab. Int J Reprod Contracept Obstet Gynecol. 2014;3162-7.
9. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G, et al. Congenital malformation in newborn: A study of 10,874 consecutive births. J Anat Soc India. 1989;38:101-11.
10. Mathur BC, Karan S, Vijaya Devi KK. Congenital malformations in the newborn. Indian Pediatr. 1975;12:179-83.
11. Suguna Bai NS, Mascarene M. An etiological study of congenital malformation in the newborn. Indian Pediatr. 1982;19:1003-7.
12. Paul VK, Bagga A. Ghai Essential Pediatrics Book (Ninth Edition). CBS Publishers. 2019;125-6.
13. Paul VK, Bagga A. Assessment of Size and Growth. Ghai Essential Pediatrics Book (Ninth Edition). CBS Publishers & Distributors Pvt Ltd. 2019.
14. WHO Growth charts. Available at: https://www.who.int/tools/child-growth-standards/standards. Accessed on 15 March 2022.
15. Pattern of congenital anomalies in newborn: a hospital-based study
16. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital-based study. Pediatr Rep. 2013;5:e5.
17. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. Indian Pediatr. 1994;31:1187-91.
18. Francine R, Salameh P, Aline H. Congenital anomalies: Prevalence of Congenital Anomalies in Neonates and Associated Risk factors. J Public Health. 2014;2(2):58-63.
19. Sridhar K. A community based study of visible congenital anomalies in rural Tamil Nadu. Indian J Plastic Surg. 2009;42:184-91.
20. Levi S, Hyjazi Y, Schaapst JP, Defoort P, Coulon R, Buckens P. Sensitivity and Specificity of routine antenatal screening for congenital anomalies by ultrasound: the Belgian Multicentric Study. Ultrasound Obstet Gynecol. 1991;1:102-10.

21. Crane J, LeFevre ML, Winborn RC, Evans JK, Ewigman BG, Bain RP, et al. A Randomized trial of prenatal ultrasonographic screening: impact on the detection, management and outcome of anomalous foetuses. Am J Obstet Gynecol. 1994;171:392-9.
22. Luck CA. Value of routine ultrasound scanning at 19 weeks: a four year study of 8849 deliveries. BMJ. 1992;304:1474-8.

Cite this article as: Kaur B, Kaur S, Thakkar T, Sood K, Navreet, Avneet. Pattern of congenital malformations in new-borns: a hospital based retrospective study. Int J Contemp Pediatr 2022;9:674-9.