International Journal of Contemporary Pediatrics
Chikkaiggaluru SN et al. Int J Contemp Pediatr. 2026 Aug;13(8):1372-1377
https://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20262410

Original Research Article

Early detection of congenital anomalies in newborns: a tertiary hospital's approach to screening and risk assessment

Samhitha Narayanappa Chikkaiggaluru[1], Deepthi Ramamurthy[2], Balakrishnan Ramanan[2]*, Pushpalatha Kariyappa[3]

[1]ESIC Dispensary, Jwalapuri, New Delhi, India
[2]Department of Community Medicine, ESIC Medical College and PGIMSR, Rajajinagar, Bengaluru, India
[3]Department of Pediatrics, ESIC Medical College and PGIMSR, Rajajinagar, Bengaluru, India

Received: 04 April 2026
Accepted: 01 July 2026

***Correspondence:**
Dr. Balakrishnan Ramanan,
E-mail: bala1096@gmail.com

ABSTRACT

Background: The spectrum of congenital anomalies ranges from minor issues like polydactyly to conditions unsustainable with life like anencephaly. The impact is on the affected children, parents or caregivers and the society at large. The most dreaded concern is death of the child. This study aims to estimate the prevalence of congenital anomalies and associated risk factors among neonates in a tertiary care hospital in Bengaluru, India.
Methods: A cross-sectional analysis was conducted over six months, involving 202 neonates. Data were collected via validated questionnaires covering socio-demographic details, comprehensive antenatal history, birth history, and the presence of congenital anomalies. Settings and design was hospital-based cross-sectional study. Statistical analysis used: The association between risk factors and congenital anomalies was assessed using Chi-square tests and Odds ratios.
Results: Out of 202 neonates included, the proportion of congenital anomalies was 8.4% (17) with congenital heart diseases (07) being the commonest followed by congenital talipes equino varus (CTEV) (04). Foetal risk factors for disability were observed among 93 (46%) of the neonates. Common neonatal risk factors observed were neonatal jaundice, Low Birth Weight, prematurity and neonatal asphyxia. Congenital anomalies were slightly higher among boys: 13 (11.7%) than girls: 4 (4.3%). Proportion of foetal risk factors was similar among boys and girls.
Conclusions: The study highlights a higher prevalence of congenital anomalies than the national average, emphasizing the need for mandatory congenital anomaly screening and establishing a comprehensive congenital malformation registry in India.

Keywords: Birth defect, Risk factors, Childhood disability, Screening, Congenital anomalies

INTRODUCTION

Congenital anomalies are defined as structural or functional anomalies that occur during intrauterine life.[1] These conditions, which may be identified before or at birth or later in life, can affect an organ or an entire organ system. The causes of congenital anomalies are often genetic, environmental, or a combination of both. For instance, neural tube defects in newborns can result from ante-natal insults to the fetus or nutritional deficiencies like maternal folate deficiency. The spectrum of congenital anomalies ranges from minor issues such as polydactyly, which are primarily cosmetic concerns, to severe conditions like anencephaly, which are incompatible with life. The impact of congenital anomalies is extensive, affecting not only the affected children but also their parents or caregivers and society at large. The most serious concern is the high death rate among these children.

According to the World Health Organization, globally about 4,10,000 children with congenital anomalies die within five years out of which 2,40,000 (59%) newborns die within 28 days of birth. Lifelong disability and social stigmatization are other major problems that are of concern.[2] Parents of children with congenital anomalies experience significantly elevated traumatic stress levels from diagnosis until the child reaches 10-12 years of age compared to parents of children without congenital anomalies.[3]

That being the case, screening newborns for congenital anomalies is critical as it helps in identifying the same at a stage when they are still amenable for medical interventions and support. Ultimately, it would not only improve the health outcomes of affected infants, but also reduce the burden of long-term disabilities and healthcare costs on families and society. Screening can be done through collection of a detailed history, complete physical examination of the newborn and specific laboratory investigations as the case may be. A classic example of early diagnosis through screening leading to high success rates of treatment is the identification of club foot (congenital talipes equinovarus); it can be picked during a simple head-to-toe examination of the newborn.[4]

Worldwide 7.9 million births occur annually with serious anomalies and 94% of these births occur in the middle- and low-income countries.[5] A meta-analysis by Bhide et al found the prevalence of congenital anomalies in India to be 184.48 per 10,000 births.[6] The Indian population has a lot of inherent risk factors, for instance, inadequate to no supplementation of pre-pregnancy folic acid leading to congenital anomalies. Despite this, congenital anomalies remain one of the least focused areas of disease surveillance in India, which lacks a nationwide congenital anomaly registry and has limited studies estimating the burden of these conditions.[7]

The study at a tertiary care teaching hospital in Bengaluru, India, aims to estimate the proportion of congenital anomalies among newborns and examine associated fetal and maternal risk factors. Addressing key knowledge gaps, it highlights the lack of comprehensive data on congenital anomalies in India due to the absence of a national registry. Additionally, it explores specific risk factors pertinent to the Indian context, including genetic, nutritional, and environmental influences. By focusing on these objectives, the study provides valuable insights into the burden of congenital anomalies in India and contributes to the development of targeted prevention and intervention strategies. This research emphasizes the importance of localized data and context-specific approaches to improving maternal and child health outcomes.

METHODS

A cross-sectional study was conducted by the Departments of Community Medicine and Paediatrics, at a tertiary care teaching hospital in South India. The hospital handles around 300 deliveries every month.

The study was conducted over a period of six months from March 2022 to August 2022; study population included the neonates born at the hospital. Informed consent was obtained from the mothers.

A detailed questionnaire was created using Google forms and validated by two experts: one from community medicine and another from paediatrics. Internal consistency was assessed by Cronbach's alpha which was >0.7. After the suggested changes were incorporated, the tool was piloted on 5% of the estimated sample size.

It consisted of the following five parts was administered to the mothers by the investigators: Socio-demographic details, comprehensive antenatal history including details like peri-conceptional folate consumption and previous obstetric history, birth history including history on mode of delivery, neonatal resuscitation, NICU admission, presence/ absence of any visible congenital anomalies and investigations done including antenatal ultrasonography.

Newborn screening involved a systematic approach to detect birth defects and abnormalities shortly after birth. A brief physical exam checks vital signs like heart rate and respiration, followed by a thorough "head to toe" and "front to back" assessment. Key components include examining the general appearance, head, spine, face, eyes, ears, mouth, chest, abdomen, genitalia, urinary tract, limbs, and chromosomal indicators. Abnormal findings were noted and prompt urgent referrals for further evaluation and management was done. All positive findings in the head-to-toe examination and systemic examination were recorded. A list of 48 anomalies will be looked for and the results of the same was compiled.

Sample size was calculated based on a study done by Taksande et al.[8] Relying on prevalence of congenital anomalies at birth to be 1.91% with 95% confidence level and an absolute precision of 2%, using Epi info software, the sample size was estimated to be 180. However, to account for factors like non-co-operation and incompleteness of data recording, a total of 200 neonates were studied. All newborns born at our hospital between March 2022 to August 2022 were included in the study.

Parents who did not give consent, newborns referred to other hospital for specialized care and newborns referred from other centres to our hospital were excluded from the study. Simple random sampling method was used to select the required sample size. Ethical clearance was duly obtained from institutional ethical committee (No. 532/L/11/12/Ethics/ESICMC&PGIMSR/Estt.Vol..IV dated 23.04.2021). Throughout the study, patient confidentiality was maintained by censoring personal

identifiers and the final report was presented in aggregate numbers only.

Data entry was done on Microsoft excel and analysed using Epi info 07 software. The categorical variables were presented as frequencies and proportions (percentage). Contingency tables were prepared for the association between risk factors and presence of congenital anomalies. Chi-square test was applied to draw the association between categorical variables. P<0.05 was considered to be statistically significant. Strength of association between risk factors and congenital anomalies was estimated by calculating Odd's ratio with and without adjusting for all the other confounding factors by logistic regression modelling.

RESULTS

Out of the 202 neonates studied, 54% were boys, 95% children from singleton pregnancies and about 65% delivered through normal delivery as shown in Table 1.

Table 1: Characteristics of the study subjects.

Characteristics		N	Percentage (%)
Gender	Boys	109	54.0
	Girls	93	46.0
Number of children	Single	192	95.0
	Multiple	10	4.9
Mode of delivery	Normal	131	64.8
	Assisted	71	35.1
Total		202	100

The proportion of congenital anomalies was found to be 8.4% as shown in figure 1, i.e., 17 of the 202 neonates had a congenital anomaly. The various congenital anomalies detected were as follows: Cardiac anomalies-8, CTEV-4, two cases of paraphimosis and one case each of Anotia, polydactyly and multicystic renal dysplasia. (Figure 2).

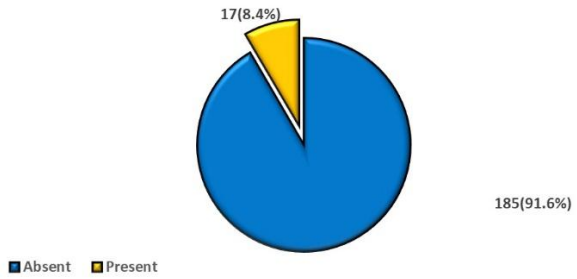

Figure 1: Proportion of congenital anomalies among the neonates included.

Table 2 shows the proportion of fetal outcome and maternal risk factors among newborns regardless of the presence or absence of congenital anomalies. The top three maternal risk factors identified were history of associated maternal illness (62.4%), parental consanguinity (17.3%) and preterm delivery (15.3%). Among the foetal prolonged hospitalization was found to be the most common maternal risk factor, present in 46% of the deliveries. Among the neonatal risk factors, prolonged hospitalization (46%), neonatal jaundice (23.8%) and low birth weight (22.8%) topped the list.

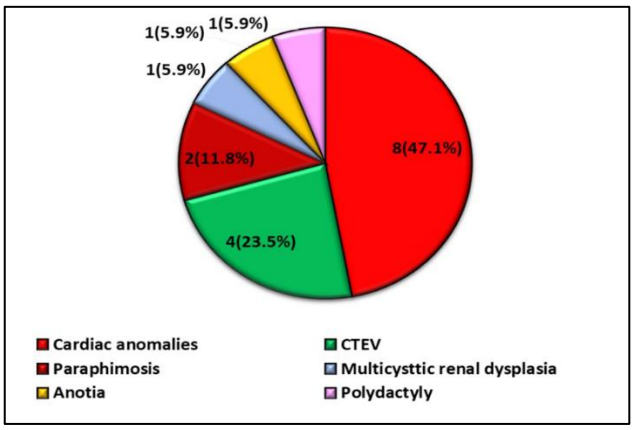

Figure 2: Various types of congenital anomalies detected among the neonates.

Table 2: Proportion of fetal outcomes and maternal risk factors and in the study participants.

Risk factors	N	Percentage (%)
Foetal outcome		
Prolonged hospitalization	93	46.04
neonatal jaundice	48	23.76
Low birth weight	46	22.77
Birth asphyxia	25	12.38
≥3 birth order	17	8.42
neonatal hypothyroidism	6	2.97
Neonatal hypoglycaemia	2	0.99
Maternal risk factors		
H/O ass maternal illness	126	62.38
Parental consanguinity	35	17.33
Preterm delivery	31	15.35
periconceptional folic acid not taken	13	6.44
Assisted conception IVF	8	3.96
H/O of previous still birth	6	2.97
h/o anomalies in previous pregnancy	6	2.97
Elderly primi	4	1.98

Table 3 shows association between various risk factors that were studied and presence of congenital anomalies. Congenital anomalies were significantly associated with perinatal non-consumption of folic acid (COR-5.92 (CI:1.42-21.86)), preterm delivery (COR-4.64 (CI:1.54-13.56)) and presence of any of the maternal or fetal risk factors (COR-6.21 (CI:1.85-27.83)) in comparison to their counter parts. However, when the variables were adjusted for risk factors and gender, positive association

was established between low birth weight and presence of congenital anomalies. It was estimated that neonates with congenital anomalies were 10.15 times (CI: 2.09-49.18) more likely to have been exposed to risk factor compared to those without congenital anomalies. Specifically, those newborns with congenital anomalies were 5.89 times (CI: 1.11-31.21) more likely to have had low birth weight.

Table 3: Association selected variables with congenital anomalies among study participants.

Variables		Congenital anomalies			
		Absent (%)	Present (%)	Crude OR (95% CI)	Adjusted OR (95% CI)
Gender	Boys	96 (88.07)	13 (11.93)	0.33 (0.09-1.02)	0.36 (0.10-1.34)
	Girls	89 (95.70)	4 (4.30)		
Perinatal folic acid consumption	No	9 (69.23)	4 (30.77)	5.92 (1.42-21.86)*	3.55 (0.64-19.80)
	Yes	176 (93.12)	13 (6.88)		
Any risk factors	No	106 (97.25)	3 (2.80)	6.21 (1.85-27.83)*	10.15 (2.09-49.18)*
	Yes	79 (84.95)	14 (15.1)		
Previous H/O abortions	No	137 (91.33)	13 (8.70)	0.88 (0.24-2.73)	0.82 (0.18-3.75)
	Yes	48 (92.31)	4 (7.69)		
Previous H/O stillbirths	No	181 (92.35)	15 (7.65)	5.93 (0.71-36.26)	9.18 (0.49-172.22)
	Yes	4 (66.7)	2 (33.3)		
Previous H/O anomalies	No	180 (91.8)	16 (8.2)	2.24 (0.09-17.44)	0.22 (0.02-3.02)
	Yes	5 (83.3)	1 (16.7)		
h/o associated maternal illness	No	69 (90.8)	7 (9.2)	0.85 (0.31-2.47)	0.43 (0.13-1.49)
	Yes	116 (92.1)	10 (7.9)		
Birth order 3 or more	No	169 (91.4)	16 (8.6)	0.66 (0.03-4.08)	1.91 (0.17-21.89)
	Yes	16 (94.1)	1 (5.9)		
Low birth weight	No	144 (92.3)	12 (7.7)	1.46 (0.44-4.32)	5.89 (1.11-31.21)*
	Yes	41 (89.1)	5 (10.9)		
Preterm	No	161 (94.2)	10 (5.8)	4.64 (1.54-13.56)*	4.26 (0.90-20.17)
	Yes	24 (77.4)	7 (22.6)		
Birth asphyxia	No	163 (92.1)	14 (7.9)	1.58 (0.34-5.62)	0.66 (0.14-3.16)
	Yes	22 (88.0)	3 (12.0)		
Assisted delivery	No	121 (92.4)	10 (7.6)	1.32 (0.46-3.68)	0.55 (0.16-1.90)
	Yes	64 (90.1)	7 (9.9)		
Parental consanguinity	No	154 (92.2)	13 (7.8)	1.53 (0.47-5.00)	0.82 (0.17-3.39)
	Yes	31 (88.6)	4 (11.4)		

*P value<0.05.

DISCUSSION

The prevalence of congenital anomalies in this study was 8.4% and most common anomaly being congenital heart disease. Systematic review and meta-analysis by Bhide and Kar estimated that the pooled national birth prevalence of congenital anomalies was 1.84% and musculoskeletal anomalies were the most common anomalies reported among newborns.[9] Research conducted at a tertiary care center in India found that the overall prevalence of congenital anomalies was 1.82%.[10] Most commonly reported congenital anomalies by Kumar et al were malformations of the circulatory system (28.0%), followed by musculoskeletal (18.6%) and urinary system (14.3%) anomalies which was similar to our study. The most common anomaly reported is congenital heart disease, which is consistent with global patterns. Sarkar et al examined 12,896 babies born, 286 had congenital malformations, resulting in a prevalence of 2.22%.[11] The prevalence is much lower than what is reported in our study as they have studied huge population over a period of several decades. The higher prevalence observed in our study could also be due to the hospital's status as a referral center, which often receives more complex cases. Another reason for increased prevalence is inclusion of minor congenital anomalies that do not require medical intervention, unlike other studies which focused solely on major congenital anomalies. Persisting with the documentation of congenital anomalies at our hospital over the coming years might produce consistent results.

The predominance of congenital heart disease in the study highlights the need for specialized care and early diagnosis, which can be crucial for the survival and quality of life of affected infants. Prenatal screening and postnatal care, including echocardiography and, if necessary, cardiac surgery, are important components of the healthcare response to this challenge.

In this study congenital anomalies were significantly associated with perinatal non-consumption of folic acid, low birth weight, preterm delivery and presence of any of the maternal or fetal risk factors. The link between low birth weight and a heightened risk of congenital malformations has been firmly established in previous research. Our observations align with these findings, showing a notably higher occurrence of congenital anomalies in preterm infants relative to their full-term counterparts, consistent with earlier reports from within this nation. Sarkar et al has reported significant association of multiparous women, consanguineous marriage, low birth weight, normal vaginal delivery, preterm child and male child with congenital anomalies than compared to their counter groups.[11]

Epidemiological risk factors for congenital anomalies study have shown significant association with increased maternal age, lower level of education, multiparity, not taking folic acid supplementation, tobacco use, and a history of previous miscarriage than compared to others.[12]

Malik et al in their community-based study reported that mothers with previous history of congenital malformation (8.3%) and abortions (13.6%) had higher prevalence of congenitally malformed babies with 2.6- and 4-times higher odds of having a malformed baby, respectively.[13]

Sunitha et al in a hospital-based study done among high-risk mothers in South India reported factors which include maternal age 25 years or younger (with an odds ratio [OR] of 1.42), paternal age under 30 years (OR=1.51), first-time pregnancy (primigravida) (OR=3.40), and parental consanguinity (OR=1.39) being significantly associated with congenital anomalies.[14]

Overall, the study emphasizes the importance of comprehensive prenatal care, including the management of maternal illnesses and monitoring for signs of preterm labor. They also highlight the need for genetic counseling, especially in populations with high rates of consanguinity like India. After birth, the focus on managing and preventing prolonged hospital stays, neonatal jaundice, and low birth weight is crucial for improving neonatal outcomes

There are several limitations to our study. Establishing a definitive cause-and-effect relationship between congenital anomalies and maternal age is challenging, primarily because of the cross-sectional nature of the study. We did not account for a variety of risk factors that might influence the results, including the use of certain medications, obesity, alcohol intake, smoking habits all of which could potentially skew the findings. Sample size of study is small; hence generalization of the study results must be done with caution. The methods used to diagnose congenital anomalies is not comprehensive; anomalies identified within 7-10 days of birth are only included.

Mandatory screening for congenital anomalies and their risk factors at health centers in India enables early identification of abnormalities, allowing for timely interventions leading to better outcomes in these children. By identifying risk factors, health centers can offer targeted education and prevention strategies to at-risk populations.

This was an effort to build digital congenital malformation registry in the hospital. Though there is a birth defect registry in India, it covers only tip of the iceberg. The establishment and inclusion of all hospitals in congenital malformation registry in India is a critical need for several reasons, and such a registry would have a profound impact on public health, prenatal care, and medical research. A dedicated registry would provide a centralized, systematic collection of data, facilitating the analysis of prevalence rates, types of malformations, and geographical variations. It would enable the identification of potential etiological factors, including environmental, genetic, and socio-economic factors, thus aiding in the implementation of preventive measures. It will also support the monitoring of trends over time, which is crucial in evaluating the effectiveness of public health interventions, such as folic acid supplementation programs, which have been shown to significantly reduce the incidence of neural tube defects. It could guide healthcare policy, enabling the government and health organizations to prioritize areas of need, develop specialized healthcare services, and initiate genetic counseling services where there is a high incidence of consanguinity-related anomalies.

CONCLUSION

In conclusion, there is increased prevalence of congenital anomalies and its risk factors which warrants the establishment of a congenital malformation registry in India and mandating all hospitals to report is an urgent requirement. It would not only serve as a cornerstone for public health policy and prenatal care improvement but also provide a wealth of data to support ongoing and future research efforts aimed at reducing the burden of congenital anomalies.

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. Congenital disorders. Geneva: WHO. Available at: https://www.who.int/health-topics/congenital-anomalies. Accessed on 08 March 2026.
2. Kar A, Dhamdhere D, Medhekar A. "Fruits of our past karma": a qualitative study on knowledge and attitudes about congenital anomalies among women

in Pune district, India. J Community Genet. 2023;14(4):429-38.
3. Oftedal A, Bekkhus M, Haugen G, Hjemdal O, Czajkowski NO, Kaasen A. Long-term impact of diagnosed fetal anomaly on parental traumatic stress, resilience, and relationship satisfaction. J Pediatr Psychol. 2023;48:181-92.
4. Barrie A, Varacallo M. Clubfoot. In: StatPearls. Treasure Island (FL): StatPearls Publishing. 2023.
5. Christianson A, Howson CP, Modell B. Global report on congenital anomaly: the hidden toll of dying and disabled children. New York: March of Dimes. 2006.
6. Bhide P, Kar A. A national estimate of the birth prevalence of birth defects in India: systematic review and meta-analysis. BMC Pediatr. 2018;18(1):10.
7. Cardoso-dos-Santos AC, Alves RS, Medeiros-de-Souza AC, Bremm JM, Gomes JD, Alves RF, et al. National congenital anomaly registers in the world: historical and operational aspects. Epidemiol Serv Saude. 2021;30(4):e2021075.
8. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: a rural medical college hospital-based data. Indian J Hum Genet. 2010;16(3):159.
9. Bhide P, Kar A. A national estimate of the birth prevalence of congenital anomalies in India: systematic review and meta-analysis. BMC Pediatr. 2018;18(1):175.
10. Kumar J, Saini SS, Sundaram V, Mukhopadhyay K, Dutta S, Kakkar N, et al. Prevalence & spectrum of congenital anomalies at a tertiary care centre in north India over 20 years (1998-2017). Indian J Med Res. 2021;154(3):483-90.
11. Sarkar S, Patra C, Dasgupta MK, Nayek K, Karmakar PR. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India. J Clin Neonatol. 2013;2(3):131-4.
12. Almeida LF, Araujo Júnior E, Crott GC, Okido MM, Berezowski AT, Duarte G, et al. Epidemiological risk factors and perinatal outcomes of congenital anomalies. Rev Bras Ginecol Obstet. 2016;38(7):348-55.
13. Malik M, Khanna P, Verma R. Association of maternal risk factors to congenital anomalies among infants: a community-based study in rural areas of Haryana, India. J Assoc Physicians India. 2019;67(9):38-41.
14. Sunitha T, Prasoona KR, Kumari TM, Srinadh B, Deepika MLN, Aruna R, et al. Risk factors for congenital anomalies in high-risk pregnant women: a large study from South India. Egypt J Med Hum Genet. 2016;18(1):79-85.

Cite this article as: Chikkaiggaluru SN, Ramamurthy D, Ramanan B, Kariyappa P. Early detection of congenital anomalies in newborns: a tertiary hospital's approach to screening and risk assessment. Int J Contemp Pediatr 2026;13:1372-7.