

**Prevalence and Pattern of Congenital Anomalies in Newborn in a Tertiary Care Centre, Located in a Hill District of Assam**Veemi Borah<sup>1</sup>, Amit Das<sup>2</sup><sup>1</sup>Associate Professor, Department of Paediatrics, Diphu Medical College, Assam<sup>2</sup>Medical Social Worker, Department of Community Medicine, Diphu Medical College, Assam

Received: 25-03-2024 / Revised: 23-04-2024 / Accepted: 26-05-2024

Corresponding Author: Dr. Veemi Borah

Conflict of interest: Nil

**Abstract:**

**Introduction:** World Health Organization (WHO) defines 'Congenital Anomaly' as structural or functional defect which occurs during intrauterine life. Globally prevalence of congenital anomaly is found to be 2-3% and about 2,40,000 infants lose their lives due to congenital malformation every year.

**Aim:** Aim of the study was to find out the, prevalence and pattern of Congenital anomalies in newborn in a tertiary care centre in Karbi Anglong of Assam, as there is paucity of information and publications on congenital anomalies in this part of Assam.

**Methods:** This study was a retrospective one done at Diphu Medical College and Hospital. Data was collected from three consecutive years from the available hospital records. Ethical approval was obtained from the Institutional Ethical Committee (Reference number- DMCH/EC/2022/105/2828). Results obtained were entered in MS Excel and analyzed in the light of available literatures.

**Results:** Among 8569 newborns, congenital anomalies were found to be present in 62 cases with a prevalence rate of 7.2 per 1000 live birth. The commonest birth defect was found to be cleft lip and cleft palate followed by the musculoskeletal system.

**Conclusion:** Congenital anomaly plays an important role in the mortality and morbidity of neonates and children. Hence proper antenatal care and counseling will help in reducing the burden of congenital anomalies.

**Keywords:** Prevalence, Congenital anomaly, Cleft lip, Cleft palate, Down's syndrome.

This is an Open Access article that uses a funding model which does not charge readers or their institutions for access and distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/4.0>) and the Budapest Open Access Initiative (<http://www.budapestopenaccessinitiative.org/read>), which permit unrestricted use, distribution, and reproduction in any medium, provided original work is properly credited.

**Introduction**

World Health Organization (WHO), defines congenital anomaly as structural or functional defects that occur during intrauterine life.[1] Congenital anomaly is the one of the major cause of total global burden of disease.[1] Every year an estimated of 2,40,000 newborn lose their lives within the first 28 days of life. Apart from the newborn deaths, congenital anomalies claim further 170,000 deaths in children between 1 month to 5 years of life.[1] The Global Burden of Disease study 2015 also identified congenital anomalies as fifth main causes of mortality in under five children and comprises 11% of the neonatal mortality.[2] Globally, prevalence of congenital anomaly is about 2-3% [3]. The burden of congenital anomalies varies with socio-economic status, nutrition, ethnicity, alcohol intake and advanced maternal age.[2,4] Still birth was observed to be a risk factor with advanced maternal age.[4] Consanguinity of marriage and exposure to certain drugs are also seen to cause congenital malformation. [5] Studies shows that 30-40% of congenital anomalies may occur due to genetic

causes, 5-10% due to environmental causes and idiopathic causes are observed in 50% of the cases[6]. Congenital anomaly may occur as an isolated(i. e single defect) or as a group of abnormalities (i.e. multiple defects) like abnormalities of Vertebrae, Anal atresia, Cardiac defects, Tracheoesophageal fistula, and/or Eosophageal atresia, Renal, Radial anomalies and Limb defects (VACTRAL).[7] Anomalies which causes a threat to an infant's health, life expectancy, physical or social life may be described as "major anomalies" and those which has less or no impact on health or long-term or short-term function as "minor anomaly".[7] Early diagnosis of diseases and timely counseling of parents helps in early intervention, thus reducing the mortality and morbidity of the neonates.[5]

**Materials and Methods**

This study was done as a retrospective one at Diphu Medical College and Hospital, located in Karbi Anglong, a hill district of Assam. Study period was between 1/5/2020 to 30/4/2023. Ethical

clearance was obtained from the Institutional Ethical Committee.

The cases were selected based on the following criteria:

**Inclusion Criteria:**

1. All live newborns delivered at Diphu Medical College and Hospital whose gestational age was more than 24 weeks and weight more than 500gms.
2. All neonates admitted at the SNCU (Special Care Newborn Unit), of Diphu Medical College and Hospital in outborn unit.

**Exclusion Criteria:** All still born babies delivered at Diphu Medical College and Hospital.

Data was collected from the available hospital records. The variables included in the study were gestational age and weight of the babies, maternal factors included maternal age, intake of alcohol, gravida and consanguinity. Babies with gestational age more than 37 weeks and weight more than 2.5 kgs were considered as term. Those below 37 weeks and weight less than 2.5kgs were considered

as preterm. Mothers who conceived for the first time was considered as primigravida and those who conceived twice or more, multigravida. Consanguinity is defined as marriage between male and female who are blood related, e.g., between brother and sister or between 1<sup>st</sup> cousins.[23] The cases were diagnosed based on the clinical, radiological and laboratory investigation wherever indicated. Data was statistically analyzed using the MS Excel.

**Results**

This study included 8569 live births as the total study population. Out of the total cases, 7867 babies were delivered at Diphu Medical College & Hospital (DMCH), and 702 were out-born cases. A total of 62 cases were diagnosed with congenital anomalies and the prevalence rate was found to be 7.2% per 1000 live births.

Males (n=35, 56.45%) were affected more than the females (n=27;43.54%).

Number of term babies (n=49, 79.03%) outnumbered pre-term babies (n=13, 20.96%). (Table 1)

**Table 1: Demography of the newborns**

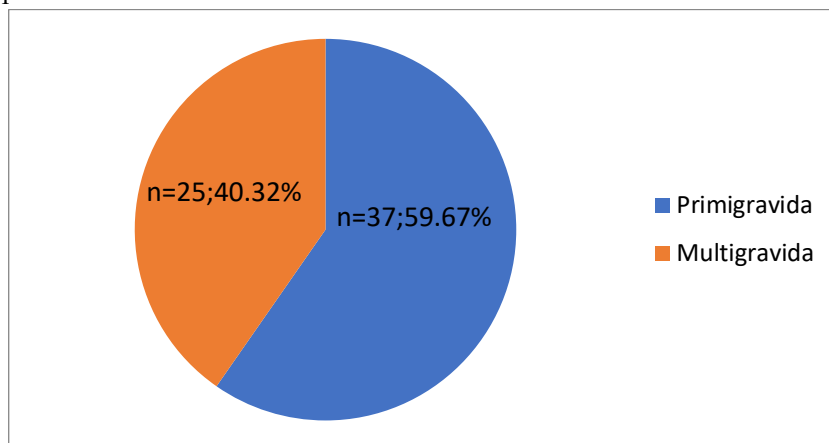
Gender		Gestational Age	
Males (n;%)	Females (n;%)	Term (n;%)	Preterm (n;%)
35; 56.45%	27; 43.54%	49; 79.03%	13; 20.96%
Total = 62 (100%)		Total = 62 (100%)	

Majority of the mothers were between the age group of 20.1 – 25 years (n=32; 51.6%) followed by 25.1–30(n=21; 33.95) years, above 30(n=05; 08.1%) years and less than 20 years(n=04;6.5%) (Table 2). Minimum and maximum age of mothers was respectively 18 and 42 years respectively. Mean age was 25.08 years and SD was found to be 4.846

**Table 2: Age group distribution of mothers of anomalous child**

Age group of mothers	No. of mothers (n)	Percentage (%)
Less than 20 yrs	04	6.45%
20.1-25 yrs	32	51.61%
25.1-30 yrs	21	33.87%
Above 30.1 yrs	05	08.06%
Total	62	100%

In this study most of the mothers were primigravida (n=37; 59.67%), followed by multigravida (n=25; 40.32%).Figure:1



**Figure 1: Percentage of Primigravida and Multigravida**

Alcohol consumption was seen in 32(51.61%) mothers whereas 30(48.38%) mothers did not consume alcohol. Out of 62, only 2, (3.22%) mothers reported with positive history of consanguinity.

More than one quarter of the newborns had multiple anomaly of cleft lip and cleft palate (n=16;

25.80%), followed by musculoskeletal system (n=12; 19.35%). The other system involved were GIT (n=7;11.29%), CNS (n=6;9.67%), CVS (n=5;8.06%), Eye & Ear anomaly (n=4;6.45%), Other congenital malformation(n=3;4.83%), Genitourinary (n=2;3.22%) and Chromosomal abnormalities(n=1;1.61%) (Table 3)

**Table 3: System involved and the type of congenital anomaly in each system**

System	Type of anomaly	n(%)
Cleft lip and cleft palate	1) Cleft lip and palate	16(25.80%)
	2) Cleft lip	03(4.83%)
	3) Cleft palate	03(4.83%)
Musculo-skeletal	1) Congenital telipes	04(6.45%)
	2) Phocomelia	01(1.61%)
	3) Congenital dislocation of hip	02(3.22%)
	4) Polydactyly	02(3.22%)
	5) Arthrogryposis	01(1.61%)
	6) Absent right arm	01(1.61%)
	7) Syndactyly	01(1.61%)
Gastro-intestinal Tract(GIT)	1) Tracheo-oesophageal fistula	03(4.83%)
	2) Congenital diaphragmatic hernia	02(3.22%)
	3) Imperforate anus	02(3.22%)
Central nervous System(CNS)	1) Meningomyelocele	03(4.83%)
	2) Hydrocephalous	02(3.22%)
	3) Anencephaly	01(1.61%)
Cardiovascular system(CVS)	1) Congenital heart defect	05(8.06%)
Ear, eye	1) Anophthalmos	01(1.61%)
	2) Microtia	01(1.61%)
	3) Accessory tragus	01(1.61%)
	4) Crumpled ear	01(1.61%)
Other congenital Malformation	1) Port-wine stain	01(1.61%)
	2) Ichthyosis	01(1.61%)
	3) Thrombocytopenia with absent radius(TAR)	01 (1.61%)
Genito-urinary	1) Undecented testis(unilateral)	01(1.61%)
	2) Hydrocele	01(1.61%)
Chromosomal abnormalities	1) Down's Syndrome	01(1.61%)

### Discussion

In our study, the prevalence rate was found to be 7.2%. Almost similar results of 6.1% and 6.3% were noticed in the studies done by other authors.[3,8] EUROCAT, the European Surveillance of Congenital Anomalies, showed higher rate of prevalence of 23.9 % per 1000 per live birth[9]. Studies by other authors showed lower rate of 1.2% and 1.7% respectively.[10,11] The difference in prevalence rate shown in different studies may be explained due to various reasons like different sampling methods, study methods, geographical variations, definitions and statistical calculation methods. The number of males were more than the females in this study, similar findings were seen in.[6,10,15] Term babies outnumbered preterm babies in our study which was similar to.[6] Type of congenital anomaly found in our study was cleft lip and cleft palate followed by involvement of the musculo-skeletal system. The type of congenital heart disease could

not be specified as it was a retrospective study and the centre was not equipped with echocardiography facility at the time of data collection. Hence the cases were diagnosed based on physical, auscultatory and radiological findings. The commonest system involved in studies done by different authors are CHD[3,8], CNS[12,13]. Involvement of musculoskeletal system was seen mostly in[10,14,15] and urinary system.[16] These differences may be due to the various etiological factors and sampling methods. The present study showed that a higher rate of congenital anomalies in primigravida and mothers between the age group between 20.1 to 25 years, which was almost similar to the findings of. [17] Congenital malformation was found to be associated more in multigravida.[18] and in mothers of older age group (>35yrs) in .[18,19,20] The variation in the results may be due to small sample size and sampling methods. Our study did not show any relation between intake of alcohol by mother with

congenital anomalies in the offspring. Results were similar to that of other authors.[21,22]. This could be explained by the fact that the amount of alcohol intake during pregnancy is not known as the study was a retrospective one and the sample size maybe small to comment on. Consanguinity was found in only n=2 (3.3%) cases and the number was too small to comment on, but studies done by other authors proved that there were significantly more congenital anomalies in newborns with history of consanguinity in parents.[15,18,23]

### Conclusion

The prevalence rate of Congenital anomaly was found to be 7.2% per 1000 live birth and proper antenatal care with counseling will definitely help to reduce the morbidity and mortality in newborns and children.

**Acknowledgement:** We would like to thank the data entry operator Mr. Denin Tisso, sister-in-charge (SNCU), Ms. Phoibi Inbuon and all the staff of SNCU for extending their help in the collection of data.

**Limitations:** This study was a retrospective one and the institution was not fully equipped with all the equipments during the period of data collection, hence few cases were selected based on clinical, laboratory and available radiological investigations. Still births could not be included in the present study as we were unable to retrieve all the data due to which the actual prevalence rate may not be reflected.

### References

1. WHO Congenital disorders. Geneva: WHO, 2023. Available: <https://www.who.int/news-room/fact-sheets/detail/birth-defects>
2. Kumar J, Saini SS, Sundaram V, Mukhopadhyay K, Dutta S, Kakkar N, Kumar P. Prevalence & spectrum of congenital anomalies at a tertiary care centre in north India over 20 years (1998-2017). *Indian J Med Res.* 2021 Mar; 154(3):483-490.
3. Chimah OU, Emeagui KN, Ajaegbu OC, Anazor CV, Ossai CA, Fagbemi AJ, Emeagui OD. Congenital malformations: Prevalence and characteristics of newborns admitted into Federal Medical Center, Asaba. *Health Sci Rep.* 2022 Apr 13; 5(3):e599.
4. Lean SC, Derricott H, Jones RL, Heazell AEP. Advanced maternal age and adverse pregnancy outcomes: A systematic review and meta-analysis. *PLoS One.* 2017 Oct 17; 12(10):e0186287.
5. Siddika M, Sen S, Islam MN, Bhuiyan MK. Pattern and Risk Factors of Congenital Anomaly in Newborn in a Tertiary Level Private Medical College Hospital, Bangladesh. *My-mensingh Med J.* 2018 Oct; 27(4):805-812.
6. Shrestha S, Shrestha A. Prevalence of Congenital Malformations among Babies Delivered at a Tertiary Care Hospital. *JNMA J Nepal Med Assoc.* 2020 May 30; 58(225):310-313.
7. DeSilva M, Munoz FM, Mcmillan M, Kawai AT, Marshall H, Macartney KK, Joshi J, Oneko M, Rose AE, Dolk H, Trotta F, Spiegel H, Tomczyk S, Shrestha A, Kochhar S, Kharbanda EO; Brighton Collaboration Congenital Anomalies Working Group. Congenital anomalies: Case definition and guidelines for data collection, analysis, and presentation of immunization safety data. *Vaccine.* 2016 Dec 1; 34(49):6015-6026.
8. Ajao AE, Adeoye IA. Prevalence, risk factors and outcome of congenital anomalies among neonatal admissions in OGBOMOSO, Nigeria. *BMC Pediatr.* 2019 Apr 3; 19(1):88.
9. Dolk H, Loane M, Garne E. The prevalence of congenital anomalies in Europe. *Adv Exp Med Biol.* 2010; 686:349-364.
10. Baruah J, Kusre G, Bora R. Pattern of Gross Congenital Malformations in a Tertiary Referral Hospital in Northeast India. *Indian J Pediatr.* 2015 Oct; 82(10):917-22.
11. Osanyin GE, Odeseye AK, Okojie OO, Akinajo OR, Okusanya BO. Fetal Congenital Anomaly in Tertiary Hospital in Lagos, South-West Nigeria: A Review of Presentation and its Outcome. *West Afr J Med.* 2019 Jan-Apr; 36(1):25-28.
12. Fajolu IB, Ezenwa B, Akintan P, Ezeaka A. 8 years review of major congenital abnormalities in a tertiary hospital in lagos, Nigeria. *Niger J Paediatr.* 2016; 43:175-1.
13. Mashuda F, Zuechner A, Chalya PL, Kidenya BR, Manyama M. Pattern and factors associated with congenital anomalies among young infants admitted at Bugando medical centre, Mwanza, Tanzania. *BMC Res Notes.* 2014 Mar 29; 7:195.
14. Bhide P, Kar A. A national estimate of the birth prevalence of congenital anomalies in India: systematic review and meta-analysis. *BMC Pediatr.* 2018 May 25; 18(1):175.
15. Congenital anomalies in neonates and associated risk factors in a tertiary care hospital: a single center study from India. Seba B, Shubhankar M, Sambhedana P. *Indian J Appl Res.* 2017;7:174-176.
16. Prevalence of congenital anomalies in a tertiary care centre in North Kerala, India. Jayasree S, D'Couth S. *Int J Reprod Contracept Obstet Gynecol.* 2018; 7:864-870.
17. Christe DM, Mohana D, Shobha S. Major congenital malformations of foetus: a bane in pregnancy. *Int J Reprod Contracept Obstet Gynecol* 2020; 9:4521-5.

18. Patel PK. Profile of major congenital anomalies in the Dhahira region, Oman. *Ann Saudi Med.* 2007 Mar-Apr; 27(2):106-11.
19. Suwatanaviroj A, Ratrisawadi V. Factors associated with congenital malformations in Thailand. *J Med Assoc Thai.* 1996 Sep; 79(9):545-9.
20. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. *Indian Pediatr.* 1994 Oct; 31(10):1187-91.
21. Kurita H, Motoki N, Inaba Y, Misawa Y, Ohira S, Kanai M, Tsukahara T, Nomiya T; Japan Environment and Children's Study (JECS) Group. Maternal alcohol consumption and risk of offspring with congenital malformation: the Japan Environment and Children & Study. *Pediatr Res.* 2021 Aug; 90(2):479-486.
22. Moges N, Anley DT, Zemene MA, et al. Congenital anomalies and risk factors in Africa: a systematic review and meta-analysis. *BMJ Paediatrics Open* 2023;7:e002022.
23. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital-based study. *Pediatric reports.* 2013 Feb; 5(1):e5.