



PREVALENCE OF FETAL CONGENITAL ANOMALIES AMONG PREGNANT WOMEN IN TERTIARY CARE HOSPITAL

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ABSTRACT **Background:** Congenital anomalies also commonly referred as birth defects, congenital disorders, congenital malformations or congenital abnormalities are conditions of prenatal origin that are present at birth, potentially impacting an infant health, development or survival. An estimated 24000 newborns die worldwide within 28th day of birth every year due to birth defects and 170000 deaths of children between ages of 1 month and 5 years and contribute to long term disability which later significantly depends on individual facilities, health care systems and societies. The proportion of under 5 deaths are increasing due to congenital anomalies as other causes of under 5 deaths are controlled (10). Fetal anomaly scan is the most effective measure of reducing the prevalence of serious congenital abnormalities and increasing the survival rate of those born with these issues. Knowledge about the prevalence of congenital anomalies is useful to obtain baseline rate and identifying clues to the etiology of conditions which help to plan and access antenatal screening for congenital anomalies especially high-risk population. **Aim and Objectives:** To analyze the prevalence of major and minor congenital anomalies in pregnant women attending the tertiary care hospital. **Methods:** This is a retrospective study done in the department of obstetrics and gynecology at Siddhartha Medical College, Vijayawada, India between January 2019 to December 2021 (Three years). All antenatal mothers attending the labor room and outpatient department of Obstetrics & Gynecology were included in the study and subjected to TIFFA Scans. All antenatal women who gave consent for the study were included. The study was started after obtaining the ethical committee approval from our institute. International statistical classification of diseases and related health problems, 10th revision (ICD-10) was used for uniformity and international comparison. The further subcategorization was done as per the WHO birth defects surveillance manual. The prevalence of individual as well as overall congenital anomalies was calculated. **Results:** A total of 12488 deliveries were done in our tertiary care hospital. Out of which 86 congenital anomalies were detected with a prevalence of 0.7 %, among which, CNS anomalies were the most common. **Conclusion:** Prenatal diagnosis of congenital anomalies provides information for proper decisions during pregnancy, fetal intervention, effective parental counselling and appropriate perinatal treatment, thereby improving perinatal outcomes.

KEYWORDS : Congenital anomalies, CNS anomalies, Prenatal diagnosis, TIFFA Scan, Parental Counselling

INTRODUCTION

Congenital anomalies are major cause of stillbirths and neonatal mortality in India, also pose a health care burden to the community. World Health Organization (WHO) defined congenital anomalies as structural or functional anomalies, including metabolic disorders which are present at the time of birth [1,2]. Major congenital anomalies occur in 2 to 3 % of human births and are an important cause of neonatal mortality and morbidity.

Though the reported incidence of congenital anomalies at birth is 2-3%, actual incidence is higher due to high number of abortions that remain unknown. Though folic acid and zinc deficiency has been proved to cause Neural tube defects, the incidence has significantly decreased due to peri-conceptional folic acid intake as implemented by National health mission guidelines [3]. According to global burden of disease study congenital anomalies are 1/5 th leading cause of under 5 mortality and they were associated with 11% of neonatal deaths[4]. Maternal anemia, Diabetes mellitus, exposure to drugs and radiation are various other risk factors attributed for causing congenital anomalies. Antenatal congenital anomalies contribute to fetal loss, still births, preterm births, neonatal mortality, and childhood morbidity with significant outcome on the mothers and their families [5,6]. Data on congenital anomalies from developing countries like India are limited and the magnitude, severity of various congenital anomalies vary with various geographical locations. Hence, we conducted this study to find the true prevalence and outcome of congenital anomalies in tertiary care center.

Methods

This was a retrospective cross sectional hospital record-based study between January 2019 to December 2021(3 years) in department of Obstetrics and gynecology SMC Vijayawada and in collaboration with Radiology departments. All the antenatal ultrasonograms were performed by Radiologist trained in fetal medicine using "ESOAT MYLAB X-6 Machine Probe Curvilinear". All the antenatal mothers attending the Obstetrics out-patient department and labor room gave

consent for the study were recruited, subjected to 1st & 2nd trimester anomaly scans. The annual delivery rate is 4000 to 4500 most of which are high risk pregnancies. All the live births at our facility from January 2019 to December 2021 were enrolled in our study. All the new born with congenital anomalies admitted in the new born unit during this period were included. Systematic clinical examination was done at birth, postnatal day 1 and pre discharge. Congenital anomalies were classified in to major and minor as per the WHO birth defects surveillance manual. Major anomaly is defined as structural changes that have significant medical, social or cosmetic consequences for the effected individual and typically require medical intervention . Anomalies were assigned international statistical classification of diseases and related health problems, 10th revision (ICD-10) codes to facilitate system wise classification of anomalies [7]. Newborns with congenital anomalies delivered in our hospital were taken in to data. The study was conducted after obtaining the ethical committee approval from our institute. Maternal parameters like age, parity, gestational age was recorded. The diagnosed anomalies were recorded in the proforma. The data was collected and processed using Microsoft Excel and descriptive statistical analysis was done.

Inclusion Criteria

a)All Antenatal women attending the Obstetrics and Gynecology outpatient department and labor room.

Exclusion Criteria

a)Antenatal women with multiple gestation.
b)Antenatal women with refusal of consent.

RESULTS

prevalence of congenital malformations

Out of total 12488 singleton pregnancies screened; 86 women were diagnosed to have congenital anomalies. Majority, i.e., 46 out of 86(53.4%) patients were multigravida. Incidence of congenital anomalies were higher among maternal age group of 22-25 years in our study. About 32 anomaly fetuses were seen in women age group 22-25

(37.2%). Antenatal mothers between 26-30 years had 29(33.7%). Around 7 congenital anomalies were noted in 31-35 (8.1%) years age group. Around 14(16.2%) congenital anomalies were noted in teenage pregnant mothers of 18-21 years. Around 4 congenital anomalies noted in >35 years age group. The maternal age wise distribution of congenital anomalies in the study group is shown in Table 1.

Table 1: The Maternal Age Wise Distribution Of Congenital Anomalies

Parameters	No of Deliveries (N= 12488), n(%)	No of Congenital Anomalies(N=78), n(%)	Percent age (%)
Maternal age (in years)	18 – 21	3459(27.7)	14(0.4)
	22 – 25	3708(29.7)	32(0.8)
	26 – 30	2884(23.1)	29(1)
	31 – 35	1486(11.9)	7(0.4)
	>35	924(7.4)	4(0.4)

The parity wise distribution of congenital anomalies in the study group is shown in Table 2

Table 2: The Parity Wise Distribution Of Congenital Anomalies

Parameters	No of Deliveries (N= 12488), n (%)	No of Congenital Anomalies(N=78), n(%)	Percentage (%)
Parity	Primi	5931(47.5)	40(0.7)
	Multipara	6557(52.5)	46(0.7)

Gestational age wise incidence of congenital anomalies in antenatal mothers

21 out of 86 cases (24.4%) of antenatal congenital anomalies were diagnosed between 16-20 weeks gestational age. Congenital anomalies in gestational age of 21-25 weeks were 27 cases (31.3%), 26-30 weeks was 23(26.7%) and >30 weeks was 15(17.4%)(Table 2).

Table 3: Gestational Age Wise Incidence Of Congenital Anomalies In Antenatal Mothers.

Gestational age (in weeks)	Total Number of Deliveries (N=12488) (n=%)	Congenital Anomalies (N=86) (n = %)	Percentage (%)
16-20	3496(28)	21(0.6)	24.4
21-25	5544(44.4)	27(0.4)	31.3
26-30	2585(20.7)	23(0.8)	26.7
>30	861(6.9)	15(0.2)	17.4

System Wise Incidence Of Congenital Anomalies.

According to the International statistical classification of disease and related health problems (7) we classified the patterns of congenital anomalies into

1. Congenital malformations of nervous system.
2. Congenital malformations of musculoskeletal system
3. Congenital malformations of Digestive system
4. Congenital malformations of circulatory system
5. Congenital malformations of genitourinary system
6. Congenital malformations of eye, ear, face and neck system

In the present study, anomalies involving the Nervous system were 40(46.5%),Renal system were 12 (14%),Digestive system was 11(12.7%), Circulatory system were 9(10.4%), musculoskeletal system were 8(9.3%) and Eye ,ear, face and neck were 6(6.9%). Anencephaly is the most common seen in 16 cases out of 40 cases involving nervous system. (Table 4), (Figure 1).



Figure 1: Anencephaly



Figure 2 : Achondroplasia

Table IV

S. NO	Systems	Congenital Anomalies (N=86)	Total (N=86) (n=0.7%)
1	CNS	Anencephaly (16)	40(46.5 %)
		Hydrocephalus (5)	
		Encephalocele+ Meningocele (6)	
		Spina bifida (3)	
		Arnold Chiari Type 4 (2)	
		Holoprosencephaly (2)	
		Dandi walkers Syndrome (2)	
		Arachnoid hematoma (1)	
		Ventriculomegaly (1)	
		Choroid cyst (1)	
2	GIT	Omphalocele (3)	11(12.7%)
		Anal atresia (3)	
		Gastroschisis (2)	
		Diaphragmatic hernia (2)	
		Bowel Dilatation (1)	
3	Musculoskeletal	Club foot (3)	13(16.6%)
		Skeletal Dysplasia (1)	
		Achondroplasia (1)	
		Absent sternum (1)	
		Limb reduction defects (1)	
		Un ossified nasal bone (1)	
4	Genitourinary system	Renal agenesis (1)	12(14%)
		Bilateral ectopic kidney (1)	
		Ambiguous genitalia (2)	
		Distal urethral obstruction (Hydronephrosis) (2)	
		Polycystic kidney disease (2)	
		PUV (1)	
		Multicyclic dysplastic kidneys (3)	
5	Circulatory System	Acynotic congenital heart diseases (4)	9(10.4%)
		Dextrocardia (1)	
		Pulmonary artery narrowing (1)	
		Truncus arteriosus (1)	
		Cystic Hygroma (2)	
6	Eye Ear Face and Neck	Cleft lip and palate (6)	6(6.9%)

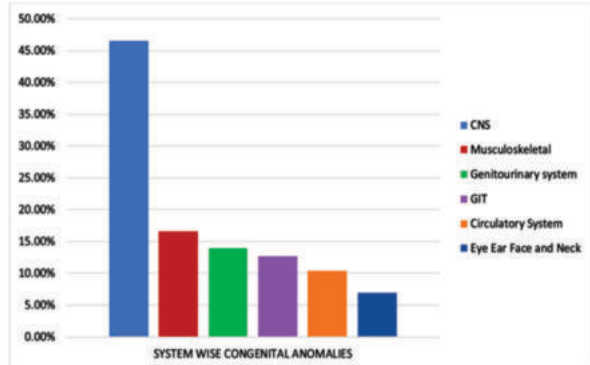
The individual organ system wise distribution ICD – 10 subclassification of clinically important major anomalies is shown in Table V

System-wise major anomalies as per International Classification of

Diseases, tenth revision classification (ICD10)

Table V

System	n (% of individual system)
Nervous system (Q00-Q07) (n=189)	
Anencephaly	16 (8.5)
Encephalocele	14 (7.4)
Microcephaly	10 (5.3)
Congenital hydrocephalus	63 (33.3)
Spina bifida	80 (42.3)
Arnold-Chiari malformation	3 (1.6)
Circulatory system (Q20-Q28) (n=621)	
Congenital malformations of cardiac chambers and connections	56 (9.0)
Congenital malformations of cardiac septa	225 (36.2)
Congenital malformations of pulmonary and tricuspid valves	54 (8.7)
Congenital malformations of aortic and mitral valves	39 (6.3)
Congenital malformation of great arteries	189 (30.4)
Respiratory system (Q30-Q34) (n=200)	
Congenital malformations of nose	6 (3.0)
Congenital malformations of larynx	7 (3.5)
Congenital malformations of trachea and bronchus	15 (7.5)
Congenital malformations of lung	164 (82.0)
Cleft lip and cleft palate (Q35-Q37) (n=37)	
Cleft palate	5 (13.5)
Cleft lip	3 (8.1)
Cleft palate with cleft lip	29 (78.4)
Digestive system (Q38-Q45) (n=144)	
Congenital malformations of oesophagus	41 (28.5)
Congenital absence, atresia and stenosis of small intestine	37 (25.7)
Congenital absence, atresia and stenosis of large intestine	39 (27.1)
Genital organs (Q50-Q56) (n=80)	
Ambiguous genitalia	44 (55.0)
Hypospadias	22 (27.5)
Urinary system (Q60-Q64) (n=316)	
Renal agenesis and other reduction defects of kidney	66 (20.9)
Cystic kidney disease	107 (33.5)
Congenital obstructive defects of renal pelvis and ureter	145 (45.9)
Musculoskeletal system (Q65-Q79) (n=412)	
Congenital deformities of hip	3 (0.7)
Congenital deformities of feet	60 (14.5)
Congenital diaphragmatic hernia	110 (26.7)



DISCUSSION

The prevalence of congenital anomalies in the present study is 0.7% comparable with the observations in Alkananda et.al [8] and it is lower than Alia [9]. The variation could be due to social factors, ethnic factors, nutritional status and geographical areas. The present study shows elderly maternal age group (> 30 years) and multipara have high risk for congenital anomalies as comparable with Singh A [10]. The most common system involved is nervous system (46.5%) followed by genitourinary system (14%). The study involves identification of

anomalies diagnosed antenatally and clinically at birth, postnatally, intervention and counselling and psychological support. Moreover, most of the Centre based studies did not used any standard protocol for classification of anomalies which might influence the reporting of system wise anomalies. Bhid et.al [11] from pune using the ICD 10 classification found results similar to our study. At our center congenital anomaly was the 3rd or 4th most common cause of death similar to the global pattern for developing countries [12,13]. Increased awareness and availability of antenatal TIFA scan leading to an increased antenatal diagnosis of CNS anomalies (Anencephaly, Encephalocele) are amenable to diagnosis and can proceed for MTP. Similarly, NTDs are well known preventable defects and account for 1/3 of neonatal deaths and significant morbidity [14]. The strength of the study was a large sample size, sufficient long duration to observe trends and use of standard methodology for classification and reporting to make it comparable to other national and international studies.

Limitations were due to retrospective study i.e., lack of recording of risk factors.

CONCLUSION

In this study prevalence of fetal congenital anomaly was found to be.% CNS defect - Anencephaly was found to be the commonest form of anomaly in our study population. Congenital anomalies responsible for significant mortality and morbidity warranting the need for national surveillance program and birth defect services which has several important implications on national health system. There is a need for integral package involving diagnosis, surgical/Medical intervention, financial support, counselling and psychological support along with follow up services, including rehabilitation. A significant proportion of birth defects is preventable or correctable warranting the need for sensitization regarding antenatal measures and postnatal corrective surgeries for healthy disability adjusted life years.

Prevention

Primary prevention can be achieved with basic reproductive health approaches which include family welfare services, promoting healthy dietary habits and lifestyle, safe food and environment, detecting, treating and preventing maternal infections, control of diseases like IDDM and epilepsy, vaccination, avoiding use of certain drugs during pregnancy and prior to conception. Preconceptionally use of folic acid supplements prevents neural tube defects.

Secondary prevention aims to reduce the number of children born with birth defects. With the use of medical genetic screening and prenatal diagnosis, birth defects are detected and the couple offered genetic counselling and therapeutic options.

Tertiary prevention is directed towards early detection and management of problem once a child with birth defects is born.

Ministry of health and family welfare (MoHFW), govt of India has made a provision for prevention, early diagnosis and management of birth defects under INAP 2014 India newborn action plan, along with basic mother and child care.

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Conflict of Interest

None.

Ethical Approval

The study was approved by the Institutional Ethics Committee

REFERENCES

- World Health Organization. Congenital anomalies. [accessed on March 23, 2018]. Available from: <http://www.who.int/mediacentre/factsheets/fs370/en/>
- Shi H, Enriquez A, Rapadas M, Martin EMMA, Wang R, Moreau J, et al. NAD deficiency, congenital malformations, and niacin supplementation. *N Engl J Med.* 2017;377:544-52. [PubMed][Google Scholar]
- Boyle B, Addor MC, Arriola L, Barisic I, Bianchi F, Csáky-Szunyogh M, et al. Estimating Global Burden of Disease due to congenital anomaly: An analysis of European data. *Arch Dis Child Fetal Neonatal Ed.* 2018;103:F22-8. [PMC free article] [PubMed][Google Scholar]
- GBD 2015 child mortality collaborators. Global, regional, national, and selected subnational levels of stillbirths, neonatal, infant, and under-5 mortality, 1980-2015: a systematic analysis for the global burden of disease study 2015. *Lancet.* 2016;388:1725-74. [PMC free article] [PubMed][Google Scholar]
- Hug L, Alexander M, You D, Alkema L. UN Inter-agency Group for Child Mortality Estimation. National, regional, and global levels and trends in neonatal mortality

- between 1990 and 2017, with scenario-based projections to 2030: a systematic analysis. *Lancet Glob Health*. 2019;7:e710–20. [PMC free article][PubMed][Google Scholar]
6. World Health Organization. Birth defects. [accessed on April 5, 2018]. Available from: http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf.
 7. GBD 2015 child mortality collaborators. Global, regional, national, and selected subnational levels of stillbirths, neonatal, infant, and under-5 mortality, 1980-2015: a systematic analysis for the global burden of disease study 2015. *Lancet*. 2016; 388:1725–74
 8. Alakananda, Choudhury SS, Neiting C (2005) A study on prevalence of congenital anomalies of foetuses among pregnant women in a tertiary care hospital and its association with socio-demographic factors. *The New Indian Journal of OBGYN* 2(1): 4650.
 9. Alia N, Ahmed I (2010) Congenital anomalies: prevalence of abnormalities in 2nd trimester of pregnancy in Madina teaching hospital, Faisalabad on grey scale ultrasound *JUMDC* 1(1): 23-28.
 10. Singh A, Ravinder K, Jammu S (2009) Pattern of Congenital Anomalies in Newborn: A Hospital Based Prospective, *Jammu (J&K)-India* 11: 346.
 11. Bhide P, Gund P, Kar A. Prevalence of congenital anomalies in an Indian maternal cohort: Healthcare, prevention, and surveillance implications. *PLoS One*. 2016;11: e0166408.
 12. Boyle B, Addor MC, Arriola L, Barisic I, Bianchi F, Csáky-Szunyogh M, et al. Estimating Global Burden of Disease due to congenital anomaly: An analysis of European data. *Arch Dis Child Fetal Neonatal Ed*. 2018;103:F22–8.
 13. Hug L, Alexander M, You D, Alkema L. UN Inter-agency Group for Child Mortality Estimation. National, regional, and global levels and trends in neonatal mortality between 1990 and 2017, with scenario-based projections to 2030: a systematic analysis. *Lancet Glob Health*. 2019;7: e710–20.
 14. Zaganjor I, Sekkarie A, Tsang BL, Williams J, Razzaghi H, Mulinare J, et al. Describing the prevalence of neural tube defects worldwide: A systematic literature review. *PLoS One*. 2016;11: e0151586.