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Research Article

A CROSS-SECTIONAL STUDY OF PREVALENCE OF CONGENITAL ABNORMALITIES ON ROUTINE ULTRASOUND SCAN OF SECOND AND THIRD TRIMESTER PREGNANCY

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Abstract

Introduction: Birth defects are a major contributor to perinatal and infant mortality, morbidity, and lifelong disability worldwide. Birth defects affect at least 3% of babies in most populations. The anomalies are the leading cause of neonatal morbidity & mortality. Defective embryogenesis or intrinsic abnormalities in the process of development result congenital anomalies. About 3,03,000 newborns die within 4 weeks of birth every year worldwide due to congenital anomalies.

Materials and methods: This is a cross-sectional study carried out in the Department of Radiodiagnosis, Dr Chandramma Dayanand Sagar Institute of Medical Education and Research, Harohalli, Kanakpura road, Karnataka, India. Pregnant women of 2nd and 3rd trimester, referred to 'Radiology' department from the outpatient and inpatient departments of 'Obstetrics and Gynecology' of the same hospital between October 2024 to September 2025 are included in the study. The Radiologist performed all the transabdominal ultrasonography on a 'Siemens Acuson X300' machine using 'CH 5-2' probe after taking verbal consent from the patient. A questionnaire was used containing the following information e.g. women's age, parity, gravidity, date of last menstrual period. The questionnaire form also included about the result of transabdominal USG

examination which included the following: singleton or multiple, dead or alive foetus, gestational age, and presence or absence of congenital anomalies.

Results: During the study period of 2024 October to September 2025, a total of 5300 numbers of 2nd and 3rd trimesters prenatal USG were done. 90 numbers of congenital anomalies were detected among the pregnant women scanned. So, the antenatal prevalence of congenital anomalies was 1.73%. Of the women having congenital abnormalities in the foetus, the majority were between 20-35 years old (73.33%) followed by those above 35 years (15.56%). The mean maternal age at the diagnosis was 25.5 years (SD ± 6.15). Women who had primary schooling represented 53.33%, with 26.67% of women were illiterate. Majority women were nullipara (42.22%) followed by primipara (31.11%). 34 cases (37.78%) were detected at 3rd trimester and majority at 2nd trimester ie. 56 cases (62.22%). The mean gestational age was 27 weeks (SD±6.42).

Conclusion: In this study prevalence of fetal congenital anomaly was found to be 1.73%. CNS defect was found to be the commonest form of anomaly in our study population. Creating awareness regarding regular ANC and importance of anomaly scan on time can help in primary prevention of disability and reducing perinatal mortality and morbidity.

Key Words: Birth defects, CNS defect, Defective embryogenesis, transabdominal ultrasonography.

INTRODUCTION

Birth defects are a major contributor to perinatal and infant mortality, morbidity, and lifelong disability worldwide. Birth defects affect at least 3% of babies in most populations. The anomalies are the leading cause of neonatal morbidity & mortality. Defective embryogenesis or intrinsic abnormalities in the process of development result congenital anomalies. About 3,03,000 newborns die within 4 weeks of birth every year worldwide due to congenital anomalies.[1]

All pregnancies are at a risk of producing congenital malformations, though only some of them are at a greater risk. There is a need for routine and thorough screening for foetal congenital anomalies. The priority goal in screening is the early detection of major foetal anomalies. The National Health Service (NHS) Foetal Anomaly Screening Program guidance recommends screening for conditions with detection rates above 50% at this scan, including anencephaly, open spina bifida and gastroschisis.[2] 'Defined ultrasound findings of uncertain significance' or 'normal variants' (referred to as 'markers') are also identified at this scan. These include echogenic bowel (EB), renal pelvicalyceal dilatation (PCD) and cardiac echogenic foci (CEF). Associations between markers and adverse pregnancy outcomes including intrauterine fetal death, chromosomal abnormalities and cystic fibrosis have been reported.[3]

Congenital anomalies may present as isolated abnormalities or part of a syndrome. The most common, severe anomalies include congenital heart defects, cleft lip with or without cleft palate, Down syndrome & neural tube defects. Laboratory test and imaging studies are available for

detection of these anomalies. Ultrasound examination is one of the most important diagnostic tools which give a great amount of information about the anatomical structure as well as some physiological aspect of the status of the foetus. The current ACR (American College of Radiology)/ AIUM (American Institute of Ultrasound in Medicine) guidelines for the performance of the second & third trimester obstetrics examination describe the standard sonographic examination.[4]

In India particularly in south region where the social support system is poor, bringing up a child with mental or physical handicap is a major burden for the parents and family. In cases where primary prevention does not possible, prenatal diagnosis by ultrasound scan provides the next best alternative. In cases where a major structural defect is identified, termination of pregnancy can be offered. The purpose of this study is to evaluate the antenatal prevalence of major congenital anomalies and malformation patterns in our hospital population of Dr Chandramma Dayanand sagar Institute of Medical Education and Research, Harohalli, Kanakpura road, Karnataka, India.

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MATERIALS AND METHODS

This is a cross-sectional study carried out in the Department of Radiodiagnosis, Dr Chandramma Dayanand Sagar Institute of Medical Education and Research, Harohalli, Kanakpura road, Karnataka, India.

Pregnant women of 2nd and 3rd trimester, referred to 'Radiology' department from the outpatient and inpatient departments of 'Obstetrics and Gynecology' of the same hospital between October 2024 to September 2025 are included in the study.

The Radiologist performed all the transabdominal ultrasonography on a 'Siemens Acuson X300' machine using 'CH 5-2' probe after taking verbal consent from the patient. A questionnaire was used containing the following information e.g. women's age, parity, gravidity, date of last menstrual period. The questionnaire form also included about the result of transabdominal USG examination which included the following: singleton or multiple, dead or alive foetus, gestational age, and presence or absence of congenital anomalies.

All the above-mentioned variables along with the detailed anatomical survey at time of scan and demographic variables including gestational age were entered in a database file and analyzed by GraphPad InStat version 3.

RESULTS

During the study period of 2024 October to September 2025, a total of 5300 numbers of 2nd and 3rd trimesters prenatal USG were done. 90 numbers of congenital anomalies were detected among the pregnant women scanned. So, the antenatal prevalence of congenital anomalies was 1.73%. Of

the women having congenital abnormalities in the foetus, the majority were between 20-35 years old (73.33%) followed by those above 35 years (15.56%). The mean maternal age at the diagnosis was 25.5 years (SD ± 6.15). Women who had primary schooling represented 53.33%, with 26.67% of women were illiterate. Majority women were nullipara (42.22%) followed by primipara (31.11%). 34 cases (37.78%) were detected at 3rd trimester and majority at 2nd trimester ie. 56 cases (62.22%). The mean gestational age was 27 weeks (SD±6.42).

Variables	Category	Numbers	Percentage
Age in years	<20	10	11.11
	20-35	66	73.33
	>35	14	15.56
Literacy	Illiterate	24	26.67
	Primary schooling	48	53.33
	>Primary schooling	18	20.00
Occupation	Housewife	90	100
	Service	-	-
Parity	0	38	42.22
	1	28	31.11
	2	16	17.78
	3	8	8.89
Gestational age	2nd trimester	56	62.22
	3rd trimester	34	37.78

Table 1: Socio-demographic characteristics of anomaly positive group of women (N=90)

Variables	Numbers	Percentage
Live foetus at time of scanning	66	73.33
Intra uterine death foetus at time of scanning	24	26.67
Multiple pregnancy	6	6.67
Multiple anomaly of foetus	8	8.89

Table 2: Results of USG of anomaly positive group of women (N=90)

Category	Pattern of anomaly	Number	Percentage
Central nervous system	Hydrocephalus	8	42.22
	Anencephaly	16	
	Microcephaly	4	
	Meningomyoceles	10	
Gastro-intestinal	Duodenal atresia	8	
	Diaphragmatic hernia	4	
	Omphelocele	6	
	Gastro schiasis	2	
Genito-urinary	Polycystic kidney	4	06.67
	Pelvi-ureteric junction obstruction	2	
Musculo-skeletal	Skeletal dysplasia	6	22.22
Others	Hydrops foetalis	12	
	Conjoined twin	4	
	Cystic hygroma	2	
	Down syndrome	2	

Table 3: Anomalies involving different system (N=90).

DISCUSSION

Advanced diagnostic technology, especially USG, has made it possible to detect increased number of birth defects in infants antenatally and during the neonatal period. In present study, attempts had been made to find out the antenatal prevalence of anomalies in our hospital who attended for USG in 2nd and 3rd trimester.[6] The antenatal prevalence of congenital malformation in the present study was 1.73%, which is comparable with the observations of Nakling et al (2005) 1.47%, Souka et al (2006) 1.21%. Higher prevalence was observed some other studies like Sallout et al (2008) 2.96%, Alia et al (2010) 2.97%, Dolk et al (2010) 2.39% and Shah et al (2013) 2.38%. On the other hand, Taboo et al (2012) and Alakananda et al (2015) showed lower prevalence than the present study.[7]

This variation may be due to different geographical area, social factor, racial difference, observer variation and equipment quality. As true prevalence of congenital anomalies depends upon several factors and therefore two studies are never strictly comparable.[8] Though elderly age group and higher parity are considered as risk factors for congenital anomaly, in our study the incidence was observed higher in primigravida and younger age group. This may be due to earlier age of marriage in our scanning population.[9]

In present study, congenital malformations of the central nervous system were the highest (42.22%) followed by malformations of the gastrointestinal system (22.22%). Similar findings were observed by Agarwal et al (1999) and Perveen et al (2007). None of cardiac defects were diagnosed in our study. The low detection rate was because the four chamber view was only included in the scan in our study population, and no targeted imaging for foetal anomalies was done.[10]

CONCLUSION

In this study prevalence of fetal congenital anomaly was found to be 1.73%. CNS defect was found to be the commonest form of anomaly in our study population. Creating awareness regarding regular ANC and importance of anomaly scan on time can help in primary prevention of disability and reducing perinatal mortality and morbidity.

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