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Research Article

Prevalence of congenital abnormalities on routine ultrasound scan of second and third trimester pregnancy

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ABSTRACT

Background: Objective of the study was to evaluate the antenatal prevalence of major congenital abnormalities and its pattern.

Methods: This is a cross-sectional study carried out in the department of 'Radiology' at 'Fakhruddin Ali Ahmed Medical College and Hospital', Barpeta. Patients in the 2nd and 3rd trimester of pregnancy, referred to 'Radiology' department from the outpatient and inpatient departments of 'Obstetrics and Gynaecology' were included in the study. Antenatal ultrasound findings were statistically analyzed on structured data collection form.

Results: Total 2650 numbers of 2nd and 3rd trimester prenatal ultrasonography (USG) were done. Out of these 45 numbers of congenital anomalies were detected. The antenatal prevalence of congenital anomalies was 1.73%. The mean maternal age and mean gestational age at diagnosis was 25.5 years (SD \pm 6.15) and 27 weeks (SD \pm 6.42) respectively. Central nervous system (CNS) defect was the commonest (42%), of which maximum number had anencephaly defect i.e. 8 (17.78%) cases.

Conclusions: Antenatal ultrasound is a non-invasive highly sensitive, accurate and cost effective imaging technique which gives good results in experienced hands. It is recommended that the obstetricians should advise regular USG at least at 2nd trimester.

Keywords: Congenital anomaly, 2nd and 3rd trimester USG

INTRODUCTION

Congenital anomaly (CA) is the structural or functional anomaly (e.g. metabolic disorder) that occurs during intrauterine life and can be identified prenatally, at birth or later in life. These defects of prenatal origin result from defective embryogenesis or intrinsic abnormalities in the development process. Based on the World Health Organization report, about 3 million fetuses and infants are born and 276000 babies die within 4 weeks of birth every year, worldwide, from congenital anomalies.¹ Worldwide surveys have shown that birth prevalence of congenital anomalies varies greatly from country to country. It is reported about 3% in the United States,²

2.5% in India,³ and 2% to 3% in the United Kingdom.⁴ The prevalence is as low as 1.07% in Japan and as high as 4.3% in Taiwan.⁵ These variations of prevalence may be explained by social, racial, ecological, and economical influences.^{5,6} The most prevalent congenital disorders are congenital heart defects,⁷ neural tube defects⁸ and Down syndrome.⁹

There are various investigating tools which are available for diagnosis of congenital abnormalities. Out of these, ultrasound has become an invaluable tool for detection of many foetal abnormalities in the antenatal period. However, recent research found that the foetal structural abnormalities like foetal skull, brain, spine, abdominal

wall, limbs, stomach and bladder can be detected at 11-14 weeks scan in only 22.3 % of the cases; therefore, a second trimester anomaly scan was suggested in a routine antenatal care to increase the prenatal detection of the foetal defect.¹⁰ In certain studies, the sensitivity of detection of foetal anomalies, before the 24th week of gestation, was 93% for the central nervous system, 45.2% for the circulatory system, 85.2% for the digestive system, 85.7% for the urinary system, 84.6% for the musculoskeletal system and 95.2% for other anomalies. Therefore, it is suggested that ultrasonography between the 20th and 22nd weeks of pregnancy can detect the majority of congenital anomalies.¹¹

So, the overall detection time varies from early to late pregnancy depending upon the gestational age of the foetus in the first antenatal check up. The accuracy of detection of foetal abnormalities depends on various factors like experience of the ultrasonographer/ultrasonologist, quality of equipment, and type of malformation.

In India particularly in Northeastern 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 Fakhruddin Ali Ahmed Medical College (FAAMC), Barpeta.

METHODS

This is a cross-sectional study carried out in the department of 'Radiology' at 'Fakhruddin Ali Ahmed Medical College and Hospital', Barpeta. 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 January 2014 to December 2014 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 2014 January to December, a total of 2650 numbers of 2nd and 3rd trimesters prenatal USG were done. 45 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 \pm 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%). 17 cases (37.78%) were detected at 3rd trimester and majority at 2nd trimester ie. 28 cases (62.22%). The mean gestational age was 27 weeks (SD \pm 6.42).

Table 1: Socio-demographic characteristics of anomaly positive group of women (N=45).

Variables	Category	Numbers	Percentage
Age in years	< 20	5	11.11
	20-35	33	73.33
	>35	7	15.56
Literacy	Illiterate	12	26.67
	Primary schooling	24	53.33
	>Primary schooling	9	20.00
Occupation	Housewife	45	100
	Service	-	-
Parity	0	19	42.22
	1	14	31.11
	2	8	17.78
	3	4	8.89
Gestational age	2 nd trimester	28	62.22
	3 rd trimester	17	37.78

Table 2: Results of USG of anomaly positive group of women (N=45).

Variables	Numbers	Percentage
Live foetus at time of scanning	33	73.33
Intra uterine death foetus at time of scanning	12	26.67
Multiple pregnancy	3	6.67
Multiple anomaly of foetus	4	8.89

The results of USG of study population showed that out of 45 numbers of fetuses with congenital anomaly, 33 cases (73.33%) were live at the time of scanning and rest of the 12 cases (26.67%) were intrauterine foetal death. Three women (6.6%) had twin pregnancy with congenital anomaly of fetuses; out of which two were conjoined

twins. 4 fetuses had multiple congenital anomalies (8.8%).

Table 3: Anomalies involving different system (N=45).

Category	Pattern of anomaly	Number	Percentage
Central nervous system	Hydrocephalus	4	42.22
	Anencephaly	8	
	Microcephaly	2	
	Meningomyoceles	5	
Gastro-intestinal	Duodenal atresia	4	22.22
	Diaphragmatic hernia	2	
	Omphalocele	3	
	Gastro schiasis	1	
Genito-urinary	Polycystic kidney	2	06.67
	Pelvi – ureteric junction obstruction	1	
Musculo-skeletal	Skeletal dysplasia	3	06.67
Others	Hydrops foetalis	6	22.22
	Conjoined twin	2	
	Cystic hygroma	1	
	Down syndrome	1	

Out of the 45 congenital anomaly fetuses, central nervous system (CNS) defect was the commonest (42%), of which maximum number had anencephaly defect ie. 8 (17.78%) cases. This is followed by Gastro-intestinal (GI) system defect which shared 22.22% of defect. Within 22.22% of miscellaneous category of anomaly, most of the cases were hydrops foetalis 6 (13.33%).



Figure 1: Anencephaly.

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



Figure 2: Meningoencephalocele.



Figure 3: Foetal ascites in hydrops foetalis.

out the antenatal prevalence of anomalies in our hospital who attended for USG in 2nd and 3rd trimester. 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%,¹² Souka et al (2006) 1.21%.¹³ Higher prevalence was observed some other studies like Sallout et al (2008) 2.96%,¹⁴ Alia et al (2010) 2.97%,¹⁵ Dolk et al (2010) 2.39%¹⁶ and Shah et al (2013) 2.38%.¹⁷ On the other hand, Taboo et al (2012)¹⁸ and Alakananda et al (2015)¹⁹ showed lower prevalence than the present study. 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. 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.²⁰ This may be due to earlier age of marriage in our scanning population.

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)²¹ and Perveen et al (2007).²² 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.

CONCLUSIONS

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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Ethical approval: The study was approved by the Institutional Ethics Committee

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