Original Article

DOI: 10.17354/ijss/2016/113

Role of Ultrasonogram in Detection of Congenital Anomalies and Prevalence of Anomalies in High-risk Pregnancies

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Abstract

Introduction: In any antenatal population, about 2-3% of the foetus are affected by congenital anomalies it is mainly due to intrinsic genetic pathology and abnormal embryogenesis during the early gestational period.

Objective: To categorize the type of major congenital anomalies in routine second and third trimester ultrasonogram (USG) and its association with high-risk pregnancy.

Methods: This is a cross-sectional study carried out in the Department of Obstetrics and Gynaecology at Government Theni Medical College and Hospital, Tamil Nadu. The study population includes about 6250 antenatal mothers in the second and third trimester, who were subjected to USG by expert radiologists and the ultrasound findings were analysed on a statistical basis in structured data collection form.

Results: About 6250 antenatal mothers of the 2nd and 3rd trimester were subjected to USG out of which 122 mothers were found to have an anomalous foetus. (1) The antenatal prevalence of congenital anomalies was 1.95%, (2) The mean gestational age and mean maternal age at diagnosis was 24 weeks (Standard deviation [SD] ± 5.15) and 28.5 years (SD ± 6.10), respectively. Central nervous system was the most common system affected out of which maximum anomaly was anencephaly.

Conclusion: USG is a cost-effective, non-invasive and sensitive tool in detecting the congenital anomalies antenatally in the hands of an experienced radiologist. Besides the first-trimester USG, the second trimester scan is mandatory in detecting anomaly in the growing foetus, as the majority of congenital anomalies are detected between 20 and 22 weeks of pregnancy.

Key words: Anencephaly, Congenital anomalies, Second and third trimester

INTRODUCTION

In any antenatal population, about 2-3% of the foetus are affected by congenital anomalies it is mainly due to intrinsic genetic pathology and abnormal embryogenesis during the early gestational period. The factors such as racial, social, environmental, and economical factors are responsible for the type and prevalence of the anomalies in a different population.

The most common anomalies noticed are in the nervous system, cardiovascular system and genitor-urinary system.

Access this article online

www.ijss-sn.com

Month of Submission : 01-2015
Month of Peer Review : 02-2015
Month of Acceptance : 02-2015
Month of Publishing : 03-2015

Although there are many investigations for detection of the anomalies, ultrasonogram (USG) is the gold standard until now in modern obstetrics. Therefore, routine anomaly USG is advised as early as 11-14 weeks and again at 20-22 weeks of gestation which detect almost 95% of the major anomalies.

In India because of the increasing awareness of detection of anomalies by ultasonogram, the percentage of mental and physical handicap is in the declining phase. Detection of anomalies in the earlier gestation age is essential to reduce the maternal risk during expulsion process and to improve future obstetric outcome. The main aim of the study is to evaluate the prevalence of the anomalies in high-risk maternal population.

METHODS

This is a cross-sectional study performed in the Department of Obstetrics and Gynaecology at Government Theni

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Medical College and Hospital. About 6250 antenatal mother of 2nd and 3rd trimester were screened for the congenital anomalies by an expert radiologist from January 2015 to December 2015.

Form B consent is obtained from all the antenatal mothers. Expert trans abdominal USG was carried on sonoray DS30 machine using 3.5-5 mHz ultra-serve curvilinear probe.

A proforma containing age, parity, investigations for the maternal disease was used.

RESULTS

During the study period of January 2015 to December 2015, a total of 6250 antenatal mothers of second and third trimester were screened. About 122 numbers of congenital anomalies were detected. The antenatal prevalence of congenital anomalies was found to be 1.95%. Of the antenatal mothers screened, the majority were between 20 and 35 years of age (94) followed by those <20 years of age (24). The mean maternal age at diagnosis was 28.5 years. Most of the women were illiterate accounting 73%, and those who had primary education represented 20%. The majority of the women were primi (50.8%) followed by second gravida (40.16%). The majority of the cases were detected at 2nd trimester, i.e., 76 cases (62.29%) the mean gestational age during the time of diagnosis was 24 weeks of gestation (Table 1).

Out of the study population (n = 122), 105 (86%) foetuses with congenital anomaly were alive at the time of scanning and the rest 17 (14%) numbers were intrauterine death. 4 Women had twin pregnancy with anomalous foetuses. Out of which one was conjoint twins. 20 foetuses had multiple anomalies (Table 2).

Out of 122 anomalous foetuses, central nervous system (CNS) defect was the most common accounting 38.5% of which maximum number had anencephaly (23.77%) followed by genitourinary system 26.22% we notice a high sensitivity in the detection rate of CNS, genitourinary and cardiac anomalies (Table 3, Figure 1 and 2).

Out of 122 anomalous, 56 cases were high-risk pregnancies.

Teenage pregnancy (24 cases) accounts for 42.8% of the total high-risk cases followed by diabetes, seizure, hypertension, elderly age (Table 4).

Table 1: Socio-demographic variables in anomaly positive cases (n=122)

Variables	Category	Numbers (%)
Age group	Adolescent (<20 years)	24 (19.6)
	Middle age (20-35)	94 (77)
	Elderly (>35)	4 (3.27)
Literacy	Illiterate	89 (72.95)
	Primary education	24 (19.67)
	Graduation	9 (7.37)
Occupation	Unemployed	109 (89.3)
	Employed	15 (12.29)

Table 2: Obstetric characteristics of anomaly positive population (n=122)

Variables	Category	Numbers (%)
Parity	0	62 (50.8)
	1	49 (40.16)
	2	9 (7.37)
	3	2 (1.63)
Gestational age	2nd trimester	76 (62.29)
	3rd trimester	46 (37.7)
Viability	Alive	105 (86)
	IUD	17 (14)

Table 3: Prevalence of anomalies in different organ systems

Category	Total (%)	Anomaly	Number (%)
CNS	47 (38.5)	Hydrocephalus	12 (9.8)
		Anencephaly	29 (23.77)
		Meningomyelocele	4 (3.2)
		Spina bifida	2 (1.6)
Genito urinary system	32 (26.22)	Polycystic kidney	2 (1.6)
		Obstructive uropathy	30 (24.6)
Cardiac	13 (10.6)	Shunt diseases	13 (10.6)
GI	10 (8.1)	Polycystic kidney disease	2 (1.6)
		Obstructive nephropathy	30 (24.6)
Musculo skeletal	8 (6.5)	Skeletal dysplasia	8 (6.5)
		Hydrops foetalis	1 (0.8)
Others	12 (9.8)	Conjoint syndrome	1 (0.8)
		Down's syndrome	3 (2.4)
		Single umbical artery	7 (5.7)

GI: Gastrointestinal

Table 4: Association between high-risk cases and congenital anomalies

High-risk	Numbers (%)
Teenage	24 (42.8)
Diabetes	9 (16.07)
Hypertension	5 (8.9)
Elderly	4 (7.1)
Previous Iscs	10 (17.85)
Seizure	4 (7.1)

Total number of high-risk cases reported 56

DISCUSSION

Routine B mode transabdominal USG has made it possible to detect increased number of birth defects antenatally. The antenatal prevalence of congenital malformation in the present study was 1.95% which is comparable with the observations of Wong *et al.*[1] which is 1.4% and

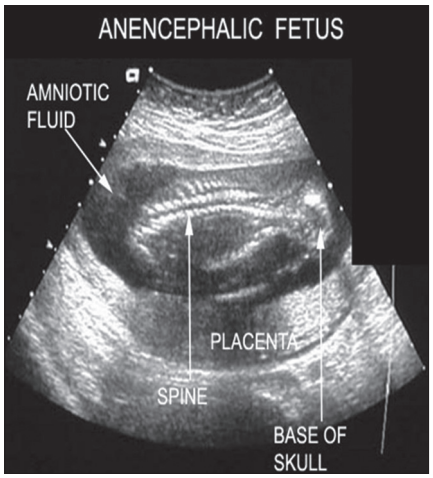

Figure 1: Ultrasound picture of an anencephalic foetus detected in second trimester

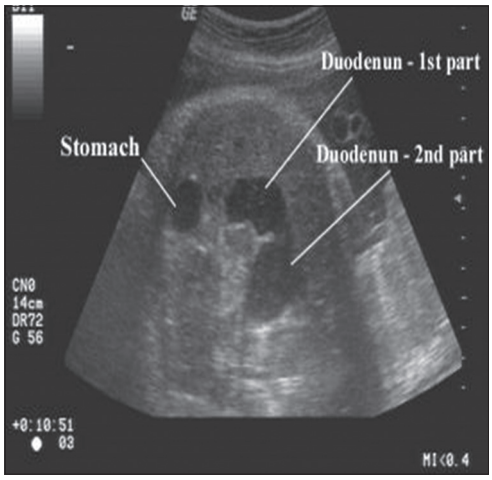

Figure 2: Ultrasound picture of a foetus showing double bubble sign in duodenal atresia

Nakling *et al.*[2] which is 1.5%, Patel *et al.*,[3] which is 1.6%. The routine ultrasonogram in antenatal mothers detects about 40% of anomalies compared to 28% those who undergo ultrasonogram on risk basis which is comparable with observations of Boyd *et al.*,[4] Temtamy *et al.*,[5] and Biri *et al.*[6]

The detection rate of anomalies by ultrasonogram is depend upon the prevalence of anomalies in the study population, the experience of the sonologist , the gestational age at the time of scanning, maternal age and medical risks.

Though elderly age group and higher parity are considered as risk factors for congenital anomaly, in our study the incidence was observed higher in younger age group and primigravida. This may be due to earlier age of marriage and early pregnancy in our screening population similar observations were present in the study of Lin *et al.*,[7] and Centers for Disease.[8] In present study, congenital malformations of the central nervous system were the highest (38.5%) followed by malformations of the genitourinary system (26.22%). Similar findings were observed by Sallout *et al.*,[9] Perveen *et al.*[10] and Mahela *et al.*[11]

Due to early marriage and early pregnancy, poor literacy, poor intake of folate, there is increased prevalence of CNS malformations in our study population. Single umbilical artery was a common anomaly detected in miscellaneous group which is around 5.7% of the total anomalies. It is comparable to Canfield *et al.*[12], Lee[13], Alia *et al.*[14] and Souka *et al.*[15]

CONCLUSION

Prevalence of congenital anomalies in our study is 1.95%. CNS is the most common system involved of which anencephaly is the maximum. Health education and creating awareness regarding the importance of detection of anomalies will be the first step in preventing and reducing adverse perinatal outcome in such pregnancies.

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How to cite this article: Aravazhi M, Balasubramaniam T, Sambath SD. Role of Ultrasonogram in Detection of Congenital Anomalies and Prevalence of Anomalies in High-risk Pregnancies. Int J Sci Stud 2016;3(12):10-13.

Source of Support: Nil, **Conflict of Interest:** None declared.