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Radiology

Determination the Detection and knowledge of Congenital Anomalies in Sudanese Pregnant Women during the First Trimester of Pregnancy by Using Ultrasonograph

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Abstract **Original Research Article**

Background Determination the detection and knowledge of Congenital Anomalies in Sudanese Pregnant Women during The First Trimester of Pregnancy Using Ultrasonography. Yet in a developing country like Sudan majority of pregnant women are not privileged to get timely diagnosis. ***Aims and Objectives:*** To assess the present status and potential of first trimester Ultrasonography in detection of fetal congenital structural malformations and evaluated the maternal knowledge of congenital. ***Methodology:*** This was a retrospective observational study conducted at antenatal Ultrasonography clinic in primary health care in soba hospital. All pregnant women at second trimester scan and women with first trimester were included. ***Results:*** Out of 2.880 pregnant women undergoing ultrasound, 500 women were at first trimester and there are no congenital anomalies were detected as there are some birth defects were seen later on, and the degree of knowledge is 46%. ***Conclusion:*** The first trimester Ultrasonography could have identified 50% of major structural defects. This focuses on the immense need of the hour to gear up for early diagnosis and timely intervention in the field of prenatal detection of congenital malformations.
Keywords: Congenital Anomalies, Pregnant Women, Ultrasonography.
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INTRODUCTION

A congenital anomaly is an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability, or is fatal. Each year, eight million children are born worldwide with congenital anomalies, of which 3.3 million before the age of five; 3.2 million of the survivors may be mentally and/or physically disabled [1]. The prevalence of birth defects is comparable all over the world; about 3% in the United States [2], 2.5% in India [3], and 2% to 3% in the United Kingdom [4]. The most prevalent conditions include congenital heart defects [5] orofacial clefts, Down syndrome [6] and neural tube defects [7].

There are number of laboratory and imaging studies available for detection of these anomalies. Out of these, ultrasound is the one which gives a great amount of information about the structure and to some extent physiological aspects of the state of fetus. Some anomalies like anencephaly can be picked as early as 12 weeks when skull primary ossification is complete [8]. The overall detection time varied from early to late pregnancy depending upon when the patient reports to hospital for antenatal checkup.

Second trimester ultrasound scan has become an essential part of antenatal care. In cases where a major structural defect is identified, termination of pregnancy is offered [9]. The morbidity and mortality of this procedure increases with advancing gestation. Therefore early detection of such abnormalities will result in the reduction of such complications. The diagnostic ability of ultrasound is well established by a number of studies [10, 11].

Detection of fetal abnormalities depends on a number of factors including the nature or type of abnormality, sophistication of equipment and experience of operator. Pediatrician should realize that our work does not stop at just to helping a mother to give birth to a healthy baby but also to give this world a healthy child and adult as well. All this is possible only when any abnormalities that could affect the baby in future should be detected at the earliest. As we all know that man has already stepped into 21st century and the

medical sciences has made a breakthrough in almost all the fields that were never been touched before.

It is such a useful and helpful device, which helps to detect not only the intrauterine well-being of fetus, but also any congenital anomalies that cause a huge amount of psychological trauma suffered by the prospective parents, who were not taking into account the time and money spent in order to ensure the birth of healthy baby. Now with advent of high-resolution real-time Ultrasonography. The concept of the prenatal care has changed profoundly. The scope of the antenatal care has expanded to include a broad range of fetal diseases such as growth disorders and congenital anomalies.

The sonologist with the help of pediatrician can now inform obstetrician a whole new range of findings besides giving differential diagnosis, patterns of inheritance, mechanism of disease, prognosis, and optimal obstetric care during pregnancy.

Imaging

The published background risk of major or minor structural congenital anomalies is estimated at 2% to 3.5% [4, 10, 11]. The following three studies emphasize that not all anomalies are detected prenatally.

Lemyre *et al*. reported their experience in a Canadian tertiary level unit. They demonstrated a residual risk of 2.9% (95% CI 2.3 to 3.7) for any congenital anomaly at birth after a second trimester level II ultrasound examination with or without amniocentesis in a population considered to be at increased risk for fetal anomaly on the basis of personal or familial history [12]. The overall rate of congenital anomalies in their population was not provided, and sensitivity of the ultrasound could therefore not be determined.

The RADIUS study provides insight on the detection of fetal anomalies using prenatal ultrasound. The overall incidence of major anomalies present at birth was 2.3%. The overall anomaly detection rate in the screened population was 35% (65/187), including almost one half of those deemed detectable by ultrasound. The detection rate of anomalies before 24 weeks' gestation was significantly higher in tertiary units (35%) than in non-tertiary units (13%) (Relative detection rate 2.7; 95% CI 1.3 to 5.8), although only one half of the anomalies detected were detected before 24 weeks [4]. Although its detection rate was lower than rates in some contemporary studies that reported detection rates as high as 61%[13], the RADIUS study highlights the potential benefits of a tertiary unit in identifying the majority of major structural anomalies present in a fetus. It is therefore suggested that all suspected fetal anomalies be re-evaluated in a tertiary unit in an attempt to provide the most detailed ultrasonographic assessment possible.

In two separate studies, false positive rates were determined to be 0.1% to 0.5% of all prenatal ultrasound examinations, the most frequently unconfirmed anomalies being ventriculomegaly, hydronephrosis, short limbs, and cysts (renal, pulmonary, abdominal, or cerebral) [14,15]. These may be true false positive results, or they may be spontaneous resolution of the condition. The use of 3-D ultrasound has been increasing consistently over the last two decades, although its contribution to prenatal diagnosis has been controversial. While assessing fetuses with congenital anomalies in the early 1990s, Merz and colleagues found that 3-D ultrasound provided additional information in 62% of cases, provided the same information in 36% of cases, and provided less information in 2% of cases [16]. In review articles published in 2005 and 2007, 3-D ultrasound was listed as particularly useful in assessing facial structures, limbs, and skeletal anomalies [17, 18].

The newest fetal imaging modality is ultrafast magnetic resonance imaging. Significant costs and difficulty of access limit the use of this modality to specific diagnoses or concerns. It appears most useful in the assessment of brain and lung anomalies, in the presence of complex multiple anomalies, when oligohydramnios is present, or when planning complex and high-risk in utero interventions [19–21].

Fetal X-ray was the first in utero imaging modality in obstetrics and was used before the advent of ultrasound for diagnostic purposes (number of fetuses, size, and position).

Congenital anomalies (CA)

The term CA refers to an anatomic abnormality that is present at the time of birth. It can be either major or minor. 14 % of CA detected antenatal or postnatal are minor which do not cause surgical or cosmetic significance.

It is useful to bear in mind the relative importance and unimportance of minor anomalies, as a few of them are at times detectable by antenatal abdominal ultrasound . Minor anomalies usually do not seriously interfere with viability or physical well-being of the infant. It is unlikely that the fetus to have two or more minor CA.

The different definitions of CA are Malformation, which is a morphologic defect of an organ, part of an organ or a large area of the body resulting from an intrinsically abnormal development process.

The first antenatal appointment

NICE recommends that the first antenatal appointment take place early in pregnancy (before 12 weeks) and that it may need to be booked as a double appointment due to the large amount of information and

assessments that are required. Information must be imparted in a way the woman can understand and backed up with written information, so she is in a position to make informed choices regarding options and care in her pregnancy.

Information and advice which should be covered in the first appointment is detailed in the separate Antenatal Care article.

Examination routinely done at the first appointment includes : (Measurement of weight and height in order to determine body mass index (BMI),measurement of baseline blood pressure (BP),testing of urine for glycosuria / proteinuria, pelvic examination ,breast examination ,weight ,blood pressure, Abdominal examination and ultrasound) [22, 23]

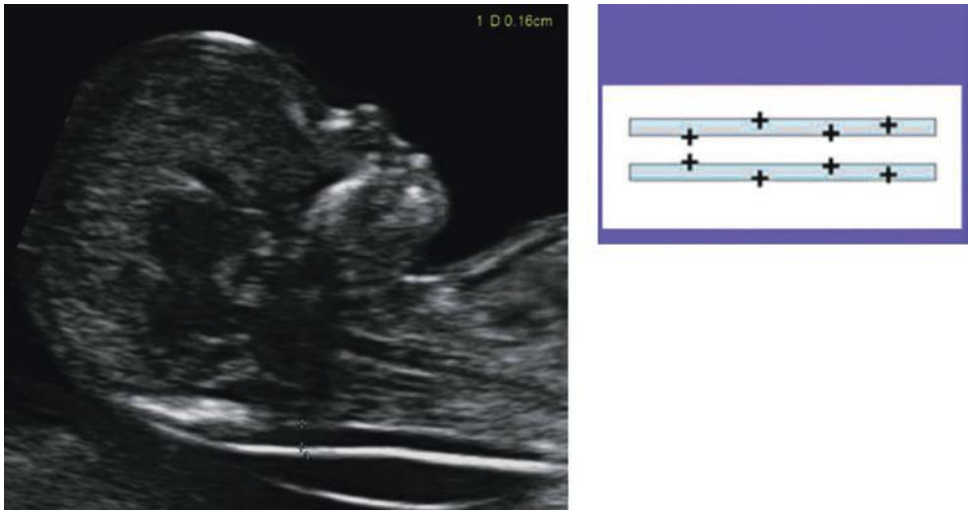


Fig-1.1: how to measurement the nuchal translucency

Table-1. 1: Normal measurement of nuchal translucency [24]

Pregnancy Mark	Normal Measurement
At 11 Weeks	Up to 2mm *Note: 9 of 10 babies with thickness 2.5-3.5mm will be normal.
12-13 Weeks	1.7mm (50^{th} percentile thickness) 2.8mm (95^{th} percentile thickness

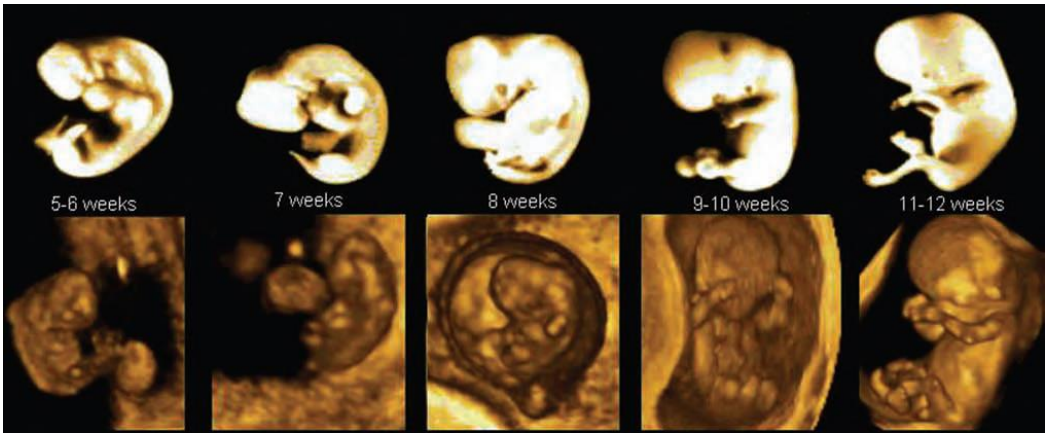


Fig-1-2: Comparison between a computer animation model of embryonic development and a series of in vivo images of the human embryo by 3D sonography, emphasizing the development of the embryo in early pregnancy (adapted, with permission, from: Azumendi G, Kurjak A, Andonotopo W, Arenas JB. Three dimensional sonoembryology. In: Kurjak A Arenas JB, eds. Donald School Textbook of Transvaginal Sonography. London: Taylor and Francis, 2005: 396–407.4

MATERIALS AND METHODS

Across sectional hospital base study design will be used in the current to identify the prevalence of congenital anomaly among Sudanese pregnant women during the first trimester of pregnancy by using abdominal ultrasound and to evaluate the accuracy of abdominal ultrasound in the diagnosis of congenital abnormalities in the first trimester of pregnancy.

This study will be conducted in the state of Khartoum, in Soba University Hospital (which established in 1974, a teaching hospital and the reference to a shift to him cases of abnormal and critical from all over Sudan)**,** the Hospital staffed with full-time maternal-fetal medicine subspecialists. Its annual delivery rate was high.

Data collection

During the prenatal period, data will be collected by using questionnaires and fetal ultrasound assessments. Intervention questionnaire sheet which will be used contains the following data Socio-demographic data (age –Residence –Height-Educational level –Occupations- past medical history – family history-……. etc) Reproductive data (Gestational age -Mode of delivery –Parity……etc) Diagnosis of abdominal ultrasound (types of structural anomalies)

Data Analysis

Data was analyzed using statistical software SPSS version 20.0. Categorical variables were analyzed using descriptive statistics

RESULTS

A total of 2,880 women visited the antenatal Ultrasonography clinic in primary health care unite in soba hospital during the study period (3/2017-9/2018). Out of which 500 women were at first trimester. All women underwent transabdominal US.

As the clinic had a poor sonography machine, the outcome reports for women explain only the gestation age and site of the embryo. No congenital anomalies were detect during the research, however we found approximately 7% birth defect

The knowledge about the congenital anomalies for women whom underwent transabdominal US were 46%. All the women under went to ultrasound were between fifteen to forty years old most of them were between twenty to thirty years old. Table and figure (3.1)

30% of the women had answer with YES that the reason for congenital anomalies is the mother, 41% said NO and 29% said they have no knowledge. Table and figure (3.2).

11% of them had answer with YES that the reason for the congenital anomalies is the fetus, 55% said NO and 34% had on knowledge. Table and figure (3.3)

16% of them had answer with YES its contagious disease, 53% said NO and 31% had no knowledge. Table and figure (3.4)

40% answer with YES it can be treated medically early, 35% said NO and the left 25% had no knowledge. Table and figure (3.5)

31% answer with YES it can be treated surgically, 32% said NO and the left 37% said they don't know. Table and figure (3.6)

58% answered YES it can be prevented, 19% said NO and the left 23% said they don't know. Table and figure (3.7)

66% said YES that it can result from drugs intake without medical supervision, 7% said NO and the other 27% said that they don't know.(3.8)

55% said YES that the age is an effective factor, 21% said NO and the other 24% said they don't know. Table and figure (3.9)

51% said YES that exposure to radiation is a leading factor, 15% said NO and the other 34% don't know. Table and figure (3.10)

14% said YES that iodine salt may decrease it, 21% said no and the 66% said they had no idea. Table and figure (3.11)

53% said YES that following before pregnancy instructions may decrease it, 18% said NO and the left 29% had no idea. Table and figure (3.12)

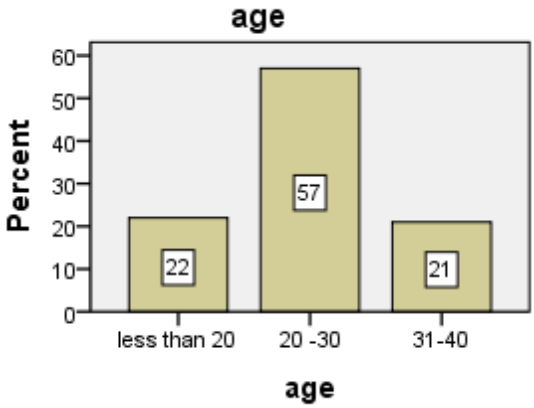
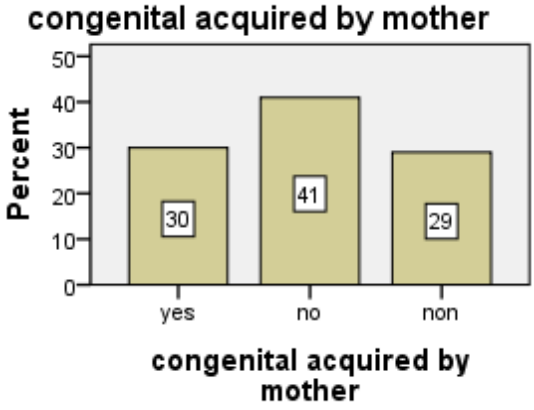


Fig-3.1



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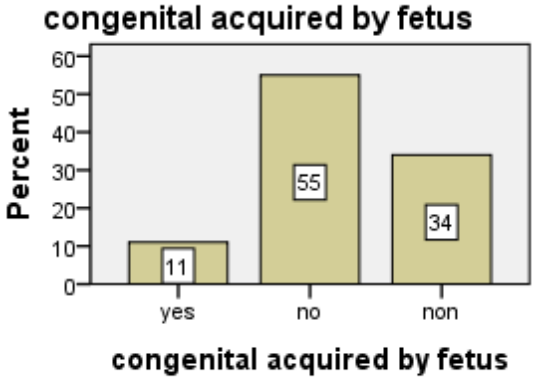

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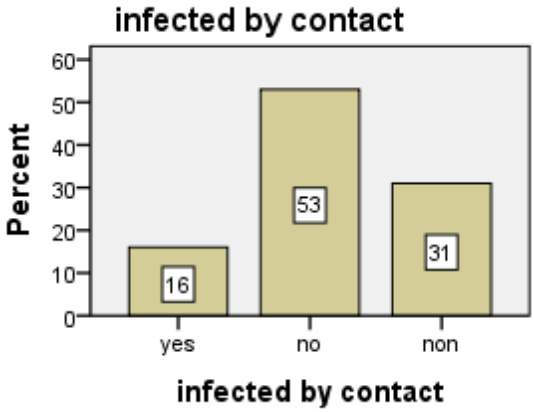

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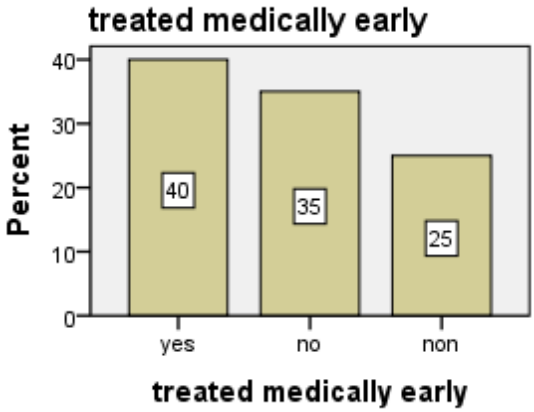
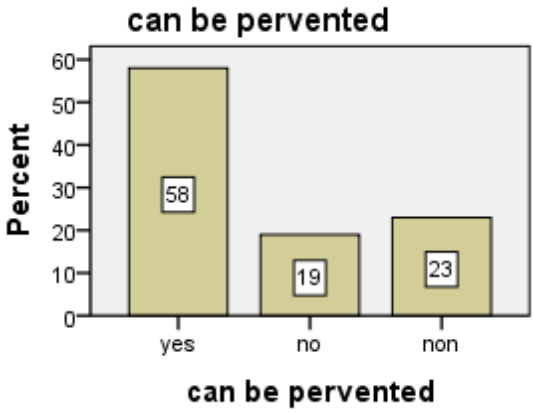


Fig-3.5

Fig-3.6



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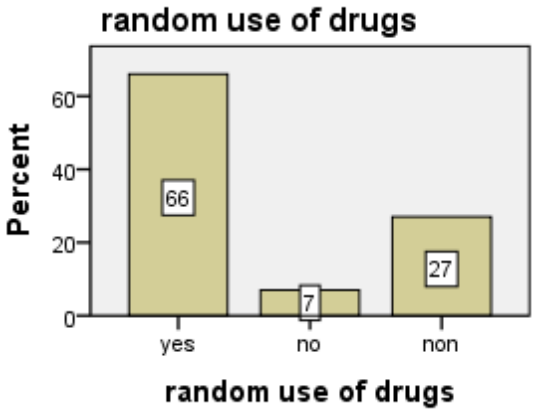

Fig-3.8

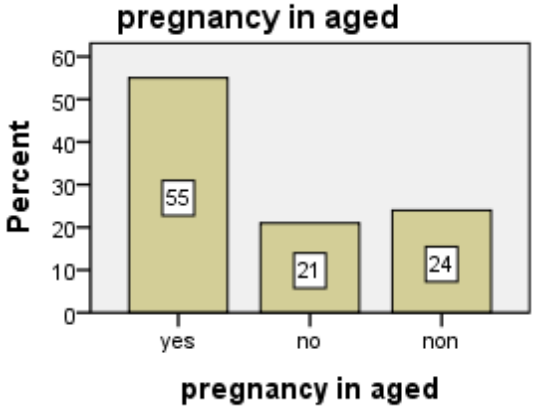

Fig-3.9

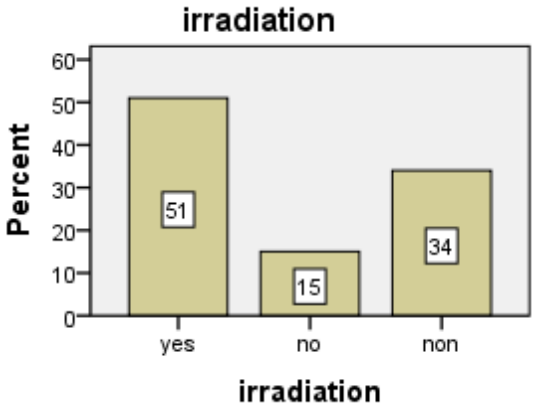

Fig-3.10

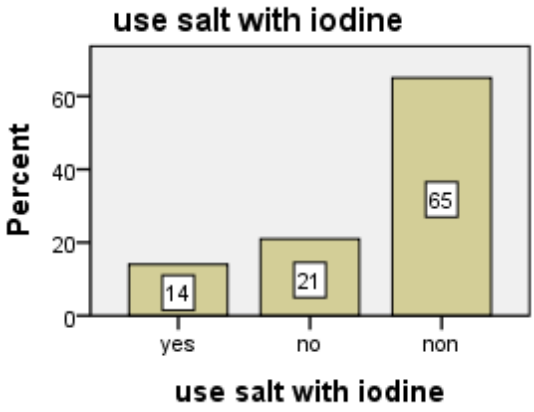

Fig-3.11

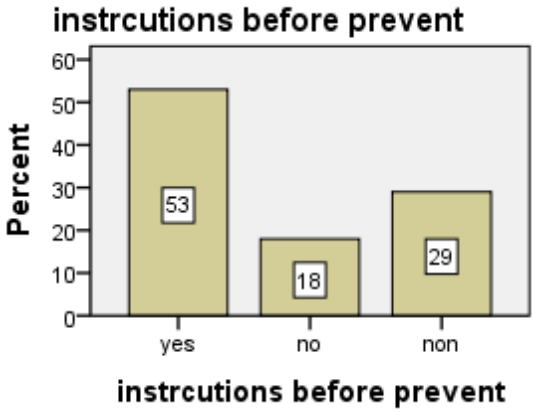

Fig-3.12

Table-3.1: Age of mother

	Frequency	Percent
Less than 20	110	22
20 – 30	285	57
31 – 40	105	21
Total	500	100

Table-3.2: congenital acquired by mother

	frequency	Percent
Yes	150	30
No	205	41
Non	145	29
Total	500	100

Table-3.3: Congenital acquired by fetus

	frequency	Percent
Yes	55	11
No	275	55
Non	170	34
Total	500	100

Table-3.4: Infected by contact

	Frequency	Percent
Yes	80	16
No	265	53
Non	155	31
Total	500	100

Table-3.5: Treated medically early

	Frequency	Percent
Yes	200	40
No	175	35
Non	125	25
Total	500	100

Table-3.6: Treated by surgical

	frequency	Percent
Yes	155	31
No	160	32
Non	185	37
Total	500	100

Table-3.7: Can be prevented

	Frequency	Percent
Yes	290	58
No	95	19
Non	115	23
Total	500	100

Table-3.8: Random use of drugs

	frequency	Percent
Yes	330	66
No	35	7
Non	135	27
Total	500	100

Table 3.9 Irradiation

	frequency	Percent
Yes	255	51
No	75	15
Non	170	34
Total	500	100

Table-3.10: Pregnancy in aged

	Frequency	Percent
Yes	275	55
No	105	21
Non	120	24
Total	500	100

Table-3.11: Use salt with iodine

	Frequency	Percent
Yes	70	14
No	105	21
Non	325	65
Total	500	100

Table-3.12: Instructions before prevent

	Frequency	Percent
Yes	265	53
No	90	18
Non	145	29
Total	500	100

DISCUSSIONS

Our study demonstrated that the first-trimester ultrasound performed at first trimester was able to detect the major structural anomalies in singleton pregnancies. This was based on the researches done in different research sites, which were used 3-4-dimensional ultrasound to revealing the major structural anomalies in singleton pregnancies.

Yet in a developing country like Sudan majority of pregnant women are not privileged to get timely diagnosis, all of them had easy termination of pregnancy. Now we will review the researches which regard to detect the congenital anomalies in first-trimester by using Ultrasonography.

Today, detection of embryonic and fetal structural abnormalities in the first trimester has frequently been reported. One has to distinguish between diagnosis during the early period until about 10 weeks when the embryo or early fetus is small and transvaginal ultrasound is applied, and diagnosis during the late period at the nuchal translucency screening, usually carried out using transabdominal ultrasound. Early first-trimester abnormalities are often diagnosed by chance on clinical indications, whereas late first-trimester diagnoses are the result of systematic screening using ultrasound markers [25].

The first trimester scan? The answer may be found in studies that assessed the performance of first trimester sonographic screening of chromosomally normal fetuses. In the 1995 study by Pandya *et al.* the incidence of structural defects among 821 chromosomally normal fetuses was approximately 4% and included cardiac, diaphragmatic, renal and abdominal wall abnormalities. Survival decreased from 97% for those with nuchal translucency thickness of 3 mm to 91% for a nuchal translucency thickness of 4 mm and 53% for a nuchal translucency thickness of ≥5 mm. In a 2006 study of 39,572 unselected women by Saltvedt *et al.* after excluding chromosomally abnormal pregnancies, the authors detected 69% of lethal anomalies by first trimester scan at 12–14 weeks' .Normal sonographic findings provide reassurance for women at high risk while detection of fetal malformation during first trimester enables discussion and decisions about possible treatments and interventions, including termination of pregnancy, during early stage of pregnancy [26].

This study to emphasize the importance of the first-trimester scan in the early detection of aneuploidy and structural fetal anomalies. In this small unselected low-risk population, the first-trimester scan detected 83% of aneuploidies and 70% of major structural anomalies. Our results are comparable to previously published studies from other centers and further exemplify the invaluable role of the first-trimester scan in the early detection of aneuploidy and structural anomalies in an unselected low-risk population [27].

In Sanjay Gandhi Postgraduate Institute of Medical Sciences. Yet in a developing country like India majority of pregnant women are not privileged to get timely diagnosis. All pregnant women attending Department of Maternal and Reproductive Health, OPD, from August 2009 till October 2013 were enrolled in the study. Patients were diagnosed prior to 12 weeks for neural tube defect, holoprosencephaly, gastroschisis, cystic hygroma, and anencephaly. All of them had easy termination of pregnancy. Out of 4080 pregnant women undergoing ultrasound, 312 (7.6%) had fetal structural malformation. Out of 139 patients who were diagnosed after 20 weeks, 47 (33.8%) had fetal structural anomalies which could have been diagnosed before 12 weeks and 92 (66.1%) had fetal

malformations which could have been diagnosed between 12 and 20 weeks. The first trimester ultrasonography could have identified 50% of major structural defects compared to 1.6% in the present scenario. This focuses on the immense need of the hour to gear up for early diagnosis and timely intervention in the field of prenatal detection of congenital malformation [28].

In Partners Human Research Committee, this study was to determine whether first-trimester aneuploidy screening sonography initially performed by credentialed sonographers was useful for detecting fetal anomalies between 11 and 14 weeks' gestation. A structural anomaly was identified in 50 of 9692 fetuses (0.5%) at the time of the first-trimester scan. The median CRL for anomalous fetuses was 55 mm (range, 36–77 mm). Of the 50 fetuses with a structural anomaly in the first trimester, 24 (48%) had an NT of 3.0 mm or greater, and 5 (10.0%) had an NT that was unable to be measured because of the severe disruptive anomaly. Anomalies diagnosed in fetuses at or before 14 weeks were lethal in 15 of 50 (30%), major in 31 of 50 (62%), and minor in 4 of 50 (8%). The most commonly identified anomalies at or before 14 weeks were abnormalities involving the fetal abdominal wall, face/profile, central nervous system, and heart. All cases of acrania, alobar holoprosencephaly, omphalocele, limb–body wall complex, ectopia cordis, and sirenomelia were diagnosed in the first trimester. Other anomalies, such as absent limbs, facial clefts, micrognathia, and congenital diaphragmatic hernias and skeletal dysplasias, were occasionally diagnosed. Other anomalies such as agenesis of the corpus callosum, minor renal anomalies, and congenital pulmonary lesions were never diagnosed in the first trimester [29].

About half of major structural abnormalities can be diagnosed in the first trimester. Increased nuchal translucency or abnormal ductus venosus blood flow appear to be associated with cardiac and skeletal defects and may facilitate early detection [30].

The overall sensitivity of 3D_VR_US for detecting structural abnormalities was 62.6% (169 of 270) and was 52.2% (141 of 270) using 2D/3D_US, *P* = .075. Sensitivity of 3D_VR_US compared to 2D/3D_US was higher for small details like polydactyly (4 of 5 vs 1 of 5) and facial clefts (5 of 5 vs 2 of 5) and lower for holoprosencephaly (2 of 5 vs 5 of 5). Malformations of skeleton and limbs were significantly more often correctly diagnosed using 3D_VR_US (*P* = .013) [31].

A search in Pub Med, MEDLINE, Embase, Cochrane Library, and ClinicalTrials.gov was performed (January 2000 to December 2012). At 14 weeks of gestation or less, fetal echocardiography detected 53% of congenital heart disease compared with 43% by complete scan (P=.040). The use of Doppler did not improve the detection rate for congenital heart defects (52% compared with 44%, respectively; P=.11). Multiple defects were identified more frequently than isolated malformations (60% compared with 44%; P=.005). The detection rate was higher combining transabdominal and transvaginal techniques (62%) than either abdominal (51%) or transvaginal (34%; P<.001). Detection rate was higher in women at high risk (65%) than unselected population (50% P=.001). Because of the natural history of fetal defects and the late development of some organ systems, a number of fetal malformations remain undetected by early ultrasonography. The overall detection rate of early ultrasonography was 501 of 996 (51%). With regard to location of fetal malformation, the highest detection rate was observed for neck anomalies (24/26 [92%]) followed by anomalies of the abdomen (96/109 [88%]), brain and spine (81/158 [51%]), heart (201/418 [48%]),limbs (36/105 [34%]), genitourinary tract (40/116 [34%]), and face (8/23 [34%]). Other types of malformations were associated with a detection rate of 15 of 41 (36%). Box 1 groups the types of malformation according to their detection rates. Fetal heart assessment was performed by echocardiography alone in 224 (53%) and included in complete anatomy examination in 194 (47%) fetuses [32].

Early detection of malformation is tremendously improved with improvement in imaging technology. Yet in a developing country like India majority of pregnant women are not privileged to get timely diagnosis. To assess the present status and potential of first trimester Ultrasonography in detection of fetal congenital structural malformations. This was a retrospective observational study conducted at Sanjay Gandhi Postgraduate Institute of Medical Sciences. All pregnant women had anomaly scan and women with fetal structural malformations were included. Out of 4080 pregnant women undergoing ultrasound, 312 (7.6%) had fetal structural malformation. Out of 139 patients who were diagnosed after 20 weeks, 47 (33.8%) had fetal structural anomalies which could have been diagnosed before 12 weeks and 92 (66.1%) had fetal malformations which could have been diagnosed between 12 and 20 weeks. The first trimester Ultrasonography could have identified 50% of major structural defects compared to 1.6% in the present scenario. This focuses on the immense need of the hour to gear up for early diagnosis and timely intervention in the field of prenatal detection of congenital malformation [33].

CONCLUSION

All the researches confirm that, the first trimester Ultrasonography could have identified 50% of major structural defects. Increased nuchal translucency or abnormal ductus venosus blood flow appear to be associated with cardiac and skeletal defects and gastroschisis and cystic hygroma may facilitate early detection This focuses on the immense need of the hour

to gear up for early diagnosis and timely intervention in the field of prenatal detection of congenital malformations .as the maternal knowledge arrange between 66% high degree and 7% low degree about congenital anomalies.

Acknowledgments

No thanks are complete unless it is first to almighty God who has given us the ability to think and do some work which benefits humankind. Then we are in debt especially to our parents, brother, husband, my children without whose love and affection we wouldn't have been able to do this work effectively and efficiently.

Abbreviations

CRL	Crown-Rump Length
CA	Congenital Anomalies
U/S	Ultra-Sonography
LNMP	Last Normal Menstrual Period
EDD	Expected Date of Delivery
NICE	National institute for health and care excellence

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