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Novel Research on Fetal Abnormalities from Ultrasound Image - A Review

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***Abstract*— Detection of malformation in early stage itself is greatest achievement in the imaging technology. This developing technology is not adopted in many developing countries so that majority of pregnant women are not privileged to get timely diagnosis. Ultrasonography is a method of screening process in the fetal imaging which has been followed in current era to find out the abnormalities. The antenatal screening programs produce the detailed anatomy of the growing fetal. The classification depends on the neural, brain and heart abnormalities found in the fetal. The first trimester period is crucial period for the fetal so that the ultrasound examination shows the improvement and also the abnormalities. wide range of neuro-pathological changes may occur during this trimester period this changes must observed carefully so that the exact diagnosis can be given to enhance the fetal development. The observations have been demonstrated in the screening of fetal such that the physiological development can be recorded clearly. World survey depicts that approx 3 in 1000 pregnancies have been come across the abnormalities and clinical practices provide a conclusion for this issues. This research articles compares various abnormalities and its critical issues of various research works and case studies.**

Keywords: Ultrasound, Trimester, Anomaly

I. INTRODUCTION

Ultrasound imaging is a real time inexpensive technology which is used to find the status of growth of the fetus. So many methods are available to detect the abnormalities in the body like MRI, X-Ray, etc., but in case of the newly developing fetus which is of below 20 weeks the above methods may produce higher anatomical details but for the assessment of the fetal it is not essential. A medical standard provides the 2D and 3D Ultrasounds which produce much better results than the other scanning methods. The application of fetal ultrasound has revolutionized the management of pregnancy and its potential complications. Simple access to a screening programs and routine use of ultrasound imaging in the first and second trimester provided the best results in assignment of gestational age, diagnosis of multiple pregnancy, prediction of adverse obstetric outcomes during late pregnancy, prenatal diagnosis of structural or karyotypic condition and diagnosis of those conditions which may be amenable to in uterotherapy. Survey mentions 297,126 women gave birth to 301,810 babies in Australia. These pregnancies were largely uncomplicated, with most births resulting in healthy babies at full term. In some cases however, there may be problems with the baby's development. Problems in fetal development may have a genetic origin, or they may be structural conditions and it is estimated that major structural conditions occur in 2 -3.5% of pregnancies. Not all conditions can be detected antenatal; detection rates of major structural conditions are reported to be approximately 60% in unselected series, and depend on the anatomical system involved and on the expertise of the ultrasound operator

All over the pregnancy ratio a survey depicts that almost 2 to 4% fetus are affected by structural abnormalities. In majority of countries worldwide, second trimester scan between 18 and 22 weeks remains the standard of care for fetal anatomical assessment; however, th e recent literature describes the advanced detection techniques which used in fetal abnormality detection during the first trimester. Besides nuchal abnormalities a wide range of central nervous system, heart, anterior abdominal wall, urinary tract, and skeletal abnormalities can be diagnosed between 11 and 14 weeks of scan. The main use of first trimester ultrasound is early detection of anomalies which may be of major or minor. This helps to give correct the disabilities and the give reassurance which avoids pregnancy termination. The recent literature suggests classification of fetal abnormalities as always detectable, potentially detectable, and undetectable till first trimester and anomaly scan. The diagnostic effectiveness of first trimester anomaly scan and echocardiography is between 10 and 13 weeks has been assessed as medium risk. The prevalence of major anomalies in their study group was 2.8%. The overall detection rate of fetal anomalies including cardiac defects was 78% and increased with raised nuchal thickness. This highlights the scope of first trimester scan apart from its conventional role in detection of chromosomal abnormality.

First trimester screening is now no more limited to detection of raised nuchal thickness (NT). Yen-Ni Chen [4] analyzed various cases to assess the prevalence and detection rate of major anomalies by applying first trimester anomaly scan and fetal echocardiography. In the fetal anomalies the normal NT and more than half of them could be detected in first trimester. Hence, even fetuses with normal NT should be offered first trimester anomaly scan and fetal echocardiography considering the ethical principles of nonmaleficence, justice, and respect for autonomy of pregnant women. Even in this era the benefits of this established technology are not in the reach of all. A vast majority of patients in India are not yet undergoing anomaly scan. We frequently encounter malformations always or potentially detectable during first trimester scan at third

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trimester or in postnatal period. It depends on both the expertise and resources available along with the awareness and sensitization in general population.

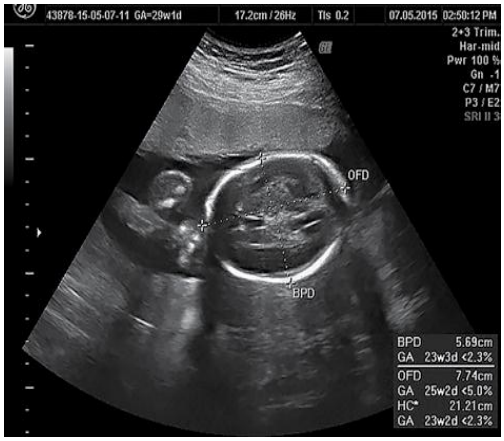

Fig 1. Foetal measurements: the difference in estimating gestational age

This fact of diagnosis is particularly more important in countries like India where medical termination of pregnancy [5] is legally allowed up to 20 weeks of gestation irrespective of malformation being lethal. Most of cases many no of patients are diagnosed with fetal abnormalities beyond 20 weeks and in that situation they are forced to seek termination services at small substandard centers. Many of such patients get deteriorated due to septic abortion and unnecessary hysterotomy and so forth. Question then arises that where lies the fault, the awareness of the patients or the expertise of the sonologist.

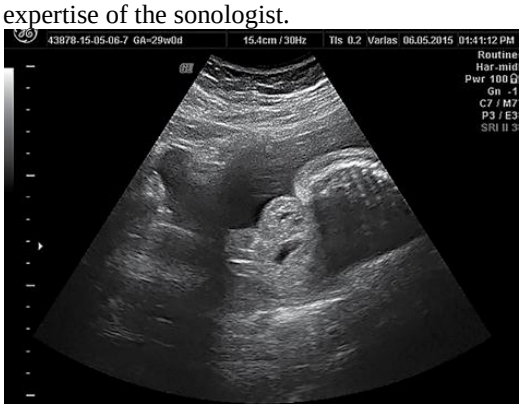

Fig 2. Abdominal wall defect with abdominal viscera of the abdominal cavity

Henceforth, the study was planned to assess the prevalence of fetal malformation in a tertiary care referral centre and to assess the present status of first trimester ultrasonography in the detection of fetal malformations in a tertiary care centre in India.

Ultrasound and fetoscopy are both valuable techniques in the antenatal diagnosis of fetal defects. They should not, in fact, be regarded as alternatives but should be used in a complementary way to be most effective. Fetoscopy permits examination of external fetal structures by direct visualization. This means that very small anatomical structures such as hands, feet, and genitalia are clearly visualized. The disadvantages, however, are that it is not always possible to see the part of the fetus where the abnormality is suspected. Furthermore, the risk of abortion exceeds that of midtrimester amniocentesis. Diagnostic ultrasound has no known risk, and both external and internal structures can be visualized. The fetus is viewed, however, in two-dimensional slices, and certain parts, such as the fetal limbs, cannot always be fully visualized. Furthermore, small structures such as the genitalia, fingers, and toes are frequently beyond the resolving power of ultrasound. Ultrasound therefore is a primary method of diagnosis for fetal structural abnormalities and, indeed, could be used to screen the whole obstetrical population. Fetoscopy is used where there is a suspicion of a small external fetal defect of some clinical significance which is beyond the resolving power of ultrasound and which cannot be diagnosed by biochemical analysis of the amniotic fluid.

II.RELATED WORKS

A vast survey has been done on finding the abnormalities in fetus in the first and second trimester periods. The literature [1] depicts the Rubella virus infection to the fetal during the first trimester of pregnancy and it may results to congenital rubella syndrome (CRS). The author discussed and described the abnormalities using ultrasound features in the prenatal scans. Article focuses the signs of CRS which is accessible to diagnosis during the pregnancy time. Many researchers analyzed different cases of CRS and depicted the results by comparing the before and/or after birth. A separate identified database [15] are used to classified them to rectify the problems or not to diagnosis. Author described the most frequently reported abnormalities were cardiac defects, pulmonary artery stenosis, microcephaly, cataract, microphtalmia, and hepatosplenomegaly.

The finding from the literature [2] provides detail and occurrence of virus infections followed in cases of fetal microcephaly, the virus infection affects the central nervous system (CNS). Most patients with zika virus infection are asymptomatic. the author analyzed about zika virus and its symptoms to the pregnant women and most common symptom of fetal microcephaly is rash. Ultrasound examinations at 18 to 20 weeks [18] of development and serial ultrasound follow-ups are also suggested by the author to find the abnormalities. The literature analysis the various cases of fetal microcephaly, intracranial anatomy and extra cranial abnormalities and also the amniocentesis, which might be needed to suspect the virus infection and the genetic abnormalities in the fetus or other congenital infection.

Another finding from the author [3] describes the heart disease which is most commonly reported in 7 per 1000 live births. Literature describes the situs anomalies leads to congenital heart disease and also defines the variations of fetal situs anomalies. In general Situs [14] refers to the arrangement of the atria, and its vessels in the human body and also author described about the Situs solitus which is considered as normal arrangement. By comparing the normal situs and solitus the literature provides the detailed statics to find the abnormalities. situs inversus is the another term which describes the inverted arrangement of the viscera and atria, situs ambiguous defines the conflict in

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arrangements that can neither be identified as solitus nor inversus. This different types of situs abnormalities [16] are clearly described by the author so that the literature uses to obtain the definition about the abnormality which is easy to classify and study.

The findings from the article [4] give a systematic review in suspicion of fetal brain abnormality and the diagnostic performance of in utero MR (iuMR) imaging. Author used this method to confirm or exclude or provide some more clear information to compare the information provided by prenatal ultrasound scans (USS) Methods [15]. The findings of prenatal USS and iuMR imaging are more accurate to judge the fetal brain abnormalities. The case study which used in literature uses 34 different cases includes the criteria which had an outcome reference diagnosis determined by postnatal imaging, surgery or autopsy.

Another finding from the author [5] describes the controversy terminology in rhinitis inpregnancy. Gestational rhinitis is a relatively common condition, which has drawn increasing interest in recent years due to a possible association with maternal obstructive sleep apnea syndrome (OSAS) and unfavorable fetal outcomes [12]. The author reviewed the current knowledge on gestational rhinitis and similar conditions regarding their physiopathology treatment prognosis. The hormonal changes and management of rhinitis during pregnancy focuses on the minimal intervention which is required for symptom of relief. The literature concluded the maternal quality of life in both the otorhinolaryngologist and gestationalrhinitis by considering the necessary precaution and the treatment measures.

The findings from the article [6] describe the advantages of fetal ultrasonography in the detection of abnormalities in fetal limbs. The author defines the fetal limb anomalies with other malformations due to unusual syndromes. The literature analysis two cases of isolated fetal limb anomalies which involve the different part of the upper limb, namely phalangeal aplasia detected through routine ultrasound fetal examination. The etiology in any of the cases a bilateral thumb anomaly of the mother's hand could not be identified. The upper limb anomaly [17] was confirmed by the clinical, radiological and anatomopathological examinations of the aborted fetuses. Author provides a method of early detection of fetal limb anomalies considered as best opportunity while selecting the best therapeutic management and also genetic counseling also taken under consideration for further pregnancies.

Another finding from the article [7] depicts the diagnostic performance of first trimester ultrasound in detecting congenital anomalies in twins. The article provides a solution to different pregnancy characteristics and early detection of structural anomalies in a large unselected population of twin pregnancies. Author proposed a systematic review based on collected materials. Retrospective analysis is used to collect data from consecutive twin pregnancies booked for clinical care from the year 1997 till 2015. Predictive accuracy of those covariates independently associated with the occurrence of fetal anomalies was assessed with logistic regression analysis and ROC curves. The author used 1064 twin pregnancies data for the analysis includes 864 dichoronic and 220 monochronic. Out of which 48 pregnancies had one or more fetuses with structural abnormalities. The data used in the article to detect the structural abnormalities in twin pregnancies which has the likelihood of first trimester detection anomalies in twins. The abnormalities are found [21][13] in cranial vault, midline brain and abdominal wall defects increasing the discrepancy in crown-rump length and nuchal translucency. Majority of structural anomalies in fetuses with chromosomal differences are identified by standard ultrasound exam in second trimester. Chromosomal anomalies have the greatest probability for prenatal detection; the greater part is diagnosed by amniocentesis prior to standard ultrasound exam in high-risk women.

The findings from the article [8] describe the anti-epileptic drugs (AEDs) in the first trimester of pregnancy and specific congenital anomalies of the fetus. The author performed a complete data collection from various cases and studies to find signals for possible links between newer AED and specific congenital anomalies. The congenital anomalies were classified according to the congenital anomaly subgroups of European Surveillance of Congenital Anomalies. The author compared various specific congenital anomalies in fetuses with a reference database. Significantly higher pervasiveness based [15] on three or more fetuses with anomalies was considered as a signal for congenital anomalies and the other AEDs. topiramate and cleft lip with/without cleft palate and hypospadias were considered as a four strong signals which is found in association with lamotrigine and anencephaly and transposition. The author concluded the terms between monotherapy with a newer AED in the first trimester of pregnancy and a specific congenital anomaly.

The findings from the article [9] describes the Body-stalk anomaly characterized by an anatomic defect in the ventral abdominal wall and exit of the viscera of the abdominal cavity, associating spine and limb deformities and absence or shortening of the umbilical cord. Ultrasound is used to suggest the prenatal diagnosis and it should be confirmed by histopathology evaluation. The literature presents a case study of body-stalk anomaly diagnosed at 23 weeks of growth by a routine ultrasound evaluation of fatal prognosis and other abdominal wall defects.

The findings from the article [10] revealed a live women fetus with large abdominal wall defect with liver, stomach, intestinal loops, kidney, cardiac apex outside the body, thoracic hypoplasia due to thoracic aortic disease modification with pulmonary hypoplasia severe kyphosis and pronounced lumbosacral scoliosis. In this case membrane [20] is not visible in surrounding of abdominal viscera and the intestinal loop movement is not present during the observation of the fetus. The placental site was on the anterior wall of the uterus, but the foetus was laying face down to the posterior wall, with the abdominal viscera being in close contact to it, showing no change in its position or any active limb movements during the examination and making harder the evaluation, despite the normal amniotic fluid volume. The umbilical cord was not seen on Doppler examination, being occulted by the foetus and fixing it in that immobile position, spine back to the placenta, raising the suspicion of a short umbilical cord.

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Another finding from the article [11] decribes the Trisomy 9 syndrome is a rare chromosomal anomaly associated with multisystem dysmorphism and central nervous system (CNS) malformations. The literature describes more about the most common Dandy-Walker malformation (DWM). It is used to indicate the spectrum of abnormalities of the posterior fossa that are categorized using CT scans, such as classic DWM. Dandy-Walker variant is one of the variable hypoplasia[19] occurs in the cerebellar vermis, with or without enlargement of the posterior fossa. Megacisterna magna is an enlarged cisterna magna, which is unaffected cerebellar vermis and fourth ventricle. The clinical and sonographic findings that have been describe the CNS abnormalities, facial abnormalities, cranial abnormalities, congenital heart defects, skeletal defects, urogenital malformations, and intrauterine growth restriction. The article provides the prenatal ultrasound findings in a second-trimester diagnosis of a nonmosaic trisomy fetus with increased nuchal translucency in the first trimester

The finding from the article [12] describes the exact gestational age and expected day of delivery for providing optimal medical surveillance. Fetal biparietal diameter (BPD) is computed by ultrasound to correlate well with gestational age before 22 week. It can be used for detecting growth abnormalities, later in the pregnancy. A portable ultrasound scanner (the Umoja scanner) that can be used by midwives in LMIC countries with limited ultrasound and technological expertise. The important finding from the above research work provides the short- and long-term outcomes of children abnormalities from pregnancies with ultrasound examination. in most of the cases the children are affected by various abnormalities which occurs in limbs, nervous systems, heart, muscular disorders, brain development. The article [22] uses a detailed study about the polyhydramnios affected fetal with normal and detailed ultrasound analysis method. The normal AFI and detailed ultrasound examinations are provides the much better results compared to other methods which used by other researchers. In addition with the outcomes of ultrasound the obstetrics, genetic syndromes, and neurodevelopment were discussed in various literatures.

III.FINDINGS FROM SURVEY

An ultrasound image containing a contour of a fetal head is recorded with the prototype scanner. The image is preprocessed: converted to grayscale, gain adjusted, smoothed, dilated/eroded and finally binary threshold. The potential contour candidates and their Cartesian coordinates are identified by applying the canny edge detector. A line connecting the two most distant edge contours across the skull is computed. The original grayscale values along this line are used to identify the top and the bottom edge points which are used for measuring the BPD value. All image processing is performed using OpenCV (Open source Computer Vision), which is optimized for tablet devices. 27 ultrasound images suitable for BPD measurement were acquired by an experienced midwife and 9 student midwives with limited or no prior ultrasound experience, on 8 different fetuses from 18 to 34 weeks. Both manual and automatic BPD measurements were computed.

TABLE 1 ANOMALY DETECTION RATE

Anomalies	Total(N)	< 22 Weeks	>22 weeks
Structural	41	28	13
Chromosomal	38	24	14

The overall computation time on a Nexus 10 tablet was 3.47 seconds; The agreement of the proposed algorithm with the reference measurements is comparable to the inter observer agreement for BPD (2.6 to 3.1 mm from literature study). Table 1 depicts the chromosomal and structural anomaly detection rate of 250 fetuses. It is observed that the abnormalities the most affected babies are come under this chromosomal and structural anomaly defects. The detection of these abnormalities is based on before or after 22 weeks of observations. The prenatal and postnatal weeks are considered for the analysis and out of this the structural abnormalities has more no of defected babies when compared to chromosomal anomalies.

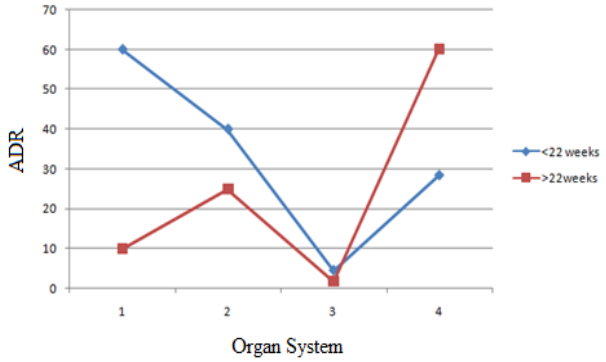

Figure 3 Anomaly detection rate versus organ system

The above figure depicts the graphical representation of prenatal detection rate along with the organ system in this the calculation made before and after 22 weeks because the prenatal duration is considered as 22 weeks of minimum time to analyze a fetus and its abnormalities.

TABLE 2 STRUCTURAL ANOMALY DETECTION RATE

Organ system	<22 weeks	>22weeks	Prenatal Detection Rate (%)
Central Nervous	60	10	70
Pulmonary	40	25	82.4
Cardiac	4.5	1.8	5.6
Other structural anomalies	28.4	60.2	38.4

It is observed that the prenatal detection rate varies for various organ systems and this data are taken from the articles which discussed in chapter III related works. The collected information is used to plot the details for error performance analysis and correlation. Table 2 depicts the structural anomaly detection rate for the 200 fetus out of which the majority of abnormalities found in central pulmonary anomalies.

IV.CONCLUSION

The application of ultrasound in finding the abnormalities is used to improve the care of pregnant women and their fetuses. Trimester scan provides the important information

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regarding plurality of the pregnancy, gestational age, cervical length, placental site, and assessment of fetal anatomy to detect fetal structural conditions. The different article identifies the various abnormalities and its possible ways to detected prenatally, the increasing ability to detect fetal structural conditions with ultrasound means that families can obtain important information about the nature of the condition so that they can make an informed decision regarding the future of the pregnancy. International expert groups have provided guidelines on first and second trimester FSA screening, as well as safety and good practice recommendations In this article various abnormalities and the methods to detect confounders including diabetes, pregnancies complicated with polyhydramnios, fetal macrosomia, congenital malformation, and genetic abnormalities, as well as neurodevelopment abnormalities and delay. Among this an important topic should be investigated further, particularly in prospective studies designed to overcome the limitations of the current scenario.

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