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Original Research Article

A study of fetal anomalies: contribution of ultrasonography and magnetic resonance imaging

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ABSTRACT

Background: Ultrasonography (USG) remains the primary modality for fetal imaging. Magnetic resonance imaging (MRI) is a suitable adjunct to USG. MRI is currently not used as a primary screening tool for antenatal period; however, it provides a reproducible fetal anatomy and can be more informative when the diagnosis on antenatal USG is inconclusive. Aim of the study was to study the contribution of antenatal USG and MRI in diagnosing fetal anomalies at a zonal hospital.
Methods: This was a prospective cross-sectional study enrolling the pregnant women between 13 to 35 weeks of gestation. The 30 consecutive fetuses suspected to have fetal anomaly on USG, were subjected to MRI after obtaining informed consent. Final diagnosis was made either radiologically (including follow up) or by clinical examination or autopsy.
Results: In 30 cases, 31 anomalies were detected on USG. There were 14 anomalies pertaining to central nervous system (CNS), 05 anomalies of genitourinary tract (GUT), 04 anomalies of thorax, 03 anomalies of gastrointestinal tract (GIT) and, 05 anomalies involving other body parts of fetus. MRI could detect 28 anomalies. USG was able to characterize a case of dorsal meningo-myelocoele better than MRI. MRI was able to show the extent of other anomalies better than USG and provided additional information of horseshoe shape in 01 case of multi cystic dysplastic kidneys. MRI could not detect 03 cases of single umbilical artery.
Conclusions: USG is the primary modality for fetal imaging. MRI can be a used as an adjunct to USG for confirmation and better delineation of anomalies.

Keywords: Fetal anomalies, Ultrasound, MRI

INTRODUCTION

USG is the primary imaging modality for fetal imaging during antenatal period. The detailed fetal anatomy evaluation is done during anomaly scan or level-II scan. This radiation free imaging modality is widely available, cost effective and comfortable for the patient and operator.[1] However, it has inherent limitations also. Small field of view (FOV), limited soft tissue contrast, significant attenuation of ultrasonic beam by increased adipose tissue in maternal abdomen (leading to poor visualization of fetal parts in obese ladies), suboptimal image quality due to associated oligohydramnios/anhydramnios, partial visualization of body parts due to inadequate fetal position or advanced gestational age, are some of the well-known limitations of USG.[1] MRI is an adjunct to USG for evaluation of pregnancy related maternal complications and also for the detailed assessment of fetal anomalies.[2] Fetal MRI has limited use in early gestational age due to the small fetus.[3] However, MRI can be performed in any trimester as long as the referring doctor and the radiologist are able

to justify the purpose.[4] The safety for 3 Tesla MRI for fetal imaging has not been established yet.[3] For obstetrics imaging, fast MRI sequences are obtained due to moving fetus.[5] In present study, antenatal USG and MRI findings were correlated with the postnatal/ post abortal examination of newborn/fetus. The additional contribution of MRI was also studied.

METHODS

This was a prospective study conducted at Base hospital, Guwahati, between Sep 2018 and Aug 2020 after prior approval from hospital ethical committee. 30 consecutive pregnant women, between 13 to 35 weeks of gestation, who were detected/suspected to have fetal anomaly on 2D gray scale USG, were enrolled in this study. The obstetrical sonogram for anatomic survey of fetus in second/third trimester included the evaluation of fetal head (ventricles, falx, cavum septum pellucidum, choroid plexus, cerebellum, cisterna magna), orbits, upper lip, neck, four-chamber view of the heart and ventricular outflow tracts, stomach, kidneys, urinary bladder, umbilical cord insertion site with number of vessels, whole spine and all four limbs. A targeted anatomic USG examination was performed on case-to-case basis. There were 25 pregnant women with gestational age between 13 to 20 weeks and rest 05 pregnant women were between 21 to 35 weeks gestation. After informed consent, all these women were subsequently subjected to MRI within two days of USG. 25 pregnancies were followed up in 3rd trimester. Subsequently, all 30 cases were followed up postnatally. The follow-up was available for 29 cases and 01 case of posterior urethral valves lost follow up postnatally. Final postnatal diagnosis was made to either radiologically or clinically or on autopsy. For imaging, the 2D gray scale USG was performed on Logic P5 from General Electric (GE) and curvilinear multi frequency probe (3.5MHz) was used in all cases. MRI was done with 1.5 Tesla Philips Achieva after informed consent. Pregnant women were positioned in head-first supine and phased array coil was used for abdomino-pelvic region. Fast T2 sequences in sagittal, coronal and axial planes of fetus. The sequences were obtained in oblique planes also depending upon the location and plane of the fetal anomaly. The slice thickness was kept at 5mm with zero gap. No intravenous gadolinium contrast or any other drug was administered to the pregnant women prior to MRI scans.

Statistical analysis

The findings were tabulated and analyzed. The categorical data was expressed as rates, proportions and percentages and descriptive analysis was done.

RESULTS

In the present study, total 30 pregnancies were enrolled with age range from 22 to 32 years and mean age of 26.7 year. The gestational age range was between 13 weeks to 35 weeks. Total 31 fetal anomalies were found. There were 14 (45.1%) anomalies related CNS, followed by the 05 anomalies of GUT (16.1%), 04 anomalies of thorax (12.9%), 03 anomalies of GIT (9.6%) and 05 (16.1%) anomalies were categorized as miscellaneous (Table 1). There were 04 Chiari-2 malformations, 02 anencephaly (Figure 1) cases, 01 occipital encephalocele (Figure 2), 01 iniencephaly, 06 cases of choroid plexus cysts, 01 case of agenesis of left kidney, 01 bilateral multi cystic dysplastic kidneys with horseshoe shape (Figure 3), 01 bilateral hydronephrosis, 01 right hydronephrosis, 01 posterior urethral valves, 02 congenital diaphragmatic hernia, 01 congenital cystic adenomatoid malformation (CCAM) (Figure 4), 01 pulmonary sequestration, 02 omphaloceles, 01 enteric duplication cyst (Figure 5), 01 bilateral club feet, 01 cystic hygroma and 03 cases of single umbilical artery (Table 2).

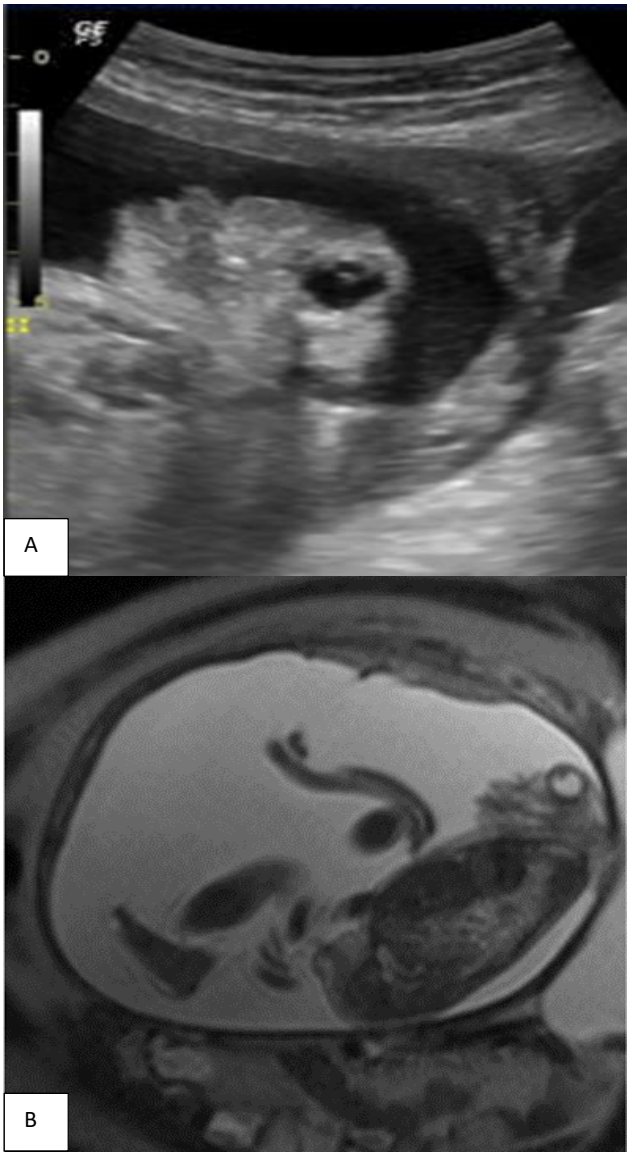

Figure 1 (A and B): Anencephaly, midline longitudinal USG image shows lack of the brain tissue above the orbits. Corresponding sagittal MRI fast T2 image shows anencephaly.

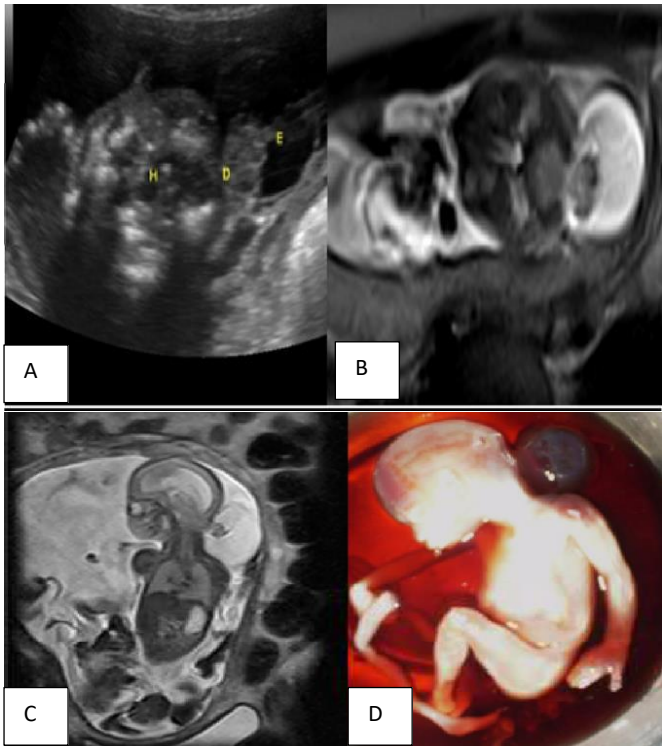

Figure 2 (A-D): Occipital encephalocoele. Transverse USG image shows fetal head (H) with a defect (D) in occipital bone and encephalocoele (E) posteriorly. Axial and oblique sagittal MRI fast T2 images shows occipital encephalocoele with defect in the occipital bone. Post abortal photo of the same fetus.

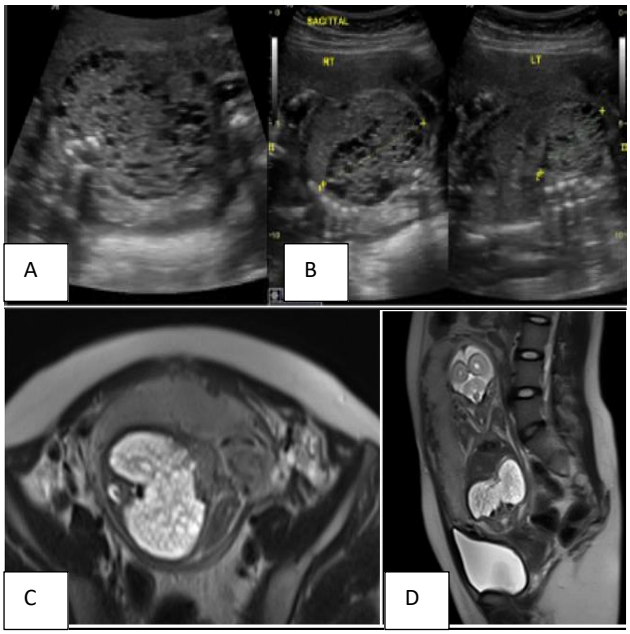

Figure 3 (A-D): Bilateral dyplastic kidneys with horseshoe configuration. Transverse and oblique sagittal images of USG bilateral enlarged and echogenic kidneys with multiple anoechoic cysts. Axial and coronal MRI fast T2 images show bilateral enlarged, heterogeneously hyperintense kidneys with fusion medially.

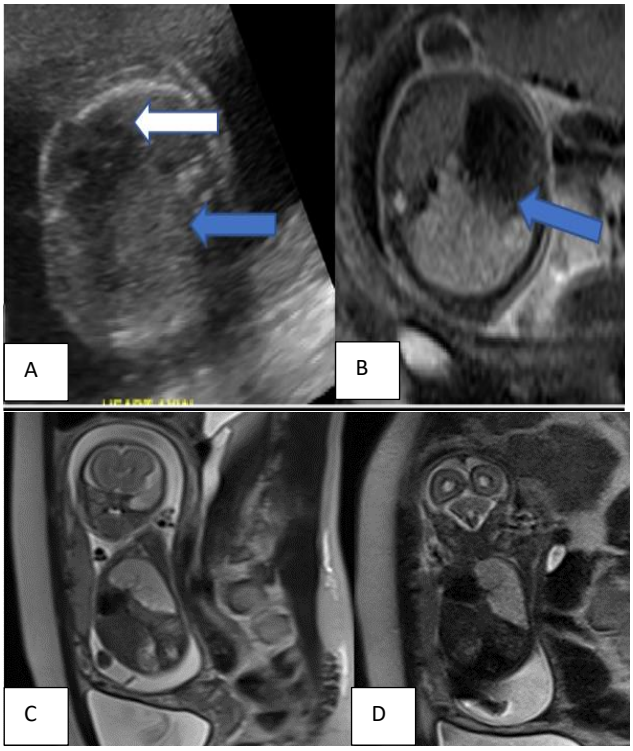

Figure 4 (A-D): CCAM. Transverse USG image showing echogenic right lung (solid blue arrow) with cardia shifted to left (white arrow). Corresponding axial MRI fast T2 image showing hyperintense right lung (blue arrow) with cardia being shifted to left side. Coronal MRI fast T2 images showing hyperintense and enlarged right lung.

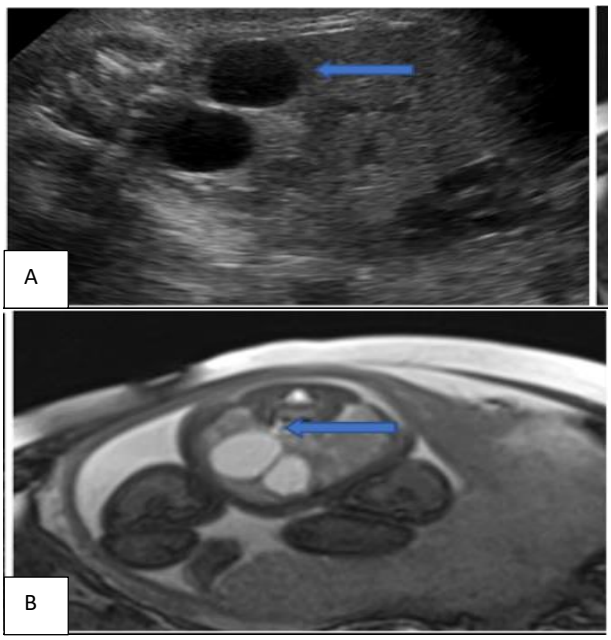

Figure 5 (A and B): Enteric duplication cyst. Transverse USG image shows a cystic lesion (blue arrow) in close relation to urinary bladder. No communication was found with bladder. Axial MRI fast T2 shows cystic lesion (blue arrow) posterolateral to urinary bladder.

Table 1: System wise anomalies.

System affected	Number	Percentage (%)
CNS	14	45.1
GUT	05	16.1
Thorax	04	12.9
GIT	03	9.6
Miscellaneous	05	16.1
Total	31	100

Table 2: Fetal anomalies.

Type of anomaly	Number
Chiari-2 malformation	04
Anencephaly	02
Occipital encephalocele	01
Iniencephaly	01
Choroid plexus cysts	06
Agenesis of left kidney	01
Bilateral multi cystic dysplastic kidneys with horseshoe shape	01
Bilateral hydronephrosis	01
Right hydronephrosis	01
Posterior urethral valves	01
Congenital diaphragmatic hernia	02
CCAM	01
Pulmonary sequestration	01
Omphalocele	02
Enteric duplication cyst	01
Bilateral club feet	01
Cystic hygroma	01
Single umbilical artery	03
Total anomalies	31

Distribution of CNS anomalies

In the present study, there were total 14 central nervous system anomalies as shown in the Figure 6: 04 cases of Chiari-2 malformation, 02 cases of anencephaly, 06 cases of choroid plexus cysts (size range 5 mm to 9 mm in maximum diameter), 01 case of occipital encephalocele and 01 case of iniencephaly. One case of dorsal spine meningo-myelocoele was better characterized on USG than magnetic resonance imaging. The communication of lesion with spinal canal and presence of thin neural tissue within strands were appreciated well on USG and magnetic resonance imaging could only detect the midline sac. Post abortal diagnosis was confirmed on autopsy. During 3rd trimester follow up, 04 choroid plexus cysts resolved completely. The two cases of choroid plexus cysts were followed up postnatally. The pregnancy was terminated in rest all cases of central nervous system anomalies.

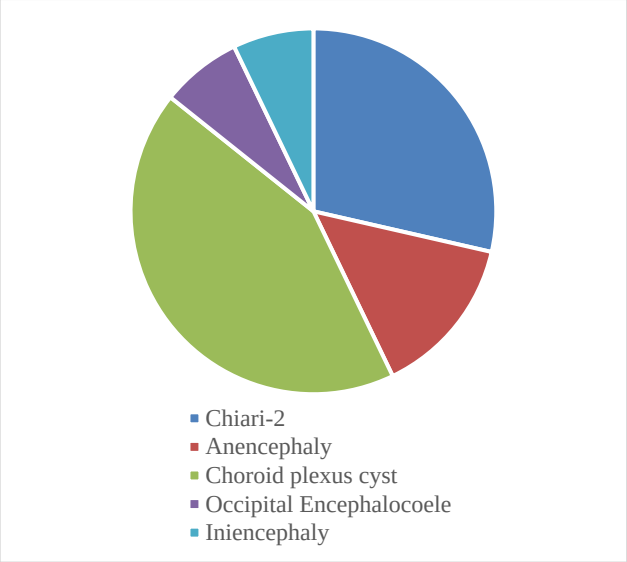

Figure 6: CNS anomalies.

Distribution of GUT anomalies

There were 05 anomalies (Figure 7) involving GUT: 01 case of agenesis of left kidney, 01 case of bilateral multi cystic dysplastic kidneys along with horseshoe shape, 01 case of bilateral hydronephrosis, 01 case of unilateral (left) hydronephrosis and 01 case of posterior urethral valve. MRI confirmed absence of left kidney in left sided renal agenesis case. In bilateral multi cystic kidneys case, MRI confirmed the findings of USG and horseshoe shape were additionally diagnosed and pregnancy was terminated in this case and cases of bilateral and unilateral hydronephrosis was followed up postnatally. Case of absent left kidney was followed up postnatally. Bilateral hydronephrosis case showed partial resolution postnatally. Unilateral hydronephrosis persisted postnatally and case was advised follow up. Posterior urethral valve neonate lost to follow up after discharge from hospital.

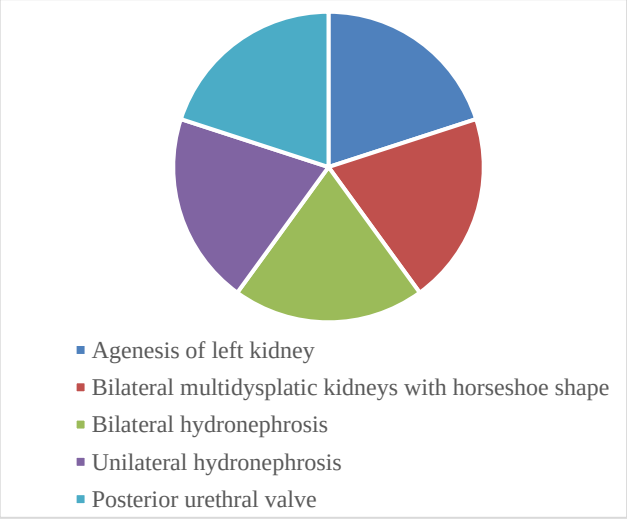

Figure 7: GUT anomalies.

Distribution of thoracic anomalies

In present study, there were 03 anomalies pertaining to thorax: 01 case of congenital diaphragmatic hernia, 01 case of pulmonary sequestration and 01 case of CCAM (Figure 8). Pregnancy was continued in all 03 cases and final diagnosis was based on radiological findings.

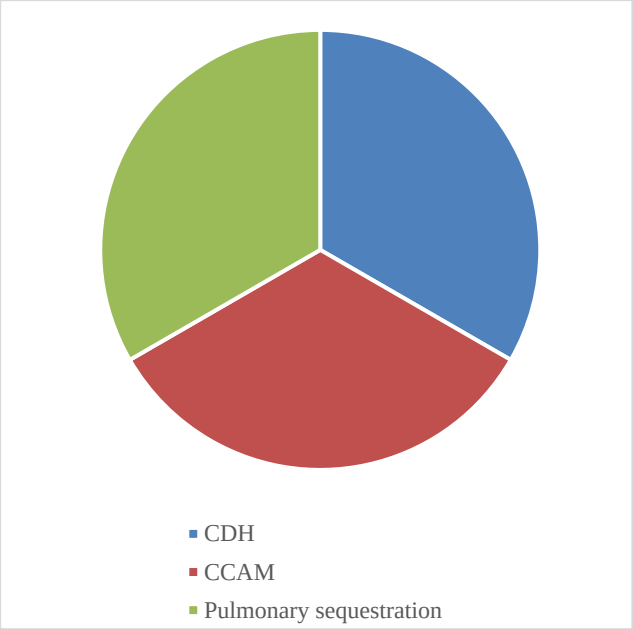

Figure 8: Thoracic anomalies.

Distribution of GIT anomalies

There were 02 cases of omphalocoeles and 01 enteric duplication cyst. Herniation of intestine was found in both cases of omphalocoele and pregnancy was terminated in these two cases. Pregnancy was terminated in both cases of omphalocoele. Gross examination and autopsy confirmed the findings. The enteric duplication cyst was followed up postnatally and the neonate was asymptomatic.

Miscellaneous anomalies

Bilateral club feet (01), cystic hygroma (01) and 03 cases of single umbilical artery were found. Pregnancy was terminated in cystic hygroma case and autopsy confirmed the diagnosis. No other anomaly was detected in club feet case postnatally. Neonatal abdominal USG was done for all single umbilical artery cases and no other anomaly could be found.

DISCUSSION

First use of USG in obstetrics and gynecology is found in classic Lancet paper published by Donald et al wherein the first ultrasound images of fetus and some masses of gynecological origin could be seen.[6] Even today, despite advancements in the field of radiology over last few decades, the role of USG in antenatal imaging cannot be overemphasized. USG provides a real time imaging which is more valuable in evaluation of fetal cardia, dynamic imaging of fetal movements and breathing. Colour and spectral doppler imaging is also an integral part of USG equipment. The widespread use of MRI in obstetrics dates back to 1980s.[7] As per Levin et al MRI can be used in pregnancy whenever the diagnosis is doubtful on USG.[3] Benson et al are of opinion that MRI is now the modality of choice whenever the diagnosis on USG is inconclusive.[7] MRI is helpful whenever the visualization of fetus is suboptimal due to its inherent limitations of USG. MRI can increase the accuracy of antenatal imaging and hence helping in management of pregnancy.[8] At present, there are multiple studies in literature on contribution of prenatal USG and MR imaging. It is shown that MRI provides additional details to USG in 36% to 57% cases for CNS anomalies.[5,9] However, this benefit of MRI is not documented for non-CNS anomalies.[1] MRI helps in detecting and characterising the fetal CNS anomalies better than USG, particularly during third trimester.[10] MRI can also be used to estimate fetal lung volume in cases like congenital pulmonary airway malformation (CPAM), congenital diaphragmatic hernia and bronchial atresia.[11] MRI has also has role in pregnancy with suspected appendicitis, some other gastrointestinal conditions, hepatobiliary and genitourinary related abnormalities.[12,13]

Sohn et al conducted a study on 56 fetuses, the CNS anomalies were found in 26 fetuses (46.4%), abdominal anomalies in 17 (30.3%), thoracic anomalies in 6 (10.7%), head and neck anomalies in 5 (8.9%) and other anomalies in 2 (3.5%) fetuses.[14] Kapoor et al conducted a study on north-western population of India and found that the incidence of CNS anomalies was maximum (32.6%), followed by gastrointestinal tract anomalies (32%), musculoskeletal anomalies (21%) and genitourinary anomalies (17%).[15] In present study, the CNS related anomalies were maximum in number (n=14, 45.2%), followed by the GUT (n=05,16.1%), thorax (n=4, 12.9%) and GIT (n=03, 9.7%). There were 05 (16.1%) anomalies categorized as miscellaneous in this study. No musculoskeletal anomaly was found in present study.

In an observational study by Whitby et al on 21 antenatal women with suspected fetal CNS anomalies on USG, the USG and MRI results could agree only in 28.6% cases. MRI provided additional information in 23.8% cases.[16] In present study, out of 14 CNS anomalies, 01 (7.1%) case of dorsal spine meningo-myelocoele was better characterized on USG than MRI. In rest 92.9%, the USG and MRI were in agreement and MRI provided the extent of lesions better.

In a study by Hosny et al, 25 pregnant women, detected to have fetal anomalies on USG, underwent MRI.[17] In their study, MRI scan changed the diagnosis in 8% (2) cases and provided additional information in a case of occult spinal diastematomyelia. In 72% (18) cases, the

MRI findings matched USG. In 39.1% cases, the US findings and MRI findings were in agreement. In a study by Hamisa et al on 23 pregnant women suspected to have fetal CNS anomalies, it was found that MRI changed the diagnosis in 14 (60.8%) cases and provided additional information in 2 (8.6%) cases.[18] In present study of 14 cases of CNS anomalies, the USG and MRI agreed in 92.8% (n=13) cases. In 01 (7.1%) case of upper dorsal meningo-myelocoele, the diagnosis of USG was confirmed on autopsy, hence, USG was better than the MRI.

Levine et al found that MRI provided additional information in both the central nervous system in 10 (55%) of 18 fetuses.[19] However, in present study, USG provided more information in 01 case of spinal meningocele (dorsal) due to higher resolution.

A study by Frates et al on 28 fetuses, the diagnosis by MRI was incorrect in 04 cases and USG diagnosis was correct in these 04 cases. In present study, the MRI imaging failed to detect 03 cases of single umbilical artery.[5]

In the study by Sohn et al on 56 fetuses, 03 fetuses with suspected intracranial abnormalities on ultrasonography were diagnosed as normal by fetal MRI and also during postnatal follow up.[14] However, in present study, there was no false positive diagnosis by USG.

The study by Cassart et al on 16 fetuses with suspected renal anomalies suggested that MRI can accurately show the urinary tract anomalies during 3rd trimester.[20] In their study, a fetus with suspected unilateral renal agenesis was found to have bilateral agenesis on MRI. Behairy et al conducted a study on 30 fetuses with sonographically suspected congenital urinary tract anomalies.[21] MRI changed the diagnosis in 06 cases and provided additional information in 04 cases. In present study, MRI confirmed the findings in 01 case of agenesis of left kidney. In 01 case of suspected bilateral multicystic dysplastic kidneys, MRI confirmed the anomaly and also, provided additional findings of horseshow shape fusion of both kidneys.

In present study, the USG was able to detect all anomalies. MRI was able to confirm the anomalies and hence providing the more confidence to the gynecologist to decide the fate of pregnancy.

Limitations

Since MRI was done only for suspected/confirmed cases of fetal anomalies, so, the actual comparison of sensitivity between USG and MRI for detection of fetal anomalies could not be done.

CONCLUSION

Ultrasound is the well accepted and primary imaging modality for anomaly scan. Additional use of colour doppler makes the USG an indispensable tool for fetal evaluation throughout the pregnancy. USG with colour doppler can easily detect presence of single umbilical artery that further leads the radiologist for targeted anomaly scan to search for other associated anomalies, if any. Role of MRI in pregnancy is to confirm or exclude or further define the anomalies detected or suspected on USG. Both of these imaging modalities are complementary in reaching a final diagnosis. Antenatal MRI can facilitate management decisions during pregnancy since it provides more confidence to the gynecologist in deciding fate of pregnancy. Also, from present study, it is also proved that MRI cannot fully replace USG for antenatal scans.

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