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REVIEW ARTICLE

The Role of 2D and 3D Ultrasound in Evaluation of Fetal Gastrointestinal Anomalies

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ABSTRACT

Aim: To evaluate the role of second mid-trimester ultrasound in prenatal detection of gastrointestinal (GI) fetal anomalies and compare the ultrasonographic findings with postnatal diagnosis.

Materials and methods: A 5-year retrospective study included 16,334 neonates delivered at a tertiary referral center. All neonates were evaluated by a second mid-trimester 2D ultrasound fetal anatomy scan. Patients with abnormal findings on 2D scan were also examined by 3D ultrasound. Postnatally confirmed GI anomalies were compared with prenatal ultrasound assessment of two sections of fetal abdomen which had analyzed the presence, size and position of the stomach, umbilical cord insertion and have assessed the amniotic fluid index (AFI).

Results: Prenatal ultrasound revealed 28 out of 38 fetal GI anomalies (73.6%). All GI anomalies initially diagnosed with 2D ultrasound were confirmed by 3D ultrasound. The major advantage of multiplanar imaging was more comprehensive anatomical information about GI anomalies. Surface rendering provided additional information in evaluating fetuses with anterior abdominal wall defects.

Conclusion: Our data indicate that standard planes obtained by 2D ultrasound can rule out a majority of fetal GI anomalies. Assessment of AFI should be an integral part of prenatal ultrasound scan in detection of GI anomalies, particularly in GI obstruction.

Keywords: GI anomalies, Esophageal and duodenal atresia, Pyloric stenosis, Hirschsprung disease, Small bowel/anal/rectal atresia, Diaphragmatic hernia, Omphalocele and gastroschisis, 2D and 3D ultrasound.

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INTRODUCTION

Congenital malformations of the gastrointestinal (GI) tract have been a significant source of morbidity and mortality in the newborn, whether associated with congenital syndromes or as isolated malformations. Some 20 to 30% of perinatal deaths can be attributed to various congenital anomalies.[1] Prenatal diagnosis of GI malformations, as well as other congenital anomalies, allows to plan the postnatal treatment as well as to provide information on fetuses with anomalies that are incompatible with life. While there have been various reports of efficacy in prenatal diagnosis of congenital anomalies, the practice guidelines issued by the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG), recommend a routine ultrasound by a trained ultrasonographer between 18 and 22 weeks of gestation for dating and evaluation of major congenital anomalies.[2] Due to development of 3D/4D US there is a tendency toward earlier diagnosis of life-threatening congenital malformations from the second to the first trimester of pregnancy.[3]

The objective of this study was to determine the success of second mid-trimester ultrasound examination in diagnosis of fetal GI anomalies.

One of the most common GI anomalies in the neonate is a congenital diaphragmatic hernia (CDH), which has a prevalence of approximately one in 3000 live births.[4] Prenatal diagnosis can allow obstetricians and pediatricians to be prepared for resuscitation in the immediate postpartum period. Abdominal wall defects, such as omphalocele and gastroschisis have a prevalence of one in 5000 live births for each anomaly. These abdominal wall defects can be diagnosed at 22 to 28 weeks gestational age and provide the opportunity for counseling pregnant patients on the course of treatment of the congenital anomaly as well as prognosis and associated chromosomal anomalies or malformations.[5] Gastrointestinal atresia and obstruction represent a significant portion of GI anomalies. Duodenal atresia occurs in one of 10,000 live births.[6] It has been shown that prenatal diagnosis of obstruction leads to earlier postpartum confirmation and decreased morbidity and complications.[7]

The successes of studies which examine the usefulness of prenatal ultrasonography in diagnosing congenital anomalies vary greatly. These variations have been attributed to the level of training to scan and interpret obstetric ultrasounds. In this study, we have also compared our results with those

of other studies which have examined the prenatal diagnosis of GI anomalies.

MATERIALS AND METHODS

A 5-year retrospective study included 16,334 neonates delivered at a tertiary referral center. The protocol was approved by the Institutional Review Board and Institutional Ethics Committee. All the neonates were evaluated by a second trimester fetal anatomy scan by 2D ultrasound. Patients with abnormal findings were assessed by 3D ultrasound, performed by a fetal imaging specialist. Commercially available equipment for 2D ultrasound was Aloka 5000 (Aloka Co, Tokyo, Japan). Voluson E8 Expert was used for 3D ultrasound imaging (GE, Riverside, CA, USA). Transabdominal 3.5 and 5 MHz or endovaginal 7.5 MHz transducers were used to obtain 2D and 3D images. For acquisition of 3D ultrasound volumes the transducer was held stationary, while mechanical sweep through fetal abdomen and/or thorax was obtained. Depending on the size of the object of interest, the acquisition time ranged from 2 to 6 seconds. In the case of fetal movement, the acquisition was repeated. Between two to five volumes were obtained per GI anomaly. Equipment used for postnatal imaging was Aloka SS1000 (Aloka, Tokyo, Japan).

Postnatally confirmed GI anomalies were compared with prenatal ultrasound assessment of two sections of fetal abdomen. The presence, size and position of the stomach, umbilical cord insertion and the AFI were analyzed.

RESULTS

A 5-year retrospective study included 16,334 neonates delivered at a tertiary referral center. There were 38 anomalies of the fetal GI system diagnosed postpartum, five neonates with esophageal atresia, two with duodenal atresia, one with pyloric stenosis, one with annular pancreas, two with Hirschsprung disease, three with small bowel atresia, two anal and one rectal atresias. Eleven neonates were diagnosed with diaphragmatic hernia, five had gastroschisis and five were diagnosed with omphalocele. All the neonates were previously evaluated mostly by a second mid-trimester fetal anatomy scan, because they were referred in the 2nd trimester of pregnancy by the providing physician to confirm the diagnosis. Two sections of fetal abdomen have been carefully analyzed for the presence, size and position of the stomach, continuity of the diaphragm and assessment of AFI. Prenatal scans revealed 28 out of 38 congenital anomalies of the fetal GI system, with a 73.6% success rate (Table 1). Five patients had esophageal atresia associated with trachea-esophageal fistula and five fetuses had bowel obstruction. The latter included two fetuses with small bowel obstruction, one with Hirschsprung disease and two with anorectal malformations which remained undiagnosed before delivery (*see* Table 1).

Table 1: Anomalies of gastrointestinal tract diagnosed in neonatal period and prenatally not diagnosed by ultrasound

Gastrointestinal anomaly	*Diagnosed postnatally in neonatal period*	*Not prenatally diagnosed by ultrasound*
	Number of patients	
Esophageal atresia with tracheo-esophageal fistula	5	5
Duodenal atresia	2	0
Pyloric stenosis	1	0
Annular pancreas	1	0
Hirschsprung disease	2	1
Small bowel atresia	3	2
Anal atresia	2	1
Rectal atresia	1	1
Diaphragmatic hernia	11	0
Omphalocele	5	0
Gastroschisis	5	0
Total	38	10 (26.4%)

All GI anomalies initially diagnosed with 2D ultrasound were confirmed by 3D ultrasound. The major advantage of multiplanar imaging by 3D ultrasound was more comprehensive anatomical information about GI anomalies, especially in patients with diaphragmatic hernia and obstructive intestinal anomalies to determine the level of the defect. Surface rendering provided additional information in evaluating fetuses with defects of anterior abdominal wall.

DISCUSSION

Of 16,334 neonates included in the study, 38 (0.2%) were diagnosed with GI anomalies; 28 were diagnosed prenatally by ultrasound (73.6%). Ten GI anomalies were not diagnosed by second mid-trimester ultrasound. Comparison of our results with earlier published prenatal ultrasound studies are exemplified in Table 2.

Some previous publications which have examined the diagnosis of duodenal atresia and GI obstruction were comparable with our results. Studies in Table 2 were performed during the period 1987 to 2012.[6-12] It was shown that neonates prenatally diagnosed with duodenal obstruction needed less readmission to the hospital after surgical correction than their counterparts who were not diagnosed prenatally. This has demonstrated the need for increased efficacy and vigilance in detecting GI anomalies which need to be corrected early postpartum for better outcomes.[7]

One major aspect of diagnosis of GI anomalies by fetal ultrasound is the timing at which the anomaly can be detected

Table 2: Comparison of our results with other obstetrical ultrasound studies performed in mid-second trimester or detection of gastrointestinal congenital anomalies

Study group	*Year of study*	*Expertise of imager and/or interpreter*	*Successful prenatal diagnosis (%)*	*Types of anomaly*
Our study	2007-2012	Tertiary referral center	73.6	GI anomalies
Choudhry[6]	1995-2004	Routine sonographer	45	Duodenal atresia
Cohen-Overbeek[7]	1991-2003	Unspecified	27.5	Duodenal obstruction
Hausler[9]	1996-2000	Skilled operators (varying levels)	34	GI obstruction
Kaasen[11]	1988-2004	Consultants in fetal medicine	60	GI anomalies
Romosan[12]	2000-2005	Specially trained midwives	29	GI anomalies
RADIUS[8]	1987-1991	Registered diagnostic medical sonographers	34.8	Assessment of major congenital anomalies

based on fetal development. While some studies have noted that obstructive anomalies cannot be diagnosed until later in pregnancy (>24 weeks), because the fetus swallows smaller amounts of fluid in early pregnancy, compared with later pregnancy.[7] Others have shown the ability to diagnose duodenal atresia and obstruction early, at or before 20 weeks.[6,9] Esophageal atresia with trachea-esophageal fistula is not easy to be diagnosed prenatally like in our cases, while if tracheo-esophageal atresia is not present with esophageal or duodenal atresia than prenatal diagnosis can be made in the first and for sure in the second trimester of pregnancy.[13,14] Prenatal diagnosis of imperforate anus, anal and/or rectal atresia and Hirschsprung disease is always puzzling by ultrasound either in the first or in the second trimester of pregnancy.[15-19]

Additionally, a wide range of efficacy in ultrasound detection of obstructive GI anomalies has been shown (0-56%), which may be attributed to various policies for prenatal ultrasound.[9] The surveillance of the amniotic fluid volume by ultrasound is an important additional instrument in detection of fetal GI anomalies. However, an excessive accumulation of amniotic fluid is not only suggestive of fetal GI anomalies, but may occur with other maternal [infections (TORCH), diabetes mellitus], fetal (cerebral, pulmonary, urogenital malformations, congenital cardiac defects, fetal tumors, chromosomal disorders, immunologic or non-immunologic hydrops) or placental conditions. Therefore, in patients with polyhydramnios an astute sonographer should look for anatomic malformations causing disability of swallowing, such as cleft lip, cleft palate, cerebral or neuromuscular deficiencies, or a blockage of the fetal GI tract such as esophageal, duodenal or intestinal stenosis or atresia. In our study, all ten fetuses with undiagnosed GI anomalies had mild to severe polyhydramnios.

Some authors have not consistently measured the same parameters in fetal ultrasound. Our study has examined various GI anomalies including abdominal wall defects, obstruction, and hernias. In one study, the correlation of fetal anomalies detected by ultrasound and the results discovered on autopsy after second trimester termination were analyzed.[11] There was a 60% agreement between ultrasound and autopsy results. The correlation decreased when the number of associated anomalies increased.

In one study, only 29% of GI anomalies were diagnosed prenatally (4/13).[12] The methodology in that study differed from our study in that they have included routine prenatal ultrasound by midwives and referral of patients to an obstetrician experienced in ultrasound, whereas in our study ultrasounds were performed at the tertiary referral center by trained obstetricians and fetomaternal specialists.

The routine antenatal diagnostic imaging with ultrasound trial (RADIUS) showed comparable adverse outcomes between the group which received two screening ultrasounds during pregnancy and a control group which had only one US examination (4.8 and 4.7% respectively),[8] and consequently questioned the efficacy of prenatal screening ultrasound. They have indicated that ultrasound diagnosis of fetal anomalies during pregnancy did not alter the prevalence of adverse outcomes. Such results attributed this to the fact that 52% of the detected anomalies were identified after 24 weeks gestation and were not legal to terminate. At the time of this study (1987-1991) the agreement of prenatal ultrasound and postnatal findings was 34.8%. However, more recent studies have demonstrated that increased efficacy of prenatal ultrasound diagnosis resulted in more efficient planning for correction, immediate neonatal care and decreased hospital admissions.[6,11,20] Our results show increased efficacy (73.6%) of prenatal diagnosis of GI anomalies, which provided the opportunity to plan for anomalous births and postpartum intervention.

Presently, 2D ultrasound is the most used modality in prenatal screening for anatomical development and congenital anomalies. Table 3 provides the comparison of 2D ultrasound, 3D ultrasound, color Doppler ultrasound and

Table 3: Advantages and disadvantages of prenatal imaging modalities

Imaging modality	*Advantages*	*Disadvantages*
2D ultrasound	• Cost-effective • Safe • Able to detect congenital fetal anomalies	• No consistent demonstration of reduced morbidity/ mortality • Success in prenatal diagnosis varies among institutions and experience of sonographers and interpreters
3D ultrasound	• Multiplanar imaging and surface rendering provide additional information • Better estimation of fetal weight • Better estimation of volume of amniotic fluid	• Requires expertise • Unclear evidence of increased diagnosis of congenital anomalies
Color Doppler ultrasound	• Ability to study blood flow and impedance in umbilical cord	• No indications or timing delineated for use • Unclear evidence of increased diagnosis of GI congenital anomalies
MRI	• High quality • Able to detect subtleties	• Expensive • Safety has not been extensively studied • Longer examination time • Unavailability of equipment/expertise

MRI in diagnosing of fetal developmental anomalies. 2D ultrasound remains also the most cost-effective means for determining anomalies, but has not been consistently shown to affect neonatal morbidity and mortality. Moreover, screening ultrasounds are not standardized and the results and efficacy depends mostly on the experience in performing and interpreting of ultrasound. Three-dimensional ultrasound provides visualization of the fetal surface as well as more accurate volume and weight measurements, however, 2D ultrasound has been shown to be in agreement with 3D- and 4D-ultrasonography up to 90.4% and added benefit has not been effectively demonstrated.[20] Multiplanar imaging derived from volumetric data improves optimization of the anatomical display.[21] However, 3D ultrasound images provide additional information in 51% of fetal anomalies, they were equivalent to 2D ultrasound in 45% and were disadvantageous in about 4%.[13] Similar to our results, these authors found that the rendered images were helpful in counseling and improving of understanding of the patients about fetal anomalies, while multiplanar imaging was more helpful in diagnostic purposes. Another advantage of 3D ultrasound is that the volume data can be stored on a removable hard disk or decompressed and sent by internet, allowing for telemedicine review by maternal fetal specialist, neonatologist and/or pediatric surgeon. Color Doppler ultrasound provides information on blood flow and impedance in the umbilical cord, which can assist in the assessment of intrauterine growth restriction and fetal developmental anomalies. However, its use is not standardized for care. The current guidelines recommend against color Doppler ultrasonography for low risk pregnancies.[2] Similarly, magnetic resonance imaging (MRI) is not presently used for screening purposes in low risk populations.[21] The benefits of MRI include higher quality imaging, but it remains expensive and its effect on the fetus has not been consistently demonstrated.[21]

With increasing standardization of fetal ultrasound diagnosis and availability of specially trained sonographers has affected increasing prenatal detection of fetal GI anomalies. Our study shows promising results of routine second mid-trimester prenatal scan in ruling out major GI anomalies such as diaphragmatic hernia, obstruction, atresia, and abdominal wall defects. Our data indicate that 3D ultrasound can be used in adjunct to standard 2D ultrasound imaging to confirm the findings and provide additional anatomical information in multiplanar and surface rendering display.[3] Rendered images are helpful in the assessment of frontal abdominal wall defects, and are appreciated by parents for educational purposes and better understanding of the severity of fetal anomaly.

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