Original Research Article

Role of ultrasound in diagnosis of fetal congenital abdominal anomalies: One year prospective study

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Abstract

Introduction: Congenital anomalies are important cause of perinatal and infant mortality, and are contributor to childhood morbidity. Fetal congenital abdominal anomalies are one of the most common anomalies. Ultrasound screening of fetus helps to identify these anomalies as early as possible in antenatal period and thus provides information for proper decisions during pregnancy. **Aim:** To find out the incidence of congenital abdominal anomalies affecting fetus and to study the outcome in patients with fetal congenital abdominal anomalies by following them postnatally. **Material and Methods:** Prospective, Observational and Non-interventional study. Ultrasound screening of 5761 fetuses were performed. Pregnancies, with ultrasound findings of fetal congenital abdominal anomalies with or without other associated anomalies, were followed up. Postnatal confirmation of anomalies were carried out by clinical examination/ imaging procedures / autopsy. **Results:** Incidence of fetal congenital abdominal anomalies was 5.56 per 1000 births. Among the fetal congenital abdominal anomalies, fetal urinary tract anomalies were the most common with an incidence of 4.69 per 1000 births. Incidence of Omphalocele, Gastroschisis and Jejunoileal atresia were 5.2, 1.7 and 1.7 per 10,000 births respectively. Concordance between antenatal and postnatal diagnosis was 90.63%. **Conclusion:** Antenatal ultrasound examination should be performed for all pregnant women to diagnose congenital anomalies. Prenatal diagnosis of congenital anomalies provides information for proper decisions during pregnancy, fetal intervention if available and appropriate treatment perinatally, thus improves perinatal and long term outcomes.

Keywords: Fetal congenital abdominal anomalies, Fetal urinary tract anomalies, Omphalocele, Gastroschisis.

Introduction

Advances in medicine have led to decline in diseases like infections and malnutrition, so the congenital malformations have emerged gaining great importance in perinatal mortality [1]. Congenital anomalies account for 8–15% of perinatal deaths and 13-16% of neonatal deaths in India [2,3]. The pattern and prevalence of congenital anomalies may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables [4]. Ultrasound is the non-invasive screening modality of choice for the detection of congenital anomalies because of its safety, cost effectiveness and detection sensitivity. Incidence of antenatally diagnosed congenital anomalies is increasing as compared to past because of advanced diagnostic facilities and routine antenatal ultrasound examination should be performed for all pregnant women to diagnose these anomalies. Prenatal diagnosis of congenital anomalies provides information for proper decisions during pregnancy, fetal intervention if available and appropriate treatment perinatally (timed delivery in tertiary care centres), thus improves perinatal and long term outcomes [5].

Anterior abdominal wall defects comprising omphalocele, gastroschisis, and bladder exstrophy remain a source of significant morbidity and mortality [6]. While esophageal atresia, duodenal

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atresia, jejunoileal atresia and anorectal atresia are the common gastrointestinal tract anomalies. Urinary tract anomalies account for 33% of all malformations detected by routine prenatal sonography [7]. Renal agenesis, Renal ectopia, Horseshoe kidney, Multicystic dysplastic kidney, Cystic Renal Dysplasia with Obstruction, Autosomal recessive polycystic kidney disease (ARPKD), Autosomal dominant polycystic kidney disease (ADPKD), Hydronephrosis and fetal megacystis are commonly diagnosed urinary tract anomalies during antenatal period.

Aim of the Study: To find out the incidence of congenital abdominal anomalies affecting fetus and to study the outcome in patients with fetal congenital abdominal anomalies by following them postnatally.

Material and Methods

This was a hospital based prospective, observational and non-interventional study, conducted in Department of Radio diagnosis in collaboration with Department of Obstetrics and Gynaecology at Rajindra Hospital, Patiala. Study was conducted for a period of one year from 1st Jan 2015 to 31st Dec 2015. Pregnant cases with gestational age ≥10 weeks undergoing routine antenatal screening having congenital abdominal anomalies with or without other associated anomalies were included. Pregnant cases with gestational age <10 weeks were excluded. Institutional review board approval for conducting this study was obtained and informed written consent for participation in this study was taken from all patients.

Ultrasound examination was performed trans-abdominally on 'Philips HD 11 XE' machine with 2-5 MHz broadband convex probe. Positive cases were followed to know the ultimate outcome of pregnancy i.e. aborted/ terminated/ delivered. Postnatal confirmation of congenital abdominal anomalies were carried out by clinical examination/ imaging procedures / autopsy. Result were statistically analysed by SPSS software.

Results

Total 5761 fetuses with gestational age ≥10 weeks were screened by ultrasound. 32 cases of fetal congenital abdominal anomalies with or without other associated anomalies were diagnosed by ultrasound during the one year study period. Incidence of fetal congenital abdominal anomalies was 5.56 per 1000 births. Among the fetal congenital abdominal anomalies, fetal urinary tract anomalies were the most common with an incidence of 4.69 per 1000 births. Incidence of Omphalocele, Gastroschisis and Jejuno-ileal atresia was found to be 5.2, 1.7 and 1.7 per 10,000 births respectively. Among fetal congenital abdominal anomalies, fetal urinary tract anomalies (84.38%) were the most common followed by fetal anterior abdominal wall defects (12.50%) and fetal GI tract anomaly (3.13%).

Table-1: Distribution of different fetal congenital abdominal anomalies detected.

Fetal Congenital Abdominal Anomalies	Number of cases	Percentage
Fetal Urinary Tract Anomalies	**27**	**84.38%**
HDN	14	43.75%
MCDK	6	18.75%
Renal Agenesis	5	15.63%
Polycystic Kidney Disease	1	3.13%
BOO	1	3.13%
Fetal Anterior Abdominal Wall Defects	**4**	**12.50%**
Omphalocele	3	9.38%
Gastroschisis	1	3.13%
Fetal GI Tract Anomaly	**1**	**3.13%**
Jejuno-ileal atresia	1	3.13%
Total	**32**	**100.0%**

Among the fetal urinary tract anomalies, hydronephrosis (43.75%) was most common followed by multicystic

dysplastic kidney (18.75%) and renal agenesis (15.63%). Among fetal anterior abdominal wall defects, omphalocele (9.38%) was 3 times more common than gastroschisis (3.13%). A single case of jejuno-ileal atresia (3.13%) was diagnosed among the fetal GI tract anomalies [Table 1].

The mean POG (Period of Gestation) at the time of antenatal diagnosis of fetal congenital abdominal anomalies was 29.48±5.53 weeks. The maternal age ranged from 21 to 37 years, with mean age was 26.34±4.22 years. The majority of affected fetuses were male: 21 (65.62%). Females were affected in 11 (34.38%) cases. Majority of cases (65.63%) of fetal congenital abdominal anomalies were not associated with any recognizable maternal risk factor. Remaining (34.37%) cases were associated with known maternal risk factors. Maternal diabetes mellitus was the most common known risk factor associated in 9.38% cases.

Out of 27 fetuses diagnosed with urinary tract anomalies, 5 (18.52%) were associated with anomalies of other system**.** Out of 5, 2 were associated with single umbilical artery, 1 with cleft lip, 1 with VSD and 1 with polydactyly. [Table 2]

Table-2: Fetal urinary tract anomalies associated with anomalies of other system

Fetal Urinary Tract Anomalies	Associated anomalies of other system
MCDK	Cleft lip
HDN	Single Umbilical Artery
Renal Agenesis	Single Umbilical Artery
HDN	VSD (Figure 1)
HDN	Polydactyly

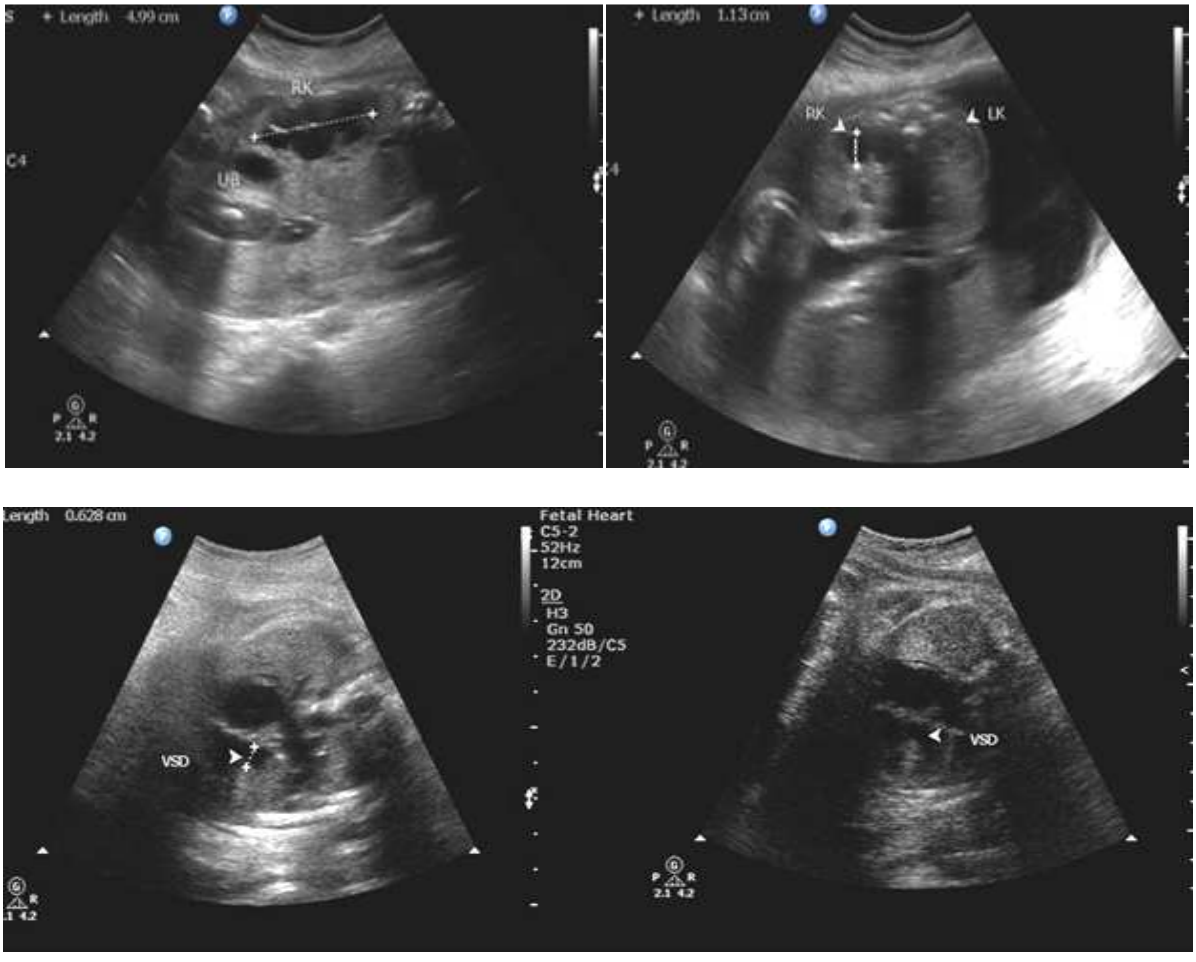

Figure-1A: Antenatal USG shows right kidney hydronephrosis and associated VSD

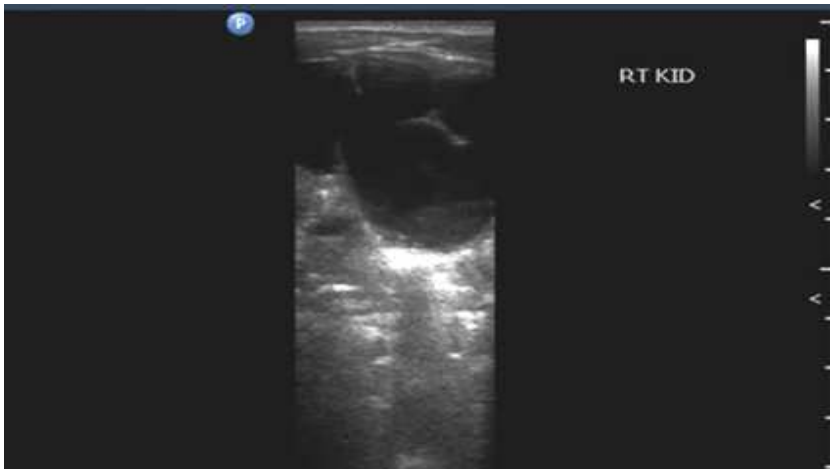

Figure-1B: Postnatal USG confirms right kidney hydronephrosis

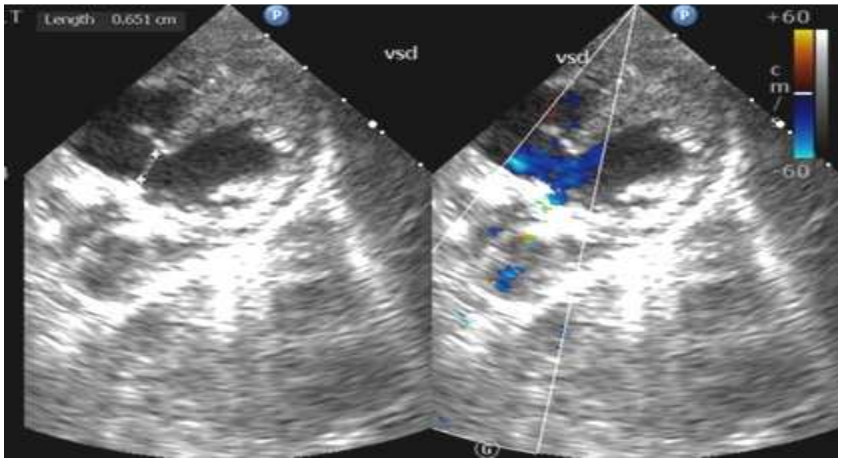

Figure-1C: Postnatal echo of fetus confirms vsd with color flow across defect

2 out of 3 cases of omphalocele were associated with anomalies of other system. One case of omphalocele was associated with hydrocephalus, spinal deformity, cleft lip, club foot and ectopia cordis (Figure-2) and other case with Myelomeningocoele.

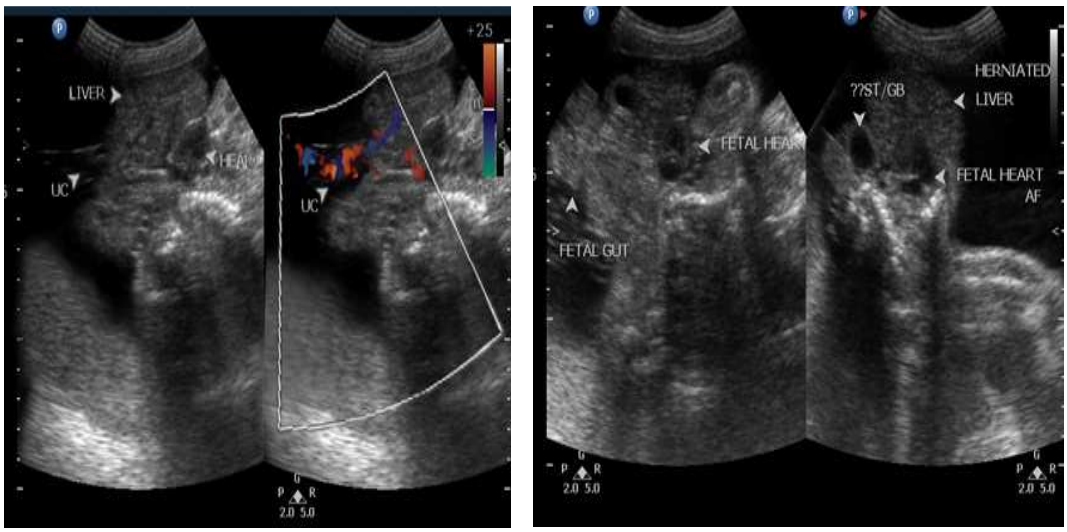

Figure-2A: Antenatal USG shows herniated liver and gut loops out of fetal abdomen at the base of umbilical cord insertion. Ectopia cordis also seen

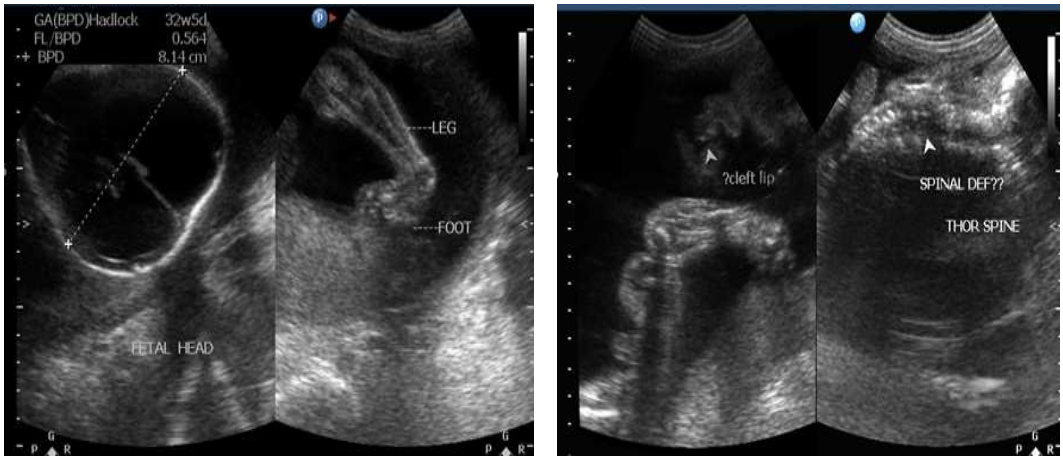

Figure-2B: Antenatal USG shows hydrocephalus, club foot Cleft lip and spinal deformity

Figure-2C: Abortus shows omphalocele with herniated liver and gut loops; club foot and cleft lip

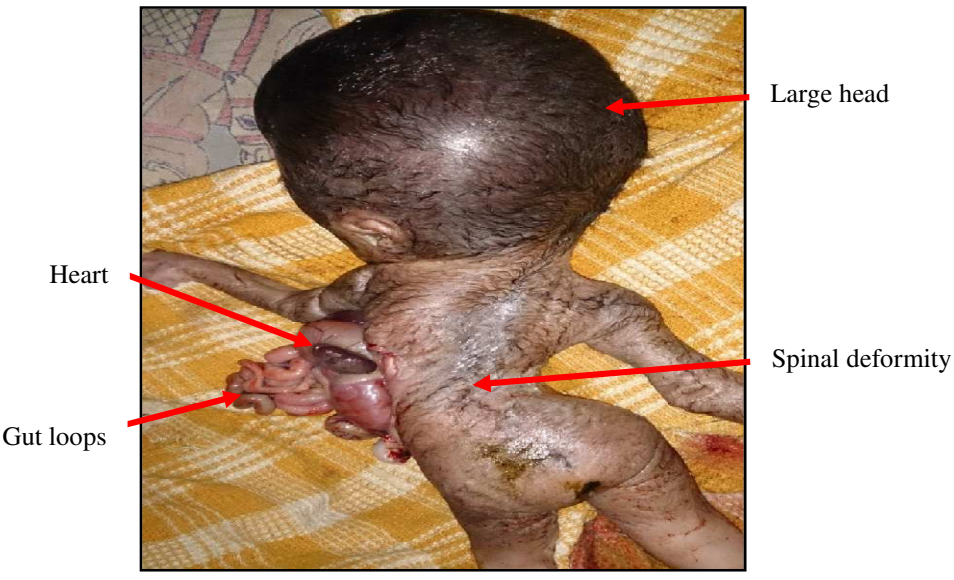

Figure- 2D: Abortus shows ectopia cordis, large head, herniated gut loops and spinal deformity

Oligohydramnions was seen in 8 (29.63%) out of 27 pregnancies with fetal urinary tract anomalies. Out of 27 cases of fetal urinary tract anomalies, 18 cases (66.67%) were delivered as term live baby and 5 cases (18.52%) as preterm live baby resulting in total 23 live born babies (85.19%). 2 cases (7.41%) underwent MTP and in 2 cases (7.41%) intrauterine death was observed. Intrauterine death occurred in fetuses diagnosed to have bilateral multicystic dysplastic kidneys and bladder outlet obstruction respectively. Termination of pregnancies were done in two cases both diagnosed to have bilateral renal agenesis.

Out of 4 cases of fetal anterior abdominal wall defect, intrauterine death was observed in 2 cases, 1 case underwent MTP and 1 case delivered as preterm live baby. A single case of fetal GI tract anomaly (jejuno-ileal atresia) was delivered as term live baby.

Postnatal confirmation of antenatally diagnosed fetal congenital abdominal anomalies was done by clinical examination/ imaging procedures / autopsy.

Agreement between antenatal and postnatal diagnosis was seen in 29 out of 32 cases resulting in concordance found to be 90.63%. Agreement between antenatal and postnatal diagnosis was seen in 24 cases of urinary tract anomalies, 4 cases of anterior abdominal wall defects and in 1 case of GI tract anomaly. In the remaining 3 cases of urinary tract anomalies variations between antenatal and postnatal diagnosis were found.

Normal renal scans were seen postnatally in one infant diagnosed to have right hydronephrotic kidney and in one infant diagnosed to have left renal agenesis antenatally. One infant found to have left hydronephrotic kidney on antenatal scans showed multicystic dysplastic left kidney on postnatal scans.

Discussion

With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal and neonatal mortality in developing countries like India [4]

Presently Ultrasound is the best screening method for prenatal detection of fetal congenital anomalies because of its safety, availability, accuracy, and cost-effectiveness [5]. Early diagnosis of congenital anomalies provides information to parents so they can make decisions about whether to continue or terminate the pregnancy and what their options are for fetal or postnatal repair.

In the present study, 32 cases of fetal congenital abdominal anomalies were diagnosed out of 5761 cases scanned, resulting an incidence of 5.56 per 1000 births. Among the fetal congenital abdominal anomalies, fetal urinary tract anomalies (27 cases) were the most common with an incidence of 4.69 per 1000 births which is comparable to the study done by Saha et al [8], reported the incidence of fetal urinary tract anomalies 4 per 1000 births. In our study, only 4 cases of fetal anterior abdominal wall defects and one case of jejuno-ileal atresia were diagnosed.

Among the fetal urinary tract anomalies, hydronephrosis (43.75%) was the most common followed by multicystic dysplastic kidney (18.75%) and renal agenesis (15.63%). This result of our study is consistent with those of previous studies done by, Kim and Song et al [9], Vega et al [10], Bondagji et al [11] and Kumar et al [12].

Table-3: Distribution of fetal urinary tract anomalies

Author and year of study	Hydronephrosis	MCDK	Renal agenesis
Kim and Song [9]	77%	10.4%	4.2%
De La Vega et al [10]	54.7%	17.9%	12.8%
Bondagji [11]	51.1%	12.8%	8.5%
Kumar et al [12]	44.9%	26.4%	8%
Present Study	43.75%	18.75%	15.63%

Early detection of congenital anomalies provides information regarding the important decision during pregnancy whether to terminate or continue pregnancy, any fetal intervention if available and proper postnatal management of infant. In the present study average gestational age for ultrasound diagnosis of congenital abdominal anomalies was 29.48±5.53 weeks. Although ultrasound imaging is highly sensitive for prenatal diagnosis of congenital abdominal anomalies, but diagnosis was not possible before 20 weeks in 90.6% cases. Major reasons for late diagnosis were either USG performed too early in pregnancy or late referral of cases to the hospital.

Agreement between antenatal and postnatal diagnosis was seen in 29 out of 32 cases resulting in concordance found to be 90.63%. Agreement between antenatal and postnatal diagnosis was seen in 24 cases of urinary tract anomalies, 4 cases of anterior abdominal wall defects and in one case of GI tract anomaly. The concordance between antenatal and postnatal diagnosis of our study is almost similar to study by Bondagji [11] and is comparable to other studies by Sanghvi et al [13], Brunisholz et al [14] and Policiano et al [15]. In contrast, the concordance of our study is much higher than studies by Helin and Persson [16] and Barakat et al [17] [Table 4].

Table-4: Concordance between antenatal and postnatal diagnosis.

Author and year of study	Percentage
Helin and Persson [16]	65%
Barakat et al [17]	68%
Sanghviet al [13]	81.5%
Brunisholzet al [14]	86%
Policiano et al [15]	88.8%
Bondagji [11]	90.1%
Present Study	90.63%

With time frame, ultrasound is becoming more accurate because of increasing experience of radiologists day by day. It is evident in literature also as shown in table above.

Limitation of Study-In our study, only 4 cases of fetal anterior abdominal wall defects and 1 case of fetal GI tract anomaly were diagnosed because of limited time duration of one year. So, it is not possible to discuss the parameters of these anomalies in detail because literatures about these anomalies are of longer duration with good number of cases available

Parameters eg sensitivity, specificity, positive predictive value, negative predictive value couldn't be assessed as only positive cases were followed postnatally. Single centre study, although highest burden of region, still can't be reflect whole region.

Conclusion

Incidence of antenatally diagnosed congenital anomalies is increasing as compared to past because of advanced diagnostic facilities and better-trained radiologists. Fetal congenital anomalies are a major cause of perinatal and infant mortality, so routine antenatal ultrasound examination should be performed for all pregnant women to diagnose these anomalies.

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Permission of IRB: Not required

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