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Original Research Article

Sequential first and second trimester ultrasound screening for early detection of fetal anomalies: a prospective observational study

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ABSTRACT

Background: First-trimester ultrasonography at 11–13^{+6} weeks plays a vital role in early detection of chromosomal abnormalities and structural fetal anomalies. When combined with a second-trimester anomaly scan at 18–24 weeks, it enhances diagnostic accuracy and improves pregnancy management.
Methods: This prospective observational study was conducted at a tertiary care centre from January 2023 to January 2025 and included. The expected prevalence of fetal anomalies (3–5%) and, this sample size 220 randomly selected singleton antenatal women between 11–13^{+6} weeks of gestation was considered adequate to assess the detection rate with reasonable accuracy. First-trimester evaluation included nuchal translucency (NT), nasal bone, ductus venosus Doppler, tricuspid regurgitation, and early fetal anatomical survey. All participants subsequently underwent a detailed anomaly scan at 18–24 weeks.
Results: This prospective observational study included 220 singleton antenatal women who underwent sequential ultrasound screening at 11–13^{+6} weeks and 18–24 weeks of gestation. A total of 11 foetuses (5%) were diagnosed with anomalies, of which 10 cases (4.55%) were detected during the first-trimester scan and 1 case (0.45%) during the second-trimester scan. Detected anomalies included chromosomal abnormalities (Trisomy 21,18, and 13), increased nuchal translucency, and structural defects involving the central nervous system, cardiac system, abdominal wall, and genitourinary system. Increased NT (>95th percentile) was observed in 6.4% cases. Anomalies were slightly more common in multigravida women and referred cases.
Conclusions: In the present study, sequential first and second trimester ultrasound screening proved effective in early detection of fetal anomalies, enabling timely counselling and appropriate pregnancy management.

Keywords: Prenatal screening, Fetal abnormalities, First trimester ultrasonography, Second trimester ultrasound, Congenital anomalies

INTRODUCTION

Congenital anomalies are an important cause of perinatal morbidity and mortality worldwide and contribute significantly to neonatal deaths and long-term childhood disability. The global prevalence of congenital anomalies is estimated to affect approximately 3–5% of all pregnancies.[1] Early detection of fetal anomalies during pregnancy is essential because it allows timely counselling, appropriate prenatal management, and informed decision-making for parents and clinicians. Prenatal screening strategies have therefore become an

integral component of modern obstetric care.[2] Ultrasonography remains the primary non-invasive modality for antenatal assessment of fetal development and structural abnormalities. Advances in ultrasound technology have substantially improved the ability to detect fetal anomalies during different stages of gestation.[3]

First-trimester ultrasound screening performed between 11 and 13^{+6} weeks of gestation has gained increasing importance for early identification of chromosomal abnormalities and major structural anomalies. This examination includes assessment of nuchal translucency thickness and early evaluation of fetal anatomy.[4] Increased nuchal translucency (NT) is one of the most established sonographic markers for chromosomal abnormalities, particularly trisomy 21, trisomy 18, and trisomy 13. It may also be associated with major structural anomalies such as congenital heart defects and genetic syndromes.[5] Additional first-trimester markers including the nasal bone, ductus venosus Doppler waveform, and tricuspid valve regurgitation further improve the detection rate of chromosomal abnormalities when incorporated into screening protocols.[6]

Early anatomical assessment during the first trimester can identify several major fetal anomalies involving the central nervous system, abdominal wall, skeletal system, and urinary tract. However, certain anomalies may not be apparent during early gestation due to ongoing fetal development.[7] Therefore, a detailed second-trimester anomaly scan performed between 18 and 24 weeks of gestation is recommended for comprehensive evaluation of fetal anatomy. This scan allows systematic assessment of the fetal brain, spine, heart, abdomen, limbs, and genitourinary structures.[8] Second-trimester ultrasonography remains the standard screening method for detecting many structural anomalies that may evolve or become more apparent later in pregnancy.[9]

Sequential ultrasound screening enables earlier identification of severe anomalies, allowing timely referral for further diagnostic tests such as fetal echocardiography, genetic testing, or invasive diagnostic procedures when required.[10] Early diagnosis of fetal anomalies also provides parents with more time for counselling and decision-making regarding pregnancy continuation and perinatal management.[11]

Furthermore, early detection allows planning of delivery in tertiary care centres where appropriate neonatal care and surgical facilities are available.[12] Despite the proven benefits of prenatal ultrasound screening, the detection rates of fetal anomalies may vary depending on factors such as gestational age, operator expertise, and availability of advanced imaging facilities.[13] Evaluating the effectiveness of sequential ultrasound screening in routine clinical practice is therefore important to optimize antenatal care strategies and improve fetal outcomes.[14] Hence, the present study was conducted to assess the role of sequential first and second trimester ultrasound screening in early detection of fetal anomalies among antenatal women attending a tertiary care centre.

METHODS

This prospective observational study was conducted in the Department of Obstetrics and Gynaecology at Shrimati shardaben general hospital a tertiary care centre between January 2023 and January 2025. The sample size was calculated using the formula for estimation of proportion ($n=Z^2pq/d^2$). Assuming a prevalence of congenital fetal anomalies of 3%, with a 95% confidence level and 2% allowable error, the minimum sample size was estimated to be about 200. To account for possible loss to follow-up and incomplete data, the final sample size was increased to 220 antenatal women.

Pregnant women attending the antenatal outpatient department were randomly selected. Women with singleton pregnancies between 11 and 13+6 weeks of gestation and crown–rump length (CRL) of 45–84 mm were included. In cases of uncertain last menstrual period or irregular cycles, gestational age was determined using CRL. Women with multifetal pregnancies, gestational age <11 weeks or ≥14 weeks, CRL outside the specified range, or those unwilling to participate were excluded. After obtaining informed consent, obstetric history and clinical examination were recorded. Ultrasound evaluation was performed primarily by transabdominal scan and transvaginal scan when required, following the 2023 ISUOG practice guidelines. The first-trimester scan included assessment of fetal anatomy, nuchal translucency, nasal bone, and uterine artery Doppler. Risk for chromosomal aneuploidies (Trisomy 21,18, and 13) was calculated using maternal age and ultrasound markers through Fetal Medicine Foundation software. High-risk cases underwent further screening with NIPT, Double Marker, or Quadruple Marker tests, with confirmation by chorionic villus sampling or amniocentesis when indicated. All participants underwent a follow-up anomaly scan between 18–24 weeks. Maternal details, ultrasound findings, screening results, and pregnancy outcomes were recorded and analysed in Microsoft Excel and jamovi software.

RESULTS

A total of 220 pregnant women were included in the study. The majority of participants belonged to the 21–30 years age group, accounting for 162 cases (73.6%). Fetal anomalies were detected in 11 cases overall. Among these, the highest number of anomalies was observed in the 21–30 years age group with 8 cases (3.63%), followed by 2 cases (0.91%) in the 31–40 years age group and 1 case (0.45%) in women aged ≤20 years. With respect to parity, the majority of the study population were multigravida women, accounting for 160 cases (72.7%), while primigravida constituted 60 cases (27.3%). Fetal anomalies were detected in 7 multigravida women (3.18%) and 4 primigravida women (1.81%). (Table 1). A total of

220 pregnant women underwent ultrasound examination during both the first trimester (11–13^{+6} weeks) and the second trimester (18–24 weeks). Transabdominal sonography (TAS) was performed in all 220 cases during both scans. Transvaginal sonography (TVS) was additionally utilized in 70 cases during the first-trimester scan and in 22 cases during the second-trimester scan to obtain better visualization of fetal structures when required (Table 2).

Table 1: Distribution of age and parity among the study population with detection of fetal anomalies on ultrasound (n=220).

Variable	Category	Total cases N (%)	Cases with fetal anomaly N (%)
Age group (years)	≤20	2 (0.9)	1 (0.45)
	21–30	162 (73.6)	8 (3.63)
	31–40	56 (25.5)	2 (0.91)
Parity	Primigravida	60 (27.3)	4 (1.81)
	Multigravida	160 (72.7)	7 (3.18)

Table 2: Mode of ultrasound examination used during first and second trimester scans (n=220).

Mode of scan	11–13^{+6} weeks scan (N)	18–24 weeks scan (N)
Transabdominal sonography (TAS)	220	220
Transvaginal sonography (TVS)	70	22

A variety of structural anomalies were identified during ultrasound examination. Among these, absent nasal bone was the most frequently observed finding, noted in 3 cases (1.4%). Certain anomalies such as diffuse skin/scalp oedema, abnormal four-chamber view of the heart, single umbilical artery, and renal anomalies were detected in 2 cases each (0.9%). Other anomalies including amniotic band, cystic hygroma, non-immune hydrops, cleft lip/cleft palate, micrognathia, left hypoplastic heart syndrome, omphalocele, ventricular septal defect, and ventriculomegaly were each observed in 1 case (0.45%). These findings demonstrate the range of fetal structural anomalies that can be detected through early ultrasound screening during pregnancy (Table 3).

A total of 215 pregnant women underwent second-trimester ultrasound examination between 18 and 24 weeks of gestation. Five cases (2.3%) underwent medical termination of pregnancy (MTP) prior to the scheduled second-trimester anomaly scan due to abnormalities detected earlier, while one additional case underwent MTP after the 18–24 weeks scan. Only one new anomaly was identified during the second-trimester scan. Among the four cases with structural anomalies noted at the 18–24 weeks scan, three had already been diagnosed during the first-trimester scan, whereas renal pyelectasis was newly identified in one case during the second-trimester examination. Renal pyelectasis does not have significant diagnostic relevance during the 11–13^{+6} weeks scan; therefore, in the present study, the majority of major fetal anomalies were detected during the first-trimester ultrasound screening (Table 4).

Table 3: Types of fetal structural anomalies identified at first-trimester ultrasound (11–13^{+6} weeks) (n=220).

Type of anomalies	Number (N)	Percentage (%)
Total anomalies detected	11	5.0
Amniotic band	1	0.45
Absent nasal bone	3	1.4
Cystic hygroma	1	0.45
Diffuse skin/scalp oedema	2	0.9
Non-immune hydrops	1	0.45
Cleft lip/cleft palate	1	0.45
Micrognathia	1	0.45
Left hypoplastic heart syndrome	1	0.45
Abnormal 4-chamber heart	2	0.9
Omphalocele	1	0.45
Single umbilical artery	2	0.9
Ventricular septal defect	1	0.45
Renal anomaly	2	0.9
Ventriculomegaly	1	0.45

(*more than 1 anomaly in fetus included).

Table 4: Types of fetal structural anomalies identified at second-trimester ultrasound (18-24 weeks) (n=215).

Type of anomalies	Number (N)	Percentage (%)
Choroid plexus cyst	1	0.45
Skeletal abnormalities	0	0
Renal pyelectasis	1	0.45
Single umbilical artery	2	0.9
MTP before follow-up 18–22 weeks scan	5	2.3
Normal scan	211	98.1

DISCUSSION

The present prospective observational study evaluated the effectiveness of sequential first- and second-trimester ultrasound screening for early detection of fetal anomalies among 220 antenatal women. A total of 11 fetal anomalies (5%) were detected in the study population, with the

majority identified during the first-trimester scan (4.55%), while only one additional anomaly (0.45%) was detected during the second-trimester scan. The overall prevalence of fetal anomalies in the present study (5%) is comparable to the findings reported in previous studies. Souka et al reported that approximately 4–5% of foetuses demonstrate structural or chromosomal abnormalities detectable during early pregnancy screening.[15] Similarly, Syngelaki et al observed that first-trimester ultrasound examination can detect a significant proportion of major fetal anomalies, with detection rates ranging between 40–60% depending on the type of anomaly.[16]

In the present study, most fetal anomalies were detected during the first-trimester scan (11–13^{+6} weeks). Becker and Wegner reported that detailed first-trimester ultrasound examination can identify many major structural anomalies.[17] Similarly, Grande et al demonstrated that systematic first-trimester anatomical assessment allows early detection of several structural abnormalities.[18] The distribution of maternal age in the present study showed that the majority of participants were in the 21–30 years age group, which is the most common reproductive age group. Similar findings were reported by Viora et al, who noted that congenital anomalies are often detected in the most common reproductive age group due to the larger number of pregnancies occurring in that population rather than increased biological risk.[19] With respect to parity, the present study observed slightly higher anomaly detection among multigravida women compared to primigravida women. However, previous studies such as those conducted by Boyd et al have reported that slightly higher frequency observed among multigravida women in this study.[20]

The types of fetal anomalies detected in the present study included chromosomal markers such as absent nasal bone, cystic hygroma, and increased nuchal translucency–related findings, as well as structural anomalies involving the cardiac system, craniofacial structures, abdominal wall, and renal system. Similar findings were reported by Borrell et al, who demonstrated that first-trimester ultrasound markers such as increased nuchal translucency and absent nasal bone are strongly associated with chromosomal abnormalities.[21] Cardiac abnormalities such as abnormal four-chamber view and ventricular septal defect were also identified in the present study. Carvalho et al demonstrated that fetal echocardiography performed in early pregnancy can significantly improve early diagnosis of congenital heart disease, allowing timely referral for specialized evaluation.[22]

In the present study, transvaginal sonography was used in a number of cases to improve visualization of fetal structures during early gestation. Timor-Tritsch et al reported that transvaginal ultrasound plays an important role in early fetal anatomical assessment, particularly during the first trimester when transabdominal imaging may be limited by fetal size or maternal factors.[23] The second-trimester anomaly scan remains an important component of routine prenatal care because some anomalies become more apparent as fetal development progresses. In the present study, only one additional anomaly was detected during the second-trimester scan. Similar observations were reported by Rossi and Prefumo, who concluded that many major anomalies can be detected during early pregnancy, while the mid-trimester scan serves as confirmation and allows detection of anomalies that develop later.[24] Another important finding in the present study was that several pregnancies with severe anomalies underwent medical termination before the scheduled second-trimester scan. Boyd et al also emphasized that early prenatal diagnosis improves counselling and allows better planning of pregnancy management and delivery.[20] The results of the present study should be considered with certain limitations, including its single-centre design and relatively small sample size. Inclusion of only antenatal attendees may introduce selection bias. In addition, early pregnancy terminations and the operator-dependent nature of ultrasound could have influenced the detection of fetal anomalies.

CONCLUSION

Sequential first- and second-trimester ultrasound screening is effective for the early detection of fetal anomalies. In the present study, the majority of anomalies were identified during the first-trimester scan, highlighting the importance of early antenatal ultrasound evaluation. The second-trimester anomaly scan remains valuable for confirming earlier findings and identifying anomalies that may become evident later in pregnancy. These findings emphasize the complementary role of both scans in comprehensive prenatal assessment. Incorporating sequential ultrasound screening can improve the overall detection of fetal anomalies during pregnancy.

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REFERENCES

1. WHO. Birth defects (congenital disorders). Geneva: World Health Organization. 2023. Available at: https://www.who.int/news-room/fact-sheets/detail/birth-defects/. Accessed on 18 February 2026.
2. Cunningham FG, Leveno KJ, Dashe JS, Hoffman BL, Spong CY, Casey BM. Obstetrics. 26th ed. New York: McGraw-Hill. 2022;749-54.

3. Rumack CM, Levine D. Diagnostic ultrasound. 5th ed. Philadelphia (PA): Elsevier. 2018.
4. Nicolaides KH. Screening for fetal aneuploidies at 11 to 13 weeks. Prenat Diagn. 2011;31:7-15.
5. Souka AP, Snijders RJM, Novakov A, Soares W, Nicolaides KH. Defects and syndromes in chromosomally normal fetuses with increased nuchal translucency thickness at 10–14 weeks of gestation. Ultrasound Obstet Gynecol. 1998;11:391-400.
6. Maiz N, Valencia C, Kagan KO, Wright D, Nicolaides KH. Ductus venosus Doppler in screening for trisomies 21, 18 and 13 and Turner syndrome at 11–13 weeks of gestation. Ultrasound Obstet Gynecol. 2009;33:512-7.
7. Syngelaki A, Hammami A, Bower S, Zidere V, Akolekar R, Nicolaides KH. Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation. Ultrasound Obstet Gynecol. 2019;54:468-76.
8. Salomon LJ, Alfirevic Z, Berghella V, Bilardo CM, Hernandez-Andrade E, Johnsen SL, et al. Practice guidelines for performance of the routine mid-trimester fetal ultrasound scan. Ultrasound Obstet Gynecol. 2011;37:116-26.
9. International Society of Ultrasound in Obstetrics and Gynecology (ISUOG). Performance of the routine mid-trimester fetal ultrasound scan. ISUOG; 2022. Available at: https://www.isuog.org/resource/isuog-practice-guidelines-updated-performance-of-the-routine-mid-trimester-fetal-ultrasound-scan.html. Accessed on 18 February 2026.
10. Liao YM, Li SL, Luo GY, Wen HX, Ouyang SY, Chen CY, et al. Routine screening for fetal limb abnormalities in the first trimester. Prenat Diagn. 2016;36(2):117-26.
11. Malone FD, Canick JA, Ball RH, Nyberg DA, Comstock CH, Bukowski R, et al. First-trimester or second-trimester screening, or both, for Down's syndrome. N Engl J Med. 2005;353:2001-11.
12. Chervenak FA, McCullough LB, Levene MI. An ethically justified, clinically comprehensive approach to peri-viability: gynaecological, obstetric, perinatal and neonatal dimensions. J Obstet Gynaecol. 2007;27:3-7.
13. Grandjean H, Larroque D, Levi S. Sensitivity of routine ultrasound screening of pregnancies in the Eurofetus database. Ann N Y Acad Sci. 1998;847:118-24.
14. Whitworth M, Bricker L, Mullan C. Ultrasound for fetal assessment in early pregnancy. Cochrane Database Syst Rev. 2015;(7):CD007058.
15. Souka AP, Pilalis A, Kavalakis I, Papantoniou N, Mesogitis S, Dalkalitsis N. Screening for major structural abnormalities at the 11- to 14-week ultrasound scan. Am J Obstet Gynecol. 2006;194:393-6.
16. Syngelaki A, Hammami A, Bower S, Zidere V, Akolekar R, Nicolaides KH. Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation. Ultrasound Obstet Gynecol. 2019;54:468-76.
17. Becker R, Wegner RD. Detailed screening for fetal anomalies and cardiac defects at the 11–13-week scan. Ultrasound Obstet Gynecol. 2006;27:613-8.
18. Grande M, Arigita M, Borobio V, Jiménez JM, Fernández S, Borrell A. First-trimester detection of structural abnormalities and the role of aneuploidy markers. Ultrasound Obstet Gynecol. 2012;39:157-63.
19. Viora E, Zamboni C, Mortara G, Stillavato S, Bastonero S, Errante G, et al. Trisomy 18: fetal ultrasound findings at different gestational ages. Am J Med Genet A. 2007;143A:553-7.
20. Boyd PA, Devigan C, Khoshnood B, Loane M, Garne E, Dolk H, et al. Survey of prenatal screening policies in Europe for structural malformations and chromosome anomalies, and their impact on detection and termination rates for neural tube defects and Down's syndrome. BJOG. 2008;115:689-96.
21. Borrell A, Grande M, Pauta M, Rodríguez-Revenga L, Figueras F. Chromosomal microarray analysis in fetuses with growth restriction and normal karyotype: a systematic review and meta-analysis. Fetal Diagn Ther. 2018;44:1-9.
22. Carvalho JS, Moscoso G, Tekay A, Campbell S, Thilaganathan B, Shinebourne EA. Clinical impact of first and early second trimester fetal echocardiography on high-risk pregnancies. Heart. 2004;90:921-6.
23. Parisi N, Rizzo G, Cecchini F, Schiattarella A, Khalil A, Mappa I, et al. Outcome of apparently isolated fetal posterior fossa anomalies: systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2025;66:571-81.
24. Rossi AC, Prefumo F. Accuracy of ultrasonography at 11–14 weeks of gestation for detection of fetal structural anomalies: a systematic review. Obstet Gynecol. 2013;122:1160-7.

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