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Importance of Early Imaging in Detecting Fetal Structures: A Case Series

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Abstract: ***Objective: To highlight the vital role of early ultrasound imaging in the first and early second trimesters for detecting fetal structural anomalies and placental abnormalities that significantly impact prenatal counseling and clinical management. Methods: This was a case series of pregnancies evaluated at an academic medical centre. Cases were identified by querying the obstetric ultrasound database for fetuses with structural abnormalities or placental findings detected between 11 to14weeks' gestation. Ultrasound reports, images, medical records, and outcomes were reviewed to compile relevant examples illustrating the range of conditions diagnosed by early imaging. Nuchal translucency (NT) thickness, presence of double aortic arch, and cystic hygroma were evaluated as markers for potential fetal anomalies. Results: In three cases (Cases 1, 2, and 3), the ultrasound findings were normal, with no increased NT thickness, double aortic arch, or cystic hygroma detected. These cases were managed with routine prenatal care. However, in two cases (Cases 4 and 5), increased NT thickness was observed, along with the presence of double aortic arch and cystic hygroma, raising concerns for potential chromosomal or structural abnormalities. These findings prompted further diagnostic investigations and appropriate interventions. Conclusion: Early prenatal imaging plays a crucial role in detecting fetal structural anomalies, such as increased NT thickness, double aortic arch, and cystic hygroma. Timely identification of these markers allows for prompt follow-up investigations, targeted interventions, and comprehensive counseling, ultimately improving fetal and maternal outcomes. This case series underscores the importance of routine prenatal imaging and vigilant monitoring for optimal management of pregnancies with potential fetal anomalies.***

Keywords: Pregnancy Prenatal Diagnosis, Ultrasonography, Prenatal Congenital Abnormalities, Fetal Diseases

1. Introduction

Prenatal ultrasound has become an integral part of modern obstetric care, providing invaluable information about fetal anatomy, growth, and well-being throughout pregnancy. While the 20-week anatomical survey remains the primary screening tool for structural abnormalities, there is increasing evidence that earlier imaging in the late first trimester and early second trimester can detect many fetal anomalies and placental abnormalities at an even more premature stage.[1,2]

Early detection of fetal structural defects and placental issues is crucial for several reasons. Firstly, it allows for timely counseling of parents regarding the nature and prognosis of the condition, which can guide difficult but deeply personal decisions about continuing or terminating the pregnancy. Secondly, it provides a wider window for further diagnostic testing and evaluations, such as amniocentesis, fetal echocardiography, or fetal MRI. Thirdly, certain congenital anomalies may benefit from fetal interventions or specialized delivery planning when diagnosed prenatally.[3,4] Finally, early imaging can also identify factors that increase the risk of adverse pregnancy outcomes like preterm birth or intrauterine growth restriction, allowing appropriate monitoring and management.[5]

The comprehensive ultrasound of the first trimester is carried out between weeks 11 and 13+6 of pregnancy. A thorough assessment of foetal anatomy, an appraisal of the uterine and adnexal regions, and a general overview and foetal biometry are all included in the full first trimester ultrasound examination. The purpose of this ultrasound scan is to supplement the ultrasound performed in the second trimester and, in most cases, to offer early confirmation of normalcy.[6]

Foetal sonographic features known as antenatal soft ultrasonography markers are usually not abnormalities per se, but rather suggest a higher age-adjusted risk of an underlying foetal aneuploidy or certain non-chromosomal abnormalities. Depending on the results of a biochemical test or the mother's age, the presence or lack of these minor indicators can be utilised to modify a patient's preexisting risk for aneuploidy.[7]

This becomes particularly important in screening for trisomy 21, as approximately 75% of fetuses affected by trisomy 21 will not have ultrasound-detectable major congenital anomalies at the time of the second trimester anatomic survey.

While the 18-22 week scan remains the primary screening tool, studies have demonstrated that up to 54% of fetuses with structural abnormalities can be detected before 14 weeks, and over 82% before 18 weeks.[8,9] This underscores the importance of high-quality first and early second trimester imaging by skilled sonographers to maximize the detection of fetal anomalies at the earliest possible gestation. We present a series of cases that highlight the vital role of early imaging in the prenatal detection of structural defects, placental abnormalities, and other conditions, and discuss the implications for clinical practice.

2. Methodology

This was a case series of pregnancies evaluated at Radiology department referred from the OBGY department of D.Y Patil Medical College, between 11 to 14 weeks of gestation.

At our institution, all pregnant women are offered routine ultrasound examinations in the first and second trimesters based on national guidelines. A dating scan is performed between 8-13 weeks to confirm viability, accurately date the pregnancy, and provide a baseline nuchal translucency measurement. An anatomical survey is then performed at 18-22 weeks to thoroughly evaluate fetal anatomy.

However, additional ultrasounds may be obtained earlier for specific indications such as vaginal bleeding, elevated aneuploidy risk, or follow up of suspected findings. Ultrasounds before 14 weeks are performed trans-vaginally to optimize resolution, with transabdominal scanning after 14 weeks gestation.

The patients were screened on GE Logique P9 R3 machine. A curvilinear 1-5 Hz probe was used and reviewed by radiologists. Fetal anatomic structures are systematically assessed in line with national guidelines.

For each selected case, detailed information was collected from the medical records on maternal demographics, gestational age at ultrasound detection, ultrasound findings, results of any genetic testing or additional imaging, ultimate diagnosis, pregnancy outcome, and clinical management. Relevant ultrasound images were also captured to illustrate key findings.

The institutional review board approved this study and informed consent was taken from all study subjects.

Descriptive statistics were used to summarize maternal characteristics. Each case is presented to highlight the role of early imaging in antenatal diagnosis, with a focus on implications for parental counseling, further testing, clinical decision-making, and prenatal management. Ultrasound images are provided to correlate with described findings.

3. Cases

Case 1: A 34-year-old G2P1L1**, whose first preganancy was normal and in second pregnancy** underwent routine prenatal ultrasound examination, which revealed normal nuchal translucency (NT) thickness and absence of double aortic arch or cystic hygroma, indicating a low risk for chromosomal abnormalities and structural defects. On NT scan the CRL was noted to be 56.5 mm which corresponded to 12 weeks 2 days. Subcutaneous edema was noted at head, neck and torso region.

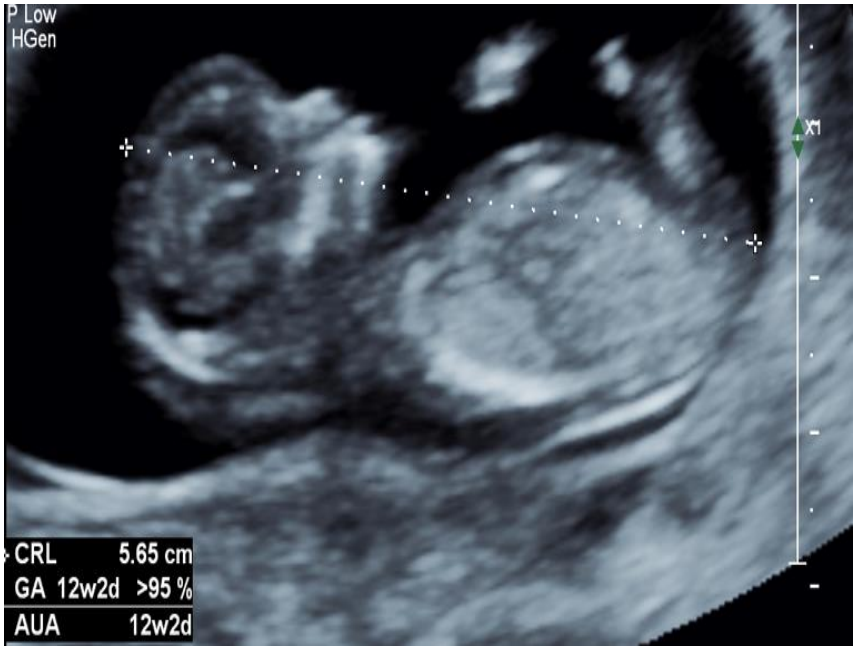

Image 1: Case 1 showing NT scan with CRL:56.5mm

Case 2: During an 8-week dating ultrasound for a 32-year-old woman, a prenatal ultrasound scan showed normal NT thickness, and no evidence of double aortic arch or cystic hygroma, suggesting a reassuring fetal anatomy at the time of examination.

Case 3: A 20-week anatomy scan revealed normal NT thickness, with no detectable double aortic arch or cystic hygroma, indicating a low risk for certain fetal anomalies.

Case 4: During a routine 13-week NT scan, demonstrated increased NT thickness, along with the presence of a double aortic arch and cystic hygroma. These findings raised suspicion for potential chromosomal or structural abnormalities, prompting further diagnostic investigations.

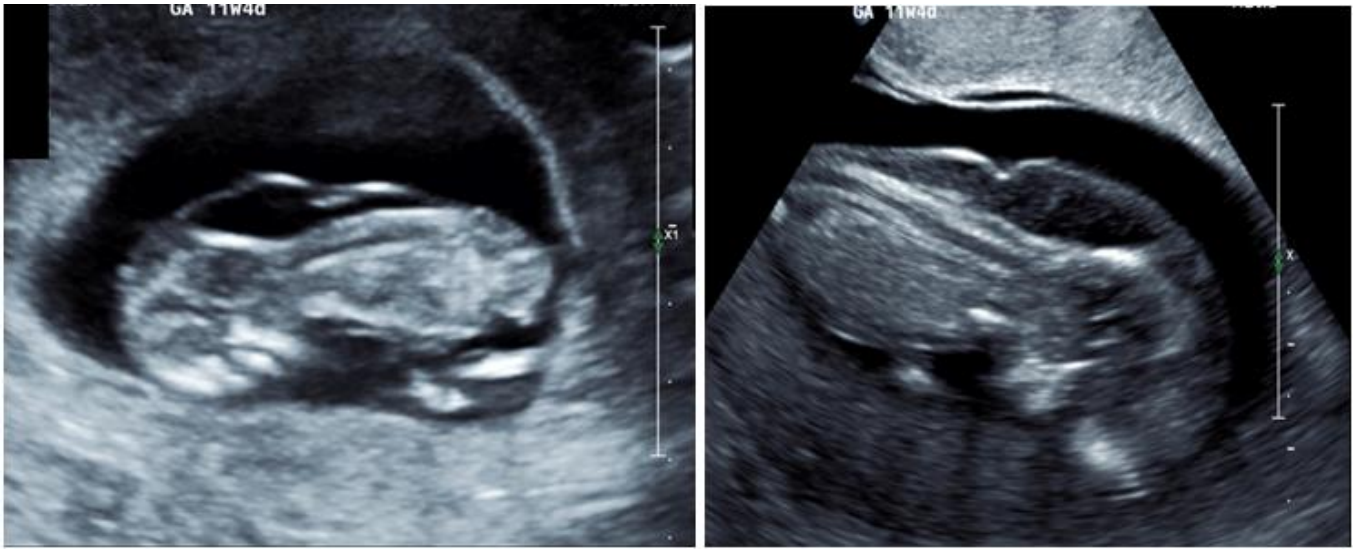

Image 2: Case 4 with cytic hygroma

Case 5: A 35-year-old woman with a poor obstetric history received an 11-week ultrasound that demonstrated increased NT thickness, accompanied by the presence of a double aortic arch and cystic hygroma, similar to Case 4. These findings necessitated additional follow-up and potential interventions. Considering the presence of cystic hygroma and double aortic arch, possibility of chromosomal abnormality was considered and CVS was performed for FISH and microarray. The reports of CVS came as FISH Trisomy 21

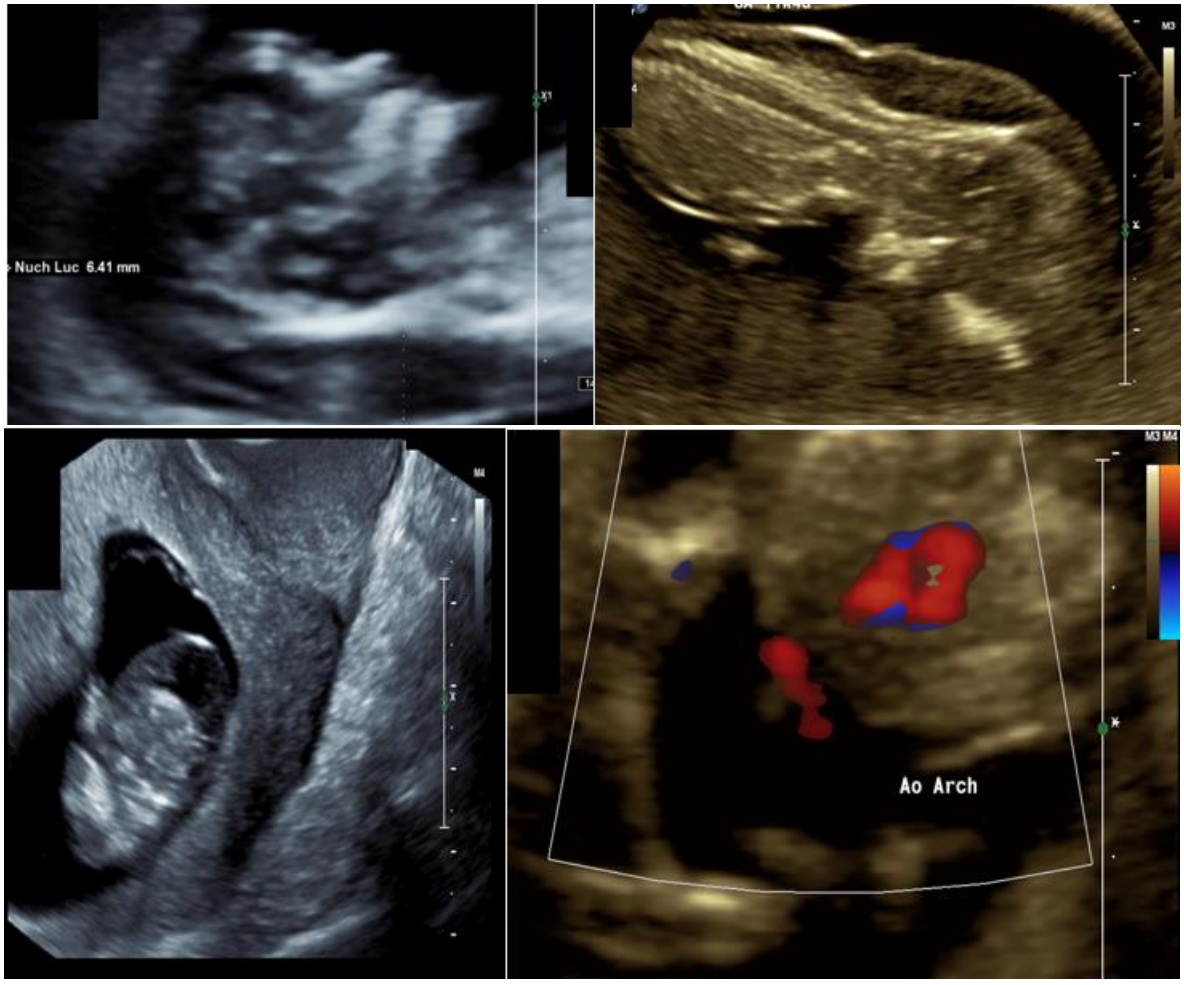

Image 3: Case 5 with increased nuchal thickness, cystic hygroma and double aortic arch

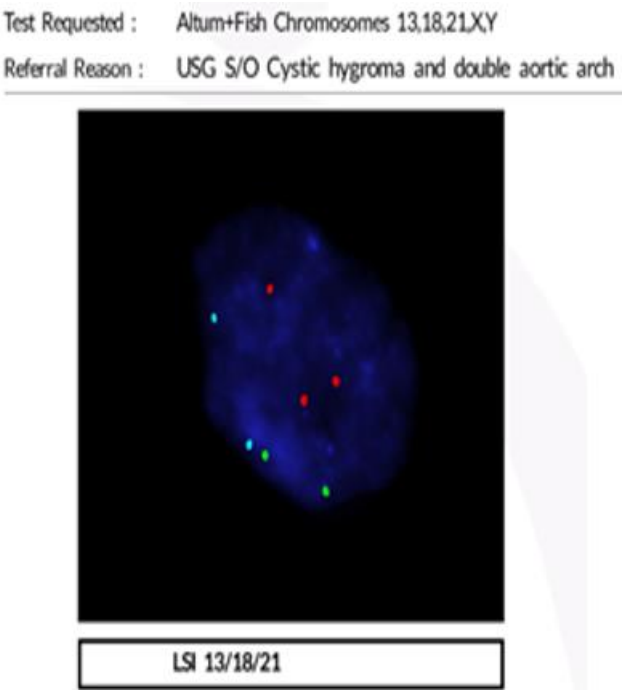

Image 4: Chromsomal test

Table 1: Study cases

	NT Thickness	Double Aortic Arch	Cystic Hygroma
Case 1	Normal	Absent	Absent
Case 2	Normal	Absent	Absent
Case 3	Normal	Absent	Absent
Case 4	Increased	Present	Present
Case 5	Increased	Present	Present

4. Discussion

This case series highlights the critical role that early imaging in the first and early second trimesters can play in promptly detecting a wide range of fetal structural anomalies and placental abnormalities. Across multiple examples, prenatal diagnosis was achievable through routine ultrasound screening between 8-14 weeks' gestation, allowing timely parental counseling, further diagnostic evaluation, and planning of prenatal management.

The ability to make accurate diagnoses in the late first trimester is supported by previous research. A systematic review by Rossi et al. found that at 11-14 weeks, ultrasound could detect up to 54% of fetuses with structural abnormalities, with particularly high rates for neural tube defects (90%), abdominal wall defects (80%), and severe kidney anomalies (70%).[8] Another study demonstrated that over 82% of fetal malformations were identifiable by 18 weeks.[9] Our findings align with this literature, showcasing diverse anomalies like anencephaly, omphalocele, and polycystic kidneys that were all diagnosed before 14 weeks.

Early diagnosis has several important implications. It provides parents more time to understand and come to terms with life-altering diagnoses like lethal anomalies, which can inform deeply personal decisions around continuation or termination of pregnancy.[10] It also allows ample opportunity for genetic counseling, confirmatory testing like amniocentesis or fetal imaging, and multi-disciplinary consultation to develop tailored care plans.[11]Furthermore, certain anomalies may benefit from fetal interventions like fetal surgery or specialized neonatal management when diagnosed early enough in gestation.[3]

In some cases, we observed anomalies that were visible on initial first trimester imaging but missed, delaying diagnosis until the mid-trimester anatomy scan. This highlights the critical importance of meticulous attention to fetal anatomy at these early time points by experienced sonographers. Targeted training programs with systematic protocols for evaluating the first trimester fetus have been shown to significantly improve detection rates of major anomalies.[12]

Our findings also emphasize the importance of early placental imaging. We report a case where a large subchorionic hemorrhage prompted hospitalization and monitoring based on first trimester findings alone. Studies indicate that suspected placental abnormalities on early ultrasound like subchorionic hematomas correlate with increased risks of adverse pregnancy outcomes like preterm birth, making early recognition critical for appropriate surveillance.

A limitation of our study is that this study was conducted at a single academic institution, which may impact generalizability. Multi-center prospective studies with standardized protocols would more definitively establish the incremental yield of first trimester imaging. However, the cases we present, supported by prior literature, provide compelling evidence that incorporation of high-quality first and early second trimester ultrasounds can meaningfully increase antenatal detection of fetal and placental abnormalities. Earlier diagnosis allows for earlier intervention, ultimately optimizing prenatal care.

In conclusion, comprehensive evaluation of the fetus and placenta should begin with routine first trimester ultrasound between 11-14 weeks. Diligent inspection of fetal anatomy by experienced providers at this point is critical to maximize the prenatal detection of structural defects and placental issues that may portend significant implications for obstetric management and prenatal care. Efforts to enhance first trimester imaging should be encouraged alongside continued routine mid-trimester anatomy surveys.

5. Conclusion

This case series demonstrates the vital importance of comprehensive first and early second trimester ultrasound examinations for detection of fetal structural anomalies and placental abnormalities. Across multiple examples, we illustrate how diligent sonographic evaluation at these early gestational ages allowed prompt antenatal diagnosis of a diverse range of conditions that have significant implications for prenatal counseling, clinical management, and maternal-fetal outcomes. Early detection facilitated timely information for parents to understand the nature and prognosis of the diagnosed anomaly, providing a crucial window to consider their values and make informed, personal decisions around continuing or terminating the pregnancy. It also enabled advanced diagnostic testing, multidisciplinary consultation, and development of

individualized care plans tailored to the specific condition and clinical scenario.

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