Early detection matters; the emerging role of first-trimester ultrasound in identifying foetal anomalies: A case-based discussion

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Introduction

Ultrasound scan (USS) is a noninvasive, inexpensive diagnostic tool for screening for anatomical abnormalities providing detailed images with high sensitivity and specificity, interpreted by trained expertise (1). In prenatal care, USS is a widely accepted diagnostic tool to identify fetal structural anomalies in addition to basic assessment of pregnancy details and aneuploidy screening (1-3). The accuracy can be improved by combining abdominal and transvaginal ultrasonography, the use of high-resolution probes and 3D ultrasonography along with the experience of the operator (3).

In Sri Lanka, there is no standard guidelines for the timing of ultrasound scans during pregnancy. In most centres a routine dating scan is performed either at booking visit or in between 11 - 14 weeks of gestation. Routine anomaly scan is performed at 16 - 22 weeks to detect foetal anomalies. In current Sri Lankan practice, it is mandatory to perform anomaly scans in high-risk pregnancies. In addition, growth scans are performed from 20 weeks onwards and subsequently at various intervals as indicated.

Early detection of major anomalies has advantages in decision-making, multidisciplinary care and as an initial step of extensive evaluation (4). Foetal abnormalities are most accurately identified in well-developed foetuses during the second-trimester ultrasound scan, which typically reveals both major and minor structural anomalies. However, by this stage, the mother has often already formed an emotional attachment to the foetus, making decision-making regarding the pregnancy considerably more challenging. Because of this reason, there is an increasing trend on performing first-trimester ultrasound screening to enable earlier detection of foetal abnormalities, facilitating timely counselling, management planning, and informed parental decisions.

This case-based discussion highlights the use of first trimester scanning to detect foetal anomalies in 2 patients presented with major anatomical abnormalities. The reports elaborate on the global transition towards the first-trimester anomaly screening which is not yet widely established in Sri Lanka.

Case presentations

Case 1

A 34-year-old woman, presented in her first pregnancy to antenatal clinic at Teaching Hospital, Peradeniya. She was conceived with dichorionic diamniotic twins, and was diagnosed with omphalocele in both twins on an USS examination at 13 weeks of gestation (Figure 1). It was a planned pregnancy following intrauterine insemination. She had a non-consanguineous marriage with no family history of congenital anomalies. She was not exposed to chemicals, radiation, or infections during the pregnancy. She was not on medications other than her routing vitamin, iron and folic acid supplements.

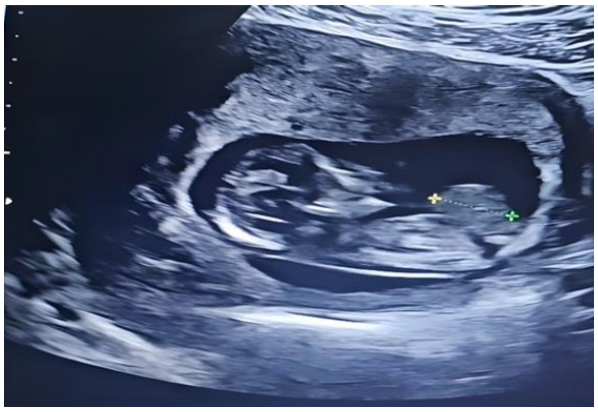

Figure 1: Ultrasound image of one of the twin foetuses at 13 weeks of gestation demonstrating an omphalocele (illustrated by the dotted line).

Case 2

A 28-year-old primigravida on routine obstetric follow-up at Teaching Hospital Peradeniya was detected to have myelomeningocele of the foetus in a routine first-trimester ultrasound scan at 12 weeks of gestation (Figure 2). It was a planned pregnancy with preconception supplements of folic acid and iron. She has no family history of congenital anomalies. She was not exposed to chemicals, radiation, or infections during the pregnancy. There was no history of medications other than her routine supplements.

Figure 2: First-trimester ultrasound image showing a neural tube defect (yellow arrow) appearing as a protrusion along the fetal spine

Discussion

In Sri Lanka, the first-trimester USS are performed mainly to confirm dates and pregnancy details such as to confirm intrauterine pregnancy. It is the usual practice to perform the second-trimester anomaly scan at 16 - 22 weeks to detect fetal anatomical anomalies. However, the current global trend is to detect major anomalies by the first-trimester anatomical survey (7). Basic foetal anatomy can be identified after 11 weeks and some major deviations including central nervous system defects, anterior abdominal wall defects, cardiac and gross limb defects, and frank hydrops can be detected by performing USS around this time (8).

Neural tube defects arise following the failure of the closure (4). The absence of the skull vault and cerebral components can be confirmed at 20 weeks of gestation, but cranial defects can be detected earlier, as cranial ossification starts around the 10th week (10).

The foetal spine can be detected by ultrasound before 10 weeks of gestation as hypoechoic parallel lines (3). Most spinal lesions are detected during the second trimester, as a bony defect or a protruded cystic mass (4). High-resolution transvaginal ultrasound can detect meningomyelocele as early as 9 - 10 weeks (3).

Congenital anterior abdominal wall defects are of 3 types: omphalocele, gastroschisis, and body stalk abnormalities. Physiological bowel herniation occurs between 9 - 10 weeks of gestation which should be differentiated before confirmation of abnormalities(5). Omphalocele consists of eviscerated abdominal organs; the proximal segment of the umbilical cord which is covered by a peritoneal membrane. In contrast, gastroschisis appears irregular as bowel loops float in amniotic fluid uncovered (3).

In addition, several other entities can be detected by the first trimester USS such as cystic hygroma, cardiac abnormalities, and severe limb abnormalities. Cervical cystic hygromas can be differentiated earlier by observing thin septations in nuchal translucency (7). Major cardiac anatomical abnormalities can be identified at 10 weeks with the earliest detection of fetal cardiac activity at

5 - 6 weeks (2). Major foetal limb abnormalities such as amelia, ectrodactyly, and fusion of lower limbs can also be screened between 9-10 weeks of gestation (3).

First-trimester ultrasound scans provide a detailed assessment of pregnancy dating, viability, location, and chorionicity. It can identify pregnancy-associated gestational trophoblastic disease and gynecological conditions (2). It is beneficial as an initial step before advanced genetic testing to identify chromosomal abnormalities (4). The detection of increased nuchal translucency combined with the assessment of the foetal nasal bone, ductus venous Doppler flow, or tricuspid regurgitation is shown to reduce the false positive rate of Down syndrome (6). Early diagnosis of major foetal anatomical abnormalities allows early referral, extensive counseling, and decision-making on the continuation of pregnancy, multi-disciplinary care, and further evaluation before the limit of legal termination (7). Early termination is safer, with less maternal morbidity and less long-term psychological impact as the emotional connection with the fetus grows with time (3). A possible disadvantage is that earlier screening will identify lethal abnormalities that would undergo spontaneous abortion such as in Down syndrome 30% at 12 weeks will die in-utero before the term (6). Exclusion of major structural abnormalities by a normal foetal USS in the first trimester can reassure the mother (7).

First-trimester ultrasound has evolved beyond early pregnancy confirmation to become a useful diagnostic tool capable of identifying significant structural abnormalities when performed with a focused and systematic approach. With the increasing availability of high-resolution ultrasound machines and skilled operators in both the government and private sectors in Sri Lanka, there is a strong rationale to shift toward routine first-trimester anomaly screening. Early detection not only facilitates timely counselling and management but also reduces the emotional and ethical challenges associated with late diagnosis in the second trimester, ultimately improving maternal and foetal outcomes.

However, several limitations must be considered when introducing first-trimester ultrasound screening for anomalies. The effectiveness of early anomaly detection largely depends on the skill and experience of the operator. This fact highlights the need for dedicated training and continuous professional development in foetal imaging. The limited availability of foetal medicine specialists, particularly in peripheral or resource limited settings, can hinder the accuracy of diagnosis and the quality of counselling provided to parents. In addition, comprehensive pre-scan counselling is essential to prepare parents for possible findings and their implications. In cases where significant anomalies are detected, the availability of appropriate referral pathways, multidisciplinary support, and management options remains variable across institutions.

Conclusions

Early detection of anatomical deviations is beneficial in terms of early referral, multidisciplinary management, less emotional impact, and decision-making on pregnancy continuation. Informed written consent has been obtained from both patients for use of case details and ultrasound images for publication.

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