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Ever-Evolving Importance of the 11 to 13+6 Weeks Fetal Ultrasonography

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ABSTRACT

Introduction: First trimester anomaly scan is a widely accepted tool for detecting aneuploidies and early fetal structural defects not detected by NIPT. Combined screening by maternal age, NT and Dual marker test is a powerful tool for fetal screening.
Aim: To examine the performance of 11 to 13+6 weeks fetal scan for detecting aneuploidies, early structural defects and maternal risk assessment of developing complications. Study was also directed to assess the maternal and fetal outcomes after early detection and interventions.
Methodology: It was a prospective observational study over 6 and half years including 847 pregnant women between 11 to 13+6 weeks gestation. The study participants were offered combined screening (NT scan + Dual Marker test) and followed up till delivery.
Results: Thirteen high risk cases (1.4%) were referred to fetal medicine specialist. CVS was done in 9 patients out of which trisomy21 was confirmed in 0.1%, turner's syndrome in 0.1% and amniocentesis confirmed trisomy18 in 0.1%. Uterine Artery PI was raised in 35 patients (4%) adding Aspirin helped prevention of pre-eclampsia in 18 patients (51.4%). 215(24.5%) women were at increased risk of FGR, adding Aspirin and high protein diet prevented FGR in 46.9% cases. 2.2% cases were at increased risk of preterm delivery and addition of vaginal progesterone prevented preterm delivery in 45% cases.
Conclusion: The use of a standardized protocol improves the sensitivity of first-trimester ultrasound screening for early structural defects and aneuploidies. By early intervention in patients at increased risk of development of PE, FGR and PTD, perinatal morbidity and mortality can be reduced.

Keywords: First trimester, Combined screening, aneuploidies, nuchal translucency.

INTRODUCTION

The 11 to 13+6 weeks scan, also referred to as the first-trimester ultrasound, represents a cornerstone of modern prenatal care. This scan is critical for the early detection of chromosomal abnormalities and structural defects that are not identifiable through Non-Invasive Prenatal Testing (NIPT). Among the key parameters evaluated during this period, increased Nuchal Translucency (NT) measurements have been strongly associated with chromosomal aberrations, structural congenital anomalies, and cardiac defects. Moreover, the scan provides valuable insights into the risks of pregnancy complications such as pre-eclampsia, fetal growth restriction (FGR), and preterm birth (PTB). When combined with maternal age

and biochemical markers through dual marker testing, the screening process achieves robust efficacy in identifying trisomies 21, 18, and 13.[1]

The integration of advanced ultrasound techniques with biochemical assessments has elevated the diagnostic and prognostic capabilities of the first-trimester scan. As a result, it offers not only enhanced foetal health assessments but also the risk of fetal loss.

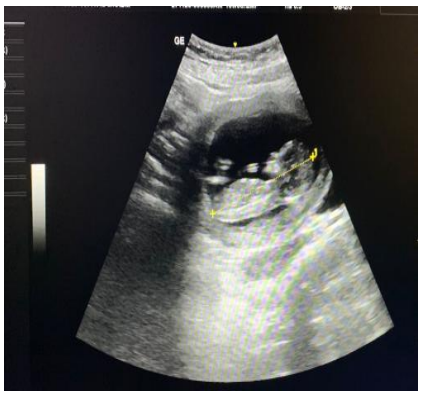
Study Aims and Objectives

This study aims to comprehensively evaluate the performance of the 11 to 13+ weeks scan in detecting foetal aneuploidies and early structural defects. Additionally, it seeks to assess the maternal risk of developing pre-eclampsia, FGR, and PTB, while analysing the maternal and foetal outcomes following timely detection and intervention. These objectives are critical for advancing prenatal care practices, especially in resource-limited settings where early screening is less commonly implemented.

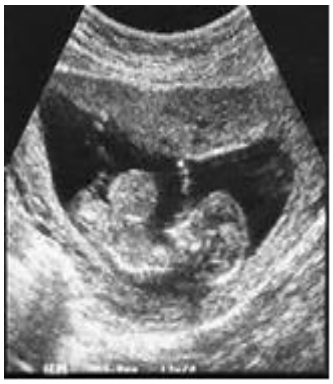
MATERIALS AND METHODOLOGY

The study was conducted as a prospective observational investigation spanning 6.5 years, from May 2016 to January 2023. A total of 874 antenatal patients were enrolled, all within 11 to 13+6 weeks of gestation and with a Crown Rump Length (CRL) between 45 and 84 mm. Maternal demographics (age, height, weight, BMI, MAP) were taken into account and all participants were subjected to thorough history taking (history of - preterm delivery / PE/ still birth / chromosomal abnormality, DM / SLE / APLA / hypertension, smoking, method of conception), high risk factors included – BOH, age >35 yrs, BMI >23 Kg/m^2, MAP>105mmHg. After this they were offered combined screening, which included NT measurement and dual marker testing, after receiving thorough counselling and providing informed consent.

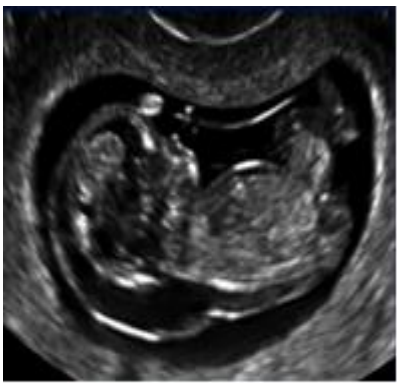
Comprehensive ultrasound evaluations, both transabdominal and transvaginal, were performed using the Versana Essential GE Machine. These scans confirmed foetal viability, ensured accurate dating, and provided detailed placental and uterine assessments and cervical length. Blood samples were analysed for Pregnancy-Associated Plasma Protein-A (PAPP-A) and free β-hCG levels, which were integrated with NT measurements to calculate risks using the FMF UK software (version 2.81).



Ultrasound showing normal CRL, NT, NB

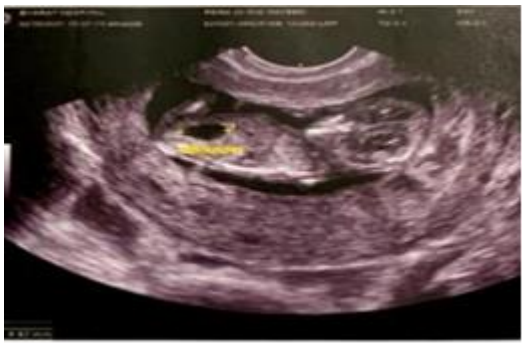


Ultrasound showing exomphalos



Ultrasound showing Increased NT

Ultrasound showing negative `a' wave of DV



Ultrasound showing fetal megacystitis

OBSERVATIONS AND RESULTS

The demographic analysis (table 1) revealed that the majority of participants (90.3%) were aged between 20 and 30 years, with only 9.3% aged between 31 and 40 years, and 0.2% aged 41 years or above. Primigravida patients constituted 58.6% of the cohort, while multigravida patients accounted for 41.4%. Key clinical findings included a 3.37% prevalence of pre-existing medical disorders, 4.9% incidence of bad obstetric history (BOH), and 6.6% cases of low-lying placenta.

Among the foetal anomalies detected (table2), major anomalies included acrania (0.3%), spinal defects (0.2%), abdominal wall defects (0.1%), exomphalos (0.1%), and hydrops (0.2%). Minor anomalies included increased NT (0.5%), absent nasal bone (0.3%), megacystis resolved on Level II ultrasound (0.1%), ductus venosus 'a' wave negativity (0.4%), and tricuspid regurgitation (0.1%). Distribution of NT measurements (table3) revealed that 96% of the cohort had values within the normal centiles, while 1% presented with significantly elevated measurements exceeding the 99th centile.

Thirteen patients (1.4%) were classified as high-risk (flowchart1) and referred to foetal medicine specialists. Chorionic villus sampling (CVS) was performed in nine cases, leading to the diagnosis of Down's syndrome in one case (0.1%) and Turner's syndrome in another (0.1%). An additional case of Edward's syndrome (0.1%) was identified through amniocentesis. The overall detection rate was 99%, with a false positive rate of 1%.

Table 1: Maternal demographics

Parameter	**Number (%)**
Age	
20 – 30 years	790 (90.3%)
31 – 40 years	82 (9.3%)
≥41 years	2 (0.2%)
Gravida	
Primigravida	513 (58.6%)
Multigravida	361 (41.4%)
Pre-existing medical disorders	29 (3.37%)
BOH	43 (4.9%)
Low-lying placenta	58 (6.6%)

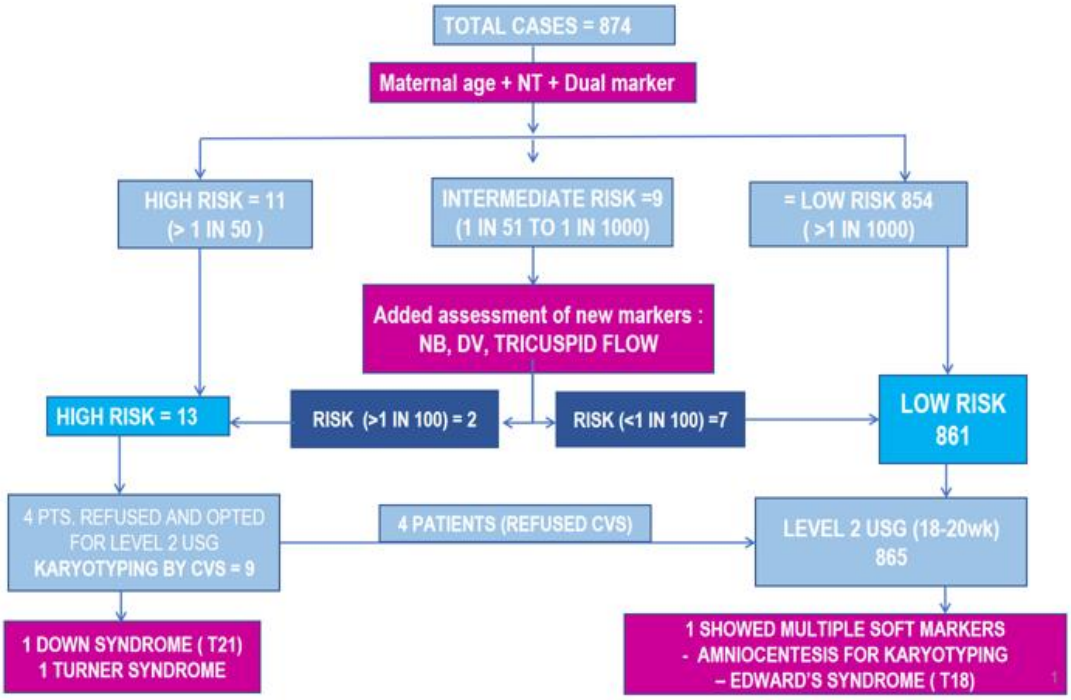
Table 2: Showing major and minor fetal defects observed

Major Fetal Anomalies (9)	
Acrania	3 (0.3%)
Spinal Defects	2 (0.2%)
Abdominal Wall Defect	1 (0.1%)
Exomphalos	1 (0.1%)

Hydrops	2 (0.2%)
Minor Anomalies (14)	
Increased NT	5 (0.5%)
Absent Nasal bone	3 (0.3%)
Megacystis (resolved on level II scan)	1 (0.1%)
DV negative	4 (0.4%)
Tricuspid Regurgitation	1 (0.1%)

Table 3: Showing Nuchal transluscency measurements of study participants

NO. OF PATIENTS	NT MEASUREMENTS	CENTILE
807	1.2 to 2.0 MM	20 to 90 centile
62	2.1 to 2.9 MM	30 to 95 centile
1	3 MM	96 centile
1	3.2 MM	97 centile
1	4.5 MM	99 centile
1	6.8 MM	> 99 centile
1	7 MM	> 99 centile



Flowchart 1: demonstrating study process and results

In total, 51 chromosomal abnormalities were identified in the study population, including 33 cases of trisomy 21, eight of trisomy 18, six of sex chromosome abnormality, one of triploidy and three of other unbalanced abnormalities. The detection rate and false-positive rate (FPR) for trisomy 21 were 93.8% and 4.84%, respectively, using biochemical markers and NT, and 100% and 3.4%, respectively, using biochemical markers, NT, NB, TR and DV flow. This approach not only improves detection rates but also minimises false positives, reducing unnecessary invasive procedures.

DISCUSSION

The above findings highlight the significant benefits of integrating newer markers such as nasal bone visibility, ductus venosus flow, and tricuspid regurgitation assessment into routine aneuploidy screening protocols and is also supported by literature available from past research studies (table 4).

Table 4: Screening performance of our study compared with other major studies

Reference	Gestational age	Ultrasound marker	T21 (DR%)	FPR (%)
Wald et al[2]	10 - 14	NT	85	6
Kagan et al[3]	11 - 14	NT, NB	91	2.5
Maiz et al[4]	11 - 13	NT, DV	96	3
Falcon et al[5]	11 - 14	NT, TR	95	5
Ghaffari et al[1]	11 - 14	NT, NB, TR, DV	90	3
Dahiya et al	11 - 13+6	NT, NB, TR, DV	93.8	4..8

Ghaffari et al screened 13,706 fetuses for chromosomal abnormalities along with maternal serum biochemical markers and maternal age, in combination with NT, NT + NB, NT + NB + TR, and NT + NB + TR + DV flow over five years. They found that NT thickness (> 2.5 mm) is the single most effective marker with a detection rate of near 70% (and a false-positive rate of 5.9%). They concluded that while individual biochemical markers have low detection rate, combined biochemical markers yield a DR of 69% at a FPR of 5.9%.

Research conducted by Chen et al[6] also supports the hypothesis and yielded similar results. They observed five cases of nasal bone loss in 9 cases of trisomy 21, 5 cases with three tricuspid regurgitation, 4 cases of venous ductus a wave flow reverse, 3 cases of fetal nasal bone loss accompanied by tricuspid regurgitation and venous ductus a wave flow reverse. One case of nasal bone loss in 2 cases of trisomy 18, 2 cases were tricuspid regurgitation and venous ductus a wave flow reverse. Two cases in 4 cases of 45X had venous ductus a wave flow reverse. There were 8 cases (0.16%) nasal bone absence in 4983 cases of normal karyotype fetus, 48 cases (0.96%) of tricuspid regurgitation and 44 cases (0.88%) of venous ductus a wave flow reverse. Thirty-two cases in 40 cases (80%) of fetal congenital heart disease were tricuspid regurgitation, 30 cases of venous ductus a wave flow reverse (75%). Eight cases of nasal bone absence normal karyotype fetus were found the nasal bone at 20 weeks gestation.

Alldred et al[7] also combined ultrasound markers with serum markers, especially PAPP-A and free ßhCG, and maternal age. They detect about nine out of 10 Down's affected pregnancies for a fixed 5% FPR. Although in their study, the absence of nasal bone appeared to have a high diagnostic accuracy (only five out of 10 affected Down's pregnancies were detected at a 1% FPR).

Despite these advancements, the implementation of first-trimester combined screening remains limited in low-resource settings, where the majority of anomaly scans are performed in the second trimester. Consequently, the potential for early interventions is often missed.

Early detection and management of pregnancy complications such as pre-eclampsia, FGR, and PTB through combined screening can significantly reduce perinatal morbidity and mortality. The study underscores the need for greater public awareness and policy support to make these advanced screening methods more accessible and affordable, particularly in resource-constrained environments.

CONCLUSION

Risk assessment using a combination of NT measurements and biochemical markers demonstrates excellent screening performance for detecting chromosomal anomalies and structural defects. Standard NT measurement can be applied when other complementary infrastructures are not available. The addition of advanced ultrasound markers such as nasal bone visibility, tricuspid regurgitation, and ductus venosus flow further enhances diagnostic accuracy. Comprehensive pre- and post-test genetic counselling remains essential for guiding patient management. By focusing on early detection and intervention, this approach holds significant potential for

improving maternal and foetal outcomes and addressing critical gaps in prenatal care.

Declaration by Authors

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Conflict of Interest: The authors declare no conflict of interest.

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