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Original Research Article

Prevalence of fetal ultrasonographic soft markers in antenatal women and their pregnancy outcomes: a cross-sectional study

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ABSTRACT

Objectives: Estimating soft marker prevalence during first and second trimester ultrasound scans and determining the relationship between nuchal translucency (NT) measurements and sociodemographic variables in the antenatal population receiving care at our facility.
Methods: This hospital-based observational study, conducted from January 2020 to November 2021, included 300 pregnant women receiving antenatal care at our institute. Soft markers studied included NT and absence of nasal bone for the first trimester and echogenic intracardiac focus (EICF), hyperechogenic bowel, choroid plexus cyst (CPC), mild renal pyelectasis, and limb shortening for the second trimester. For ultrasonographic examination, a HITACHI – ALOKA -6 USG equipment was used via transabdominal route. Wherever applicable, independent sample t test, chi-square test, and fisher's exact test were used to examine demographic factors and NT. To analyse the data, S.P.S.S version 23 was utilized.
Results: The mean age of the participants was 24.96 years, and 41% (n=123) were primigravida and 59% (n=177) were multigravida. Most pregnant women (79%, n=237) were low-risk. Prevalence of Increased NT was 2.66% (n=9), hypoplasia or absence of NB was none, EICF was 2.33% (n=7), Hyperechogenic bowel was 0.33% (n=1), CPC was none, mild renal pyelectasis was 0.66% (n=2), and limb shortening was 0.33%. Relationship between obstetric risk factors and increased NT (2=12.22, p=0.006) and mean age of participants with increased NT (t=-2.625, p=0.009) was statistically significant**.**
Conclusions: First-trimester fetal NT screening should be done for pregnant women over 30 and multigravida. Soft markers in pregnant women need a thorough diagnostic evaluation to rule out fetal abnormalities.

Keywords: Prevalence, Pregnant women, Nuchal translucency, Hyperechogenic bowel, Pyelectasis, Pregnant women

INTRODUCTION

Ultrasonography, a widely utilized technique for prenatal diagnosis, assumes a critical role in the detection of fetal malformations and contributes significantly to the preservation of fetal well-being.[1] In a subset of pregnancies, estimated to be around 5%, routine fetal ultrasound screening identifies a morphological characteristic in the fetus that is not inherently concerning but necessitates additional diagnostic evaluation to determine whether it is a normal variant (false-positive screening) or indicative of a serious fetal condition, such as a chromosomal anomaly (true-positive screening). The morphological features in question are commonly known as "soft markers" (SM) in the scientific literature.[2] In the first trimester, increased nuchal translucency and a short nasal bone have been observed as potential soft markers. Additionally, in the second trimester, hyperechogenic bowel, short nasal bones, renal pyelectasis, intracardiac foci, short femur, nuchal fold, or mild cerebral ventriculomegaly have been identified as potential markers.[3-5] USMs (Ultrasonographic Soft Markers) have

been found to have a strong correlation with the detection of fetal chromosomal disorders and the occurrence of adverse pregnancy outcomes.[6] However, the prevalence of soft markers in antenatal women located within the vicinity of our tertiary care institute is not well documented.

Therefore, we conducted this study with the objectives of estimating the prevalence of soft markers detected during ultrasound scans conducted in the first and second trimesters and determining the association between nuchal translucency (NT) measurements and various sociodemographic variables among the antenatal population receiving care at our tertiary healthcare institution.

METHODS

Study type

This hospital-based prospective observational study was conducted in the department of Obstetrics and Gynaecology with coordination from the department of Radiology at our tertiary care institute.

Study duration

The study was carried out over a period spanning from January 2020 to November 2021. The present study was started subsequent to obtaining the necessary approval from the institutional ethics review committee, in accordance with the guidelines set forth by the Helsinki Declaration of 1975, as subsequently revised in 1983.

We enrolled all pregnant women with singleton pregnancies who were seeking antenatal care during the first and second trimesters at the outpatient department (OPD) of the Department of Obstetrics and Gynaecology at our institution for the period of the study.

Participants who demonstrated failure to return within the designated timeframe of 18 to 23+6 weeks, experienced pregnancy loss at any point during the study, were unwilling, or exhibited a lack of follow-up were excluded from the analysis. Written and informed consent was obtained from all participants prior to their involvement in the study. The present study employed non-probability purposive sampling to recruit a total of 300 pregnant women who were receiving antenatal care at our institute.

The selection of participants was based on the specific eligibility criteria mentioned earlier that were established for this research activity. The collection of data regarding maternal age, type of pregnancy, gravidity, ultrasonography outcomes, and other pertinent information regarding the present study, in addition to the infant situation at the time of birth, was done by administering a pre-designed and pre-tested semi-structured questionnaire to the participants.

With regard to the standard ultrasonography performed during the first trimester, the USMs evaluated increased nuchal translucency and the absence of nasal bone. Second-trimester USMs included in this study were EICF (Echogenic Intracardiac Focus), hyperechogenic bowel, choroid plexus cyst, mild renal pyelectasis and limb shortening. A team of knowledgeable obstetricians with a minimum of two years of clinical ultrasound experience performed the ultrasound examinations. Whenever a fetus was identified with a suspected malformation or abnormal ultrasonic soft markers were observed, it underwent further examination by a senior obstetrician employed in the department. For the ultrasonographic examination, a HITACHI-ALOKA-6 USG equipment was used via transabdominal route. The study involved the systematic monitoring of patients through regular antenatal visits and post-delivery assessments in order to identify any significant congenital malformations in the fetus and assess the overall outcome of the pregnancy.

In the present investigation, increased NT thickness was defined as NT thickness ≥95th percentile.[7] The absence or underdevelopment of the nasal bone (NB) was determined by the lack of visibility or insufficient brightness of the nasal bone compared to the surrounding skin in the mid-sagittal part of the fetal face or the coronal section of the retro nasal triangle (RNT).[8] The term echogenic intracardiac focus EICF for the study referred to a specific area inside the fetal heart, namely in the region of the papillary muscle, where the echogenicity was similar to that of bone.

This characteristic was seen in either one or both ventricles of the fetal heart.[9] Hyperechogenic bowel for the study was defined as a condition in which the fetal intestine exhibited regions of echogenicity that were uniform and either equivalent to or greater than the echogenicity of the surrounding bone.[10] Cystic lesions known as choroid plexus cysts (CPCs) have been identified through sonographic examination as distinct, diminutive cysts located within the choroid plexus situated inside of the lateral cerebral ventricles.[11]

In the context of the investigation, the condition known as mild renal pyelectasis (PYE) was deemed to be present when the measurement of the anterior posterior renal pelvis diameter (APRPD) exceeded 1.5 mm.[12] The phenomenon of limb shortening was said to be present in cases where the measured-to-expected ratio is equal to or less than 0.91 for the femur, and equal to or greater than 0.89 for the humerus.[13] For the calculation of the prevalence of different types of USMs, it was calculated by, Prevalence (%)=(Number cases having the type of soft marker/Total number of participants covered in the study)×100

Statistical analysis

The data for this study was collected through a systematic process, compiled, tabulated, and subsequently entered

into a Microsoft excel spreadsheet. To analyse the data, the statistical package for social sciences (SPSS) version 23 was utilized. The results of the study were presented using descriptive statistics, specifically in terms of frequency and percentages and for age mean and standard deviation was used.

In order to investigate the potential relationship between demographic variables and NT, the independent sample t test, the chi-square test and the fisher's exact test were applied wherever applicable. In order to determine the statistical significance of the association, a threshold of $p<0.05$ at the 95% confidence interval (CI) was employed.

RESULTS

The mean age of the participants (n=300) was 24.96 years, with 44% of participants in the age group of 26–30 years. With regard to parity, 41% (n=123) of participants were primigravida and 59% (n=177) were multigravida.

The majority of the pregnant women fell into the category of low risk, i.e., 79% (n=237), followed by postdated pregnancy (Table 1).

Regarding the prevalence of the different soft markers, the prevalence of increased NT was 2.66% (n=9), hypoplasia or absence of NB was seen in none, EICF prevalence in our study was 2.33% (n=7), prevalence of Hyperechogenic bowel was 0.33% (n=1), CPC was seen in none, mild renal pyelectasis prevalence was 0.66% (n=2), and prevalence of limb shortening was 0.33% (n=1) (Figure 1) (Table 2).

Table 1. Socio demographic variables of the participants (n=300).

Variables	N (%)
Age (in years)	
<20	33 (11)
21-25	123 (41)
26-30	132 (44)
31-35	8 (2.64)
>35	4 (1.33)
Parity	
Primigravida	123 (41)
Multigravida	177 (59)
Risk factors present in pregnancies	
GDM	6 (2)
Hypothyroidism	3 (1)
Preeclampsia	14 (4.66)
Postdated	28 (9.33)
Prom	6 (2)
Preterm	6 (2)
Low risk	237 (79)

Table 2: Prevalence of different types of USMs.

Soft markers	N (%)
Increased nuchal translucency	9 (2.66)
Echogenic intracardiac focus	7 (2.33)
Hyperechogenic bowel	1 (0.33)
Mild renal pyelectasis,	2 (0.66)
Limb shortening	1 (0.33)
Hypoplasia/ Absence of nasal bone	0 (0)
Choroid plexus cyst	0 (0)

Table 3. Association of NT with different sociodemographic variables.

Variable	Increased nuchal translucency	Normal nuchal translucency	Test significance, P value
Mean age	28.89±5.41	25.24±4.06	t=-2.625, p=0.009
Gravidity	**N (%)**	**N (%)**	
Primipara	2 (1.6)	121 (97.50)	$\chi2$=1.35, p=0.244
Multigravida	7 (3.9)	170 (96.04)	
Risk Factors	**N (%)**	**N (%)**	
Gestational diabetes mellitus	1 (16.660)	5 (83.33)	
Hypothyroidism	1 (33.33	2 (66.66)	$\chi2$=12.32, p=0.006
Preeclampsia	1 (7.14)	13 (92.85)	
Low risk	6 (25.31)	231 (97.46)	

Table 4: Pregnancy outcome of the studied antenatal population.

Ultrasonographic structural abnormality	Total cases	Abnormal outcome	Abnormal rate
Increased nuchal translucency	9	1	11.11
Echogenic intracardiac focus	7	0	0.0
Hyperechogenic bowel	1	1	100
Limb shortening	1	1	100

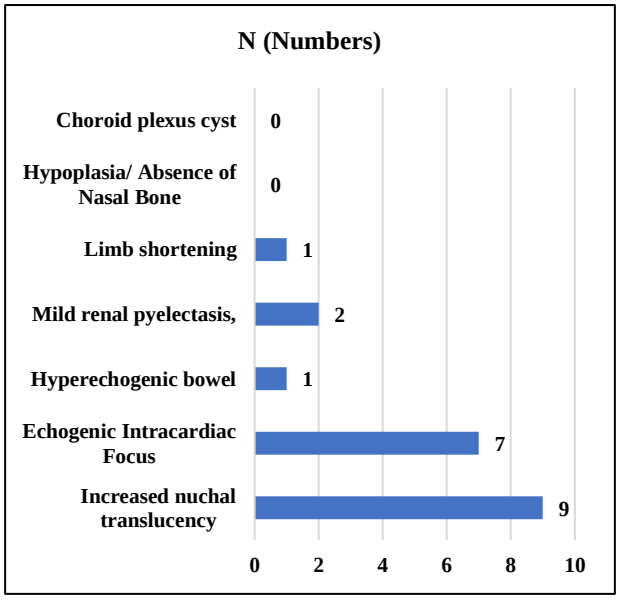

Figure 1. Prevalence of different types of USMs.

The mean age of the participants with normal NT was 25.24±4.06 years, while the mean age of participants with increased NT was 28.89±5.41 years, and this difference was statistically significant (t=-2.625, p=0.009). The relationship between the parity of the participants and NT status was non-significant (2=1.35, p=0.244). However, the relationship between the obstetric risk factors and increased NT was statistically significant (2=12.22, p=0.006) (Table 3). Participants who were having USMs were followed up to learn about the outcome of the pregnancy. The most prevalent soft marker was increased NT seen in 9 cases (2.6%), out of which 1 case (11.11%) were having adverse outcome of different types. Out of 7 cases of ECIF, no case had adverse outcome one with Hyperechogenic bowel developed Down syndrome which was also associated with increased NT and one case (100%) showing limb shortening had MTP (Table 4).

DISCUSSION

In this study, a total of 300 antenatal women were enrolled. We evaluated them for NT and NB at 11 to 13+6 weeks and other antenatal soft markers at 18 to 22+6 weeks. The prevalence of increased NT, EICF, hyperechogenic bowel, mild renal pyelectasis, and limb shortening was 2.66%, 2.33%, 0.33%, and 0.33%, respectively. Hypoplasia NB and CPC were found in none. Increased NT was found to be significantly associated (p<0.05) with the age and obstetric risk factors in the study. The prevalence of NT in our study was similar to the study by Sulaiman B et al, which was a prospective cohort study at the Obstetrics and Gynecology Department of Usmanu Danfodiyo University Teaching Hospital, Sokoto.[14] The nasal bone was present in all fetuses; this could be attributed to the small sample size of our study. In a study conducted by Sara Masihi et al, 2314 patients were enrolled and the nasal bone was absent in all three cases with trisomy 21 and in 6 of 2173 cases with the normal phenotype (0.3%).[15]

The prevalence of ECIF in our study is in line with the study by Roya Fallahian et al, 1000 pregnant women resorting to Mahdiye Hospital for the second trimester ultrasound screening from 2014 to 2015 were selected for this descriptive research and studied for the presence of ECIF (prevalence ECIF=3.8%). The prevalence of hyperechogenic bowel in our study was similar to that of Brigitte Simon-Bouy et al, where hyperechogenic fetal bowel is prenatally detected by ultrasound during the second trimester of pregnancy in 0.1% to 1.8% of fetuses.[16,17] Prevalence of mild renal pyelectasis in our study was similar to the study conducted by Signorelli M et al.[18] In our study, advanced age and obstetric risk factors were found to be associated with an increase in NT. Age above 30 years had a higher chance of increased NT, which was comparable to studies done by Pranav P, Pandya et al, RJM Snijderset et al. Among 9 fetuses with increase NT, one has abnormal outcome (trisomy 21) which has associated hyperechogenic bowel and diagnosed case of gestational diabetes mellitus.[19,20]

Two cases with the soft marker ECIF in the study have no congenital heart defects. In a study by TC Winter, Anderson et al, an echogenic intracardiac focus was seen in 147 of the 3,192 karyotypically normal fetuses (4.6%) and 16 of the 53 fetuses with trisomy 21 (30%). However, in a study by Anderson N. and Jyoti et al, isolated EIF in women aged 18 to 34 years was not associated with increased risk for trisomy 21 in mid gestation.[21,22]

CONCLUSION

Routine first trimester screening for fetal nuchal translucency should be done pregnant women especially in above 30 years of age and multigravida. By identifying increased nuchal translucency measurements, healthcare providers can offer further diagnostic testing or counselling to expectant mothers, enabling them to make informed decisions regarding their pregnancy. The identification of soft markers in pregnant individuals necessitates a comprehensive diagnostic investigation to exclude the presence of fetal abnormalities. This approach is crucial in order to mitigate the risks associated with perinatal mortality and morbidity.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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