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FETAL NUCHAL TRANSLUCENCY BY ULTRASOUND A SCREENING TOOL FOR FETAL ABNORMALITIES

KEYWORDS: nuchal translucency, ultrasound, fetal abnormalities

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ABSTRACT

AIM: *To assess fetal nuchal translucency in an unselected population of pregnant women with viable singleton pregnancy and usefulness of increased Nuchal translucency in identifying abnormal fetuses.*

SUBJECTS & METHODS :- *A Prospective study consisting of 100 antenatal women was conducted in kasturba Gandhi hospital, Chennai, after obtaining ethical clearance from IEC , madras medical college. Women with NT >95th percentile for that gestational age and CRL, were considered to be at high risk (screen positive).For screen positive women, fetal karyotyping was done after counseling them regarding the risk for having an anomalous fetus. Chi-square test and student t test were used to find the statistical significance in the noted difference of anomalies associated with the fetuses with increased NT using statistical software namely SPSS version 15.A sample size of 100 was arrived using G power 3.0.10 software which determines the significance size at 0.01 and power of 0.99.*

RESULTS :- *The mean age for women with normal NT : 24.4 years (SD-4.4), mean age for women with abnormal NT : 28.8 years(SD -6.5).All fetuses with increased NT had abnormal outcome showing that NT is an effective method of screening for fetal abnormalities. statistically significant difference seen in the incidence of increased NT among antenatal women who were older than 30 years and the P value <0.01. The diagnostic value of increased NT in relation to cardiac defects sensitivity is 100%, specificity is 96.9%, positive predictive value is 40% and the p value is < 0.001. Increased NT was significantly associated with abnormal fetuses was statistically significant p<0.001 and with syndromic fetus p value <0.001.*

CONCLUSION:-*Our study confirms that all fetuses with increased NT had adverse pregnancy outcome. 40% had chromosomal anomalies .Isolated cardiac defects were identified among 40% fetuses,20% had spontaneous miscarriage. Incidence of increased NT is higher in women more than 30 years (28.6%), against women less than 30 years (3.1%), indicating higher risk for fetal abnormalities in the former group.*

INTRODUCTION:-

Prenatal diagnosis of fetal structural malformations during either the first or the second trimester has helped to reduce perinatal morbidity and mortality and also helped in early termination of an anomalous fetus. Although many major fetal defects are diagnosable in the first trimester, the diagnostic accuracy is significantly higher in the mid-second trimester owing to the larger size and more advanced development of the fetus.

Mid –second trimester screening method is not an ideal tool and search is on for early markers. A great deal of interest has been directed towards shifting prenatal screening for chromosomal abnormalities and fetal structural abnormalities to the first trimester (11-14 weeks of gestation) using the sonographic measurement of the fetal nuchal translucency(NT), alone or in combination with serum markers (Pregnancy associated plasma protein A {PAPP-A} + free beta human chorionic gonadotrophin {beta-HCG}).

Sonographic screening of aneuploidy became a reality in 1985 when Beryl Benacerraf demonstrated thickened nuchal fold in a downs syndrome fetus.

Dr. Langdon Down (it is after his name that downs syndrome has been named) 100 years back , reported that skin of affected fetus at the back of the neck was too large and swollen. This excess skin thickness can be easily studied by ultrasound as nuchal translucency (at 11-14 weeks of gestation).

Fetal nuchal translucency thickness at 11-14 weeks scan has been combined with the maternal age to provide an effective method of assessing the risk for chromosomal anomalies. In addition increased nuchal translucency identifies a higher proportion of major cardiac defects, skeletal defects and a wide range of genetic syndromes. For example Nicolaides (2004) reported approximately 90% sensitivity for trisomies 18 & 13 with a 1% false positive rates. There is a strong association between increasing nuchal translucency and fetal cardiac anomalies. (atzei and colleagues 2005; simpson and colleagues 2009).

It is widely accepted that the measurement of NT to screen fetal abnormalities should be combined with a search for detectable malformations. The American college of obstetricians and gynaecologists recommends that when the nuchal translucency measurement is 3.5mm or greater with a normal fetal karyotype, then targeted sonographic examination , fetal echocardiography or both should be considered.

Visualization of normal fetal anatomy in the first trimester provides reassurance to the patient and reduces anxiety. Early detection of fetal structural malformations allows timely referral to a tertiary care centre.

Assessment of risk for chromosomal defects in the first trimester rather than second trimester provides option for earlier invasive diagnostic testing and consequently results in fewer traumas for those couples choosing termination of affected pregnancy. NT as a screening tool for fetal aneuploidy are less sensitive in younger women because of lower prevalence rates. Gestational age also affects the accuracy of downs syndrome detection. Testing sensitivity is approximately 5% lower if performed at 13 weeks instead of 11 weeks. A study group of the Royal college of obstetricians and gynaecologists has recommended that there is now sufficient evidence to support routine first trimester nuchal translucency screening with appropriately trained sonographers using high resolution equipments, provided that the results should be subjected to an external agency for regular audit.

REVIEW OF LITERATURE:-

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NUCHAL TRANSLUCENCY – DEFINITION :-

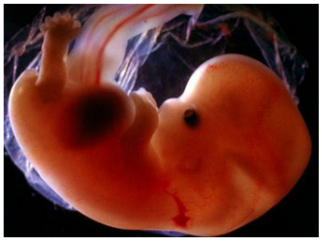
The nuchal translucency is defined as the maximum thickness of the subcutaneous translucency between the skin and soft tissues overlying the cervical spine of the fetus and is typically observed in the first trimester (11 to 14 weeks).

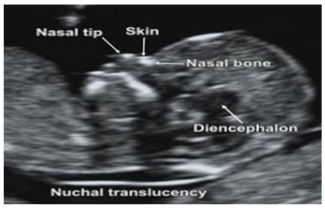
NORMAL NT MEASUERMENTS:-

Fetal nuchal translucency thickness increases with crown-rump length (CRL) and therefore it is essential to take gestation into account when determining whether a given translucency thickness is increased. The 50th percentile values for NT thickness are 1.2mm and 1.9 mm for a CRL of 45mm (11th week) and 85 mm (13 weeks + 6 days) respectively, and the 95th percentile values are 2mm and 2.8mm respectively.

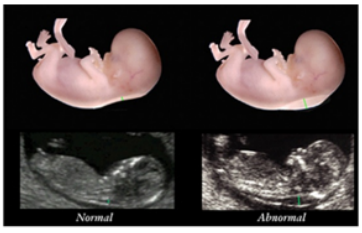
Currently the more accepted method is to base the cut off on a progressive rise, using 95thpercentile as the threshold for an abnormal measurement resulting in a more sensitive and specific indicator for the detection of anomalous fetus.

First Trimester Fetus





Midsagittal view for measuring NT by ultrasound



Normal NT Abnormal NT

RISK FOR TRISOMY BASED ON NT:-

Less than 2.5 mm : risk is 5 times lesser for that age.
More than 2.5 mm : 12 fold increased risk.

PATHOPHYSIOLOGY:-

The heterogeneity of conditions associated with increased NT suggests that there may not be a single underlying mechanism for the collection of fluid under the skin of the fetal neck. Possible mechanisms include,

1. Cardiac dysfunction
2. Venous congestion in the head and neck
3. Altered composition of extracellular matrix
4. Failure of lymphatic drainage
5. Fetal anaemia
6. Fetal hypoproteinemia
7. Fetal infection

TRANSIENT NATURE OF NUCHAL TRANSLUCENCY BETWEEN 11-14 WEEKS:-

There is a brief opportunity between 11-14 weeks of gestation when the fetal lymphatic system is developing and the peripheral resistance of the placenta is high. After 14 weeks the lymphatic system is likely to have developed sufficiently to drain away any excess fluid and changes to the placental circulation. So after this time any abnormalities causing fluid accumulation may seem to correct themselves and then go undetected by measuring NT.

The reasons for selecting 14 weeks of gestation as the upper limit are :-

1.To provide women who have affected fetuses, the option of an earlier and safer form of termination.

2.The incidence of abnormal accumulation of nuchal fluid in chromosomally abnormal fetuses is lower at 14-18 weeks of gestation than at <14 weeks of gestation .

3.The success rate for taking a measurement at 11-14 weeks of gestation is 98% to 100% which falls to 90% at more than 14 weeks of gestation because the fetuses is often in a vertical position which makes it more difficult to obtain the appropriate image.

The reason for selecting 11 weeks of gestation as the earliest gestation was:

Screening necessitates the availability of diagnostic test and in the early 1990s , it was appreciated that CVS before 10 weeks of gestation was associated with transverse limb reduction defects. It was realized that many major fetal abnormalities could be diagnosed at NT scan the minimum gestation was 11 weeks.

MEASUREMENT OF NUCHAL TRANSLUCENCY:-

Nicolaides et al described the measurement criteria necessary to achieve uniformity of the measurement.

1. NT can be measured successfully by transabdominal ultrasound examination in about 95% of cases .

2. The minimum fetal CRL should be 45mm and the maximum of 84 mm. The optimal age for measurement of fetal NT is 11 to 13 weeks + 6 days.

3. Fetal NT increases with CRL and therefore it is essential to take gestation into account when determining whether a given translucency thickness is increased.

4. A good mid saggital section of fetus as for measurement of fetal CRL , should be obtained and the NT should be measured with the fetus in neutral position. When the fetal neck is hyperextended the measurement can increase by 0.6mm and when the neck is flexed , the measurement can decrease by 0.4mm(whitlow et al 1998). Only the fetal head & upper thorax should be included in the image.

5. Calipers are placed at the inner borders of the NT space.

NT – CALCULATION OF PATIENT SPECIFIC RISK :-

1. Every woman has high risk that her fetus has chromosomal defect . In order to calculate the individual risk ,it is necessary to take into account the background or a "priori risk", which depends on maternal age and gestation and multiply this by a series of factors or likelihood ratios,which depends on the results of a series of screening tests carried out during the course of pregnancy.

2. The likelihood ratio for a given sonographic or biochemical measurement is calculated by dividing the percentage of chromosomally abnormal fetuses by the percentage of normal fetuses with that measurement.

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3. Prospective studies in more than 2,00,000 pregnancies , including more than 900 fetuses with trisomy 21, have demonstrated that NT screening can identify more than 75% of fetuses with trisomy21 , for a false positive rate 5%.

OUTCOMES OF FETUSES WITH INCREASED NT:-

1. Chromosomal defects
2. Fetal death
3. Lethal fetal abnormalities
4. Developmental delay
5. Chromosomally normal fetuses with increased NT
6. Cardiac defects, pulmonary defects, abdominal wall defects, Central nervous system defects, gastrointestinal defects, fetal anaemia, genitourinary defects, neuromuscular defects, Facial defects, skeletal defects.

METHODS OF SCREENING	DETECTION RATES (DR%) for Trisomy 21
Maternal age (MA)	30%
NT alone	70%
Maternal serum free beta HCG + PAPPA-A at 11-13 weeks	70%
Maternal age + maternal serum biochemistry at 15-18 weeks	50-70%
Maternal age + fetal NT at 11-13 weeks	70-80%
Maternal age+ fetal NT + maternal serum free beta HCG+PAPP-A at 11-13weeks	85-90%
Maternal age+ fetal nasal bone(NB)+ fetal NT at 11-13 weeks	90%
Maternal age +fetal NT+ maternal serum beta HCG+ PAPP-A at 11-13 weeks	95%
Stepwise sequential screen NT + 1st trimester serum with risk calculated, then quadruple screen with final risk including 1st & 2nd trimester results	95%
Fully integrated screen PAPP-A and NT in the 1st + quadruple in the 2nd trimester with 1 risk calculated	96%
Contingent sequential screen 1st trimester screen and quad test	94%

REVIEW OF STUDIES:-

The following table summarizes the details of diagnostic accuracy of increased NT among various studies.

study	Study population	GA (wks)	NT (cut-off)	sensitivity	specificity	PPV	FPR
Nicolaides etal (1994)	1273	10-13	>or= 3mm	85%	95.9%	35.5 %	5%
Pandya et al (1995)	1763	10-13	>or= 2.5mm	75%	92%	-	8%
Bewley etal (1995)	1127	8-13	>=mm	40%	94%	-	6.1%
Taipele etal (1997)	2281	10-14	>=3 mm	62.3%	99.4%	24%	0.6%
Economoides et al (1998)	96,127	11-14	>99th centile	81%	99.6%	-	0.4%
Schwarzler et al (1999)	4523	10-14	>2.5 mm	76%	95.3%	8.2%	4.7%
Naidoo P et al (2008)	428	11-14	>95th	85%	99.7%	25%	-

MATERIALS & METHODS:-

A prospective study consisting of 100 antenatal women was conducted in government kasturba Gandhi hospital,Chennai after obtaining ethical clearance from IEC , madras medical college.

Study period :- August 2015 to August 2016

INCLUSION CRITERIA:-

1. Antenatal women with reliable dates.
2. Antenatal women with singleton viable intrauterine gestation.

EXCLUSION CRITERIA:-

1. Antenatal women with unreliable dates.
2. multiple gestations
3. Antenatal women who will not continue their check up until termination of pregnancy at KGH.

A detailed history of the patient was taken. Risk factors of having fetal abnormality were noted. A detailed systemic and obstetric examination was made . All preliminary investigations were made.

Antenatal women between 11-14 weeks of gestation were offered counseling before the screening. During the counseling, the patients were made aware of the benefits of ultrasound at 11-14 weeks of gestation even if not willing to participate in this study.

Women were counseled about the interpretation of the results of the screening procedure, the possibility for an invasive procedure such as chorionic villous sampling or amniocentesis and also the risks associated with invasive procedure.

After counseling, antenatal women were enrolled after written informed consent in the study and detailed ultrasonography was done. The scans were carried out by the trained sonologist. The following were noted namely fetal CRL, viability, nuchal translucency, any structural abnormalities , uterine anomaly, adnexa, cervix and internal os were noted .Then the estimated chance for having fetal abnormalities based on NT measurements were discussed with the antenatal women and her family. These antenatal women were followed up until termination of pregnancy

CRITERIA FOR DOING INVASIVE TESTING:-

Women with NT> 95thpercentile for that gestational age and CRL were considered to be at high risk (screen positive).

For screen positive women, fetal karyotyping was done after counseling them regarding the risk for having an anomalous fetus. Fetuses with normal karyotyping were followed up with scan to detect anomalies at 18-20 weeks and fetal echocardiography at 20-22weeks.If lethal anomaly detected, counseling and option for termination of pregnancy was given. If no lethal anomaly was identified, pregnancy was continued till term and delivered. After delivery the baby was evaluated for anomalies by paediatrician and appropriate investigations were done.

The following were endpoints to assess the adverse outcome of pregnancy.

1. Pregnancy loss – spontaneous fetal loss.
2. Intrauterine death
3. Termination of pregnancy
4. Fetal aneuploidy (trisomy 21,13,18,turner's syndrome
5. Lethal congenital anomaly

Postnatally identified anomalies in the neonate such as cardiac defects / pulmonary defects/ abdominal defects/ skeletal defects / genetic syndromes.

INTERPRETATION & ACTION:-

1. Normal NT - Routine antenatal care and follow up until delivery.
2. Increased Nuchal translucency -karyotyping(by CVS/ Amniocentesis/ cordocentesis).
3. Karyotyping abnormal – counseling
4. Karyotyping normal – continue pregnancy after anomaly scan.
5. Lethal anomalies detected - counseling
6. No lethal anomalies - fetal 2D echo
7. Fetal 2D echo (N) - continue pregnancy

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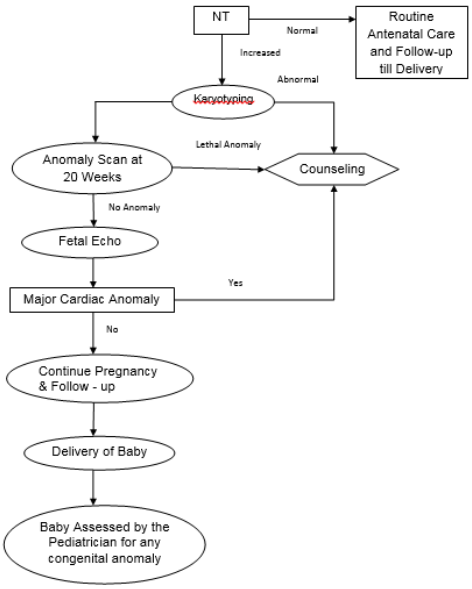
8. Lethal cardiac anomaly - counseling
9. Following delivery - evaluation by paediatrician.



STASTISTICAL METHODS:-

Chi-square test and student t test were used to find the statistical significance in the noted difference of anomalies associated with the fetuses with increased NT using the statistical software namely SPSS version 15.

SAMPLE SIZE CALCULATION:-

A sample size of 100 was arrived by using G power 3.0.10 software which determines the significance size(alpha) at 0.01 and the power of(1-beta) 0.99.

RESULTS AND ANALYSIS:

Table 1

Maternal Age in Years and NT

Maternal Age in Yrs	Normal NT		Increased NT		Pregnancy Outcome for Abnormal NT
	No.	%	No.	%	
<= 20	19	95%	1	5%	Turners Syndrome
21-25	41	100%	0	0%	
26-30	30	93.80%	2	6.30%	1.CHD-ASD 2.CHD-VSD
31-35	2	66.70%	1	33.30%	Spontaneous Fetal Loss, Normal Karyotype, no detectable anomalies
>35	3	75%	1	25%	Downs Syndrome

Mean age for women with normal NT: 24.4 Years (SD-4.3)
Mean age for women with abnormal NT: 28.8 Years (SD-6.5)

Maternal Age and NT

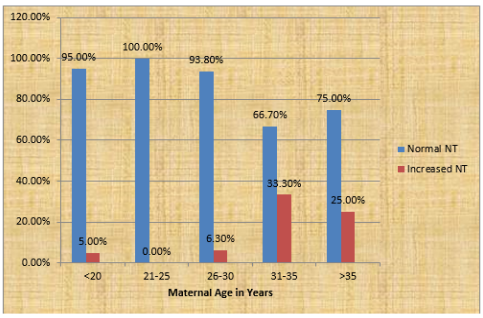


Table 2

Gestational age and NT

Gestational age In Weeks	Number (n-100)		Normal NT (n=95)		Increased NT (n=5)	
	Number	Percentage	Number	Percentage	Number	Percentage
11-11+6	24	24%	22	91.60%	2	8.40%
12-12+6	42	42%	40	95.20%	2	4.80%
13-13+6	34	34%	33	97%	1	3%

Majority (42%) of the women were screened between 12-12+6 Weeks

Gestational Age and NT

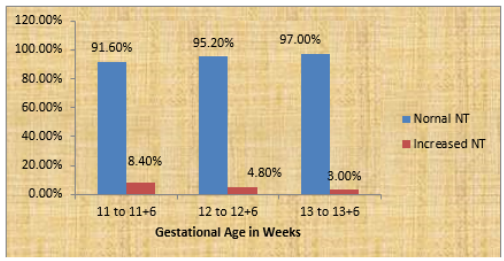


Table 3

Diagnostic value of increased NT in relation to cardiac defects

Diagnostic Value of NT	Number
True Positives	2
True Negatives	95
False Negative	-
False Positive	3
Sensitivity	100%
Specificity	96.90%
PPV (%)	40%
P Value	<0.001**

Statistically significant at < 1% level

Sensitivity of increased NT in identifying isolated cardiac defects is 100%

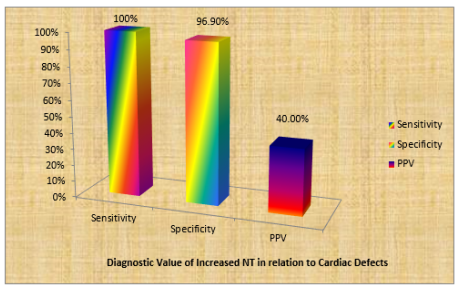


Table 4

Increased NT and Structural Anomaly in Fetus

Nuchal Translucency	No Structural Anomaly		Fetal Structural Anomaly		P Value
	Number	Percentage	Number	Percentage	
Normal	95	100%	0	0%	<0.001**
Increased	1	20%	4	80%	

** - Significant at <1% level

Increased NT was significantly associated with abnormal fetus. The association was statistically significant (p<0.001).

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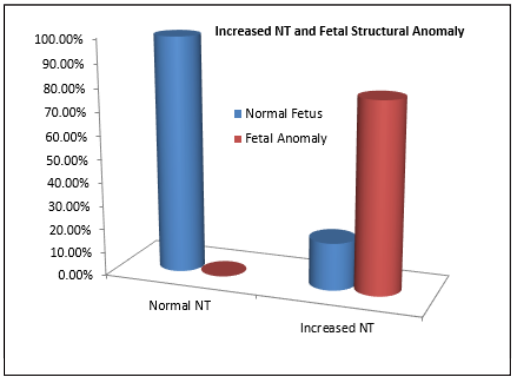
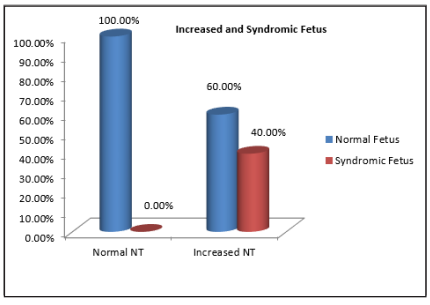


Table 5
Increased NT and Syndromic Fetus

Nuchal Translucency	No syndromic fetus		Syndromic fetus		P Value
	Number	Percentage	Number	Percentage	
Normal	95	100%	0	0%	<0.001**
Increased	3	60%	2	40%	

** - significant at <1% level

Increased NT was significantly associated with syndromic fetus. The association was statistically significant (P value <0.001).



DISCUSSION

One of the major goals of antenatal screening of fetal nuchal translucency at 11-14 weeks of gestation is early identification of the anomalies in the fetus.

In our study, total of 100 antenatal women were screened for fetal nuchal translucency as a marker for aneuploidies and congenital anomalies of the fetus. Majority (42%) of the women were screened between 12-12+6 weeks of gestation. Majority (41%) of the women were between 21-25 years of age. 7% of the women were above 30 years.

In our study, 95% of pregnant women had normal nuchal transluceny (NT < 95th percentile). NT screen positivity rate (NT > 95th percentile) is 5% which is similar in the Literature. A study by Naidoo petal, 2008, reported an incidence of increased NT as 15%.

The mean age for women with normal NT was 24.4 years (S.D 4.3). Mean age for women with abnormal NT was 28.8 years (SD 6.5)

Among women aged less than 30 years the incidence of increased NT 3.2%, whereas among women above 30 years, the incidence is 28.6%. This difference is statistically significant (p value < 0.01) but this could be due to small sample size in the later age group in our study.

With advancing age, the incidence of abnormal nuchal translucency is increased in our study. This is similar to the study by Szabo Jetal, 1995 where increased NT was 5.4% in women >35 years and 1.3% in women <35 years.

Fetuses with normal NT were normal at birth.

Among the 5 pregnant women with increased NT, One (20%) pregnant women had a spontaneous fetal loss at 5 months amenorrhoea. The products of conception revealed a karyotypically normal fetus by cytogenetic analysis. Souka et al, 2001, reported an incidence of 5.15% miscarriages among fetuses with increased NT and normal karyotyping.

All the 5 fetuses with increased NT had abnormal outcome in the form of aneuploidies (40%) (Downs syndrome and Turners syndrome), structural heart disease (40%) (VSD,ASD) and spontaneous fetal loss (20%). Both the pregnancies with fetal aneuploidy underwent medical termination of pregnancy on parental request. Study by Alexioy et al reported an incidence of aneuploidies of 19% among fetuses with increased NT. Study by Bilardo et al revealed 45% adverse pregnancy outcome which included aneuploidies, structural anomalies, miscarriages and termination of pregnancy on parental request.

Among the NT screen positive fetus, 60% had normal karyotyping and 40% had abnormal karyotyping (Downs syndrome and Turners syndrome). This is similar to a study done by Bilardo et al, Amsterdam, 2007. In the latter study which included 675 fetuses with increased NT, 67% & 33% had normal and abnormal karyotyping respectively.

Among fetuses with increased NT, four (80%) had abnormal fetuses (septal defects, aneuploidies) unlike those with normal NT among whom none had abnormal outcome. This difference noted is statistically significant (p Value < 0.001). This finding makes NT measurement in first trimester as an essential screening tool.

NT measurement for detecting isolated cardiac anomaly is 100% sensitive, 97% specific but with a positive predictive value of 40%. Hence increased NT should be considered an indication for fetal Echocardiography. Study by Atzei et al, 2005, reported an incidence of cardiac defects as 18% especially if the NT is more than 99th percentile.

In our study, 40% (2 out of 5) had isolated cardiac anomalies among the increased NT group. Cardiac defects were ASD and VSD.

Bilardo et al study quoted an incidence of 4% isolated cardiac defects among 54 fetuses with increased NT and normal karyotype.

Among 2 live births with increased NT measurement and normal karyotyping and normal anomaly scan, both the babies had cardiac septal defects. Even these cardiac defects will not warrant a MTP even if detected in the anomaly scan. This is in contrary to the study done by Bilardo et al 2007, his study quotes adverse outcome of 4% among fetuses with increased NT, normal karyotyping and normal ultrasound at 18-22 weeks. So mere increase in NT is not an indication to terminate pregnancy.

In our study, distribution between primigravida were 53% and multigravida were 47%. 4 women with increased NT were multi gravida and 1 women with increased NT was a primi gravida.

36% of pregnant women had significant risk factors in the past obstetric history (eg. previous history of fetal anomalies, previous history of Downs syndrome, prior history of recurrent pregnancy loss). Among the above, only one pregnant women (20%) had increased NT and she was also overt diabetic on insulin.

There were no significant correlation of increased NT with present obstetric risk factors throughout the pregnancy except for one pregnant woman with overt diabetes mellitus who was 37 years old and also fetal karyotyping showed downs syndrome.

Majority of the babies had birth weight between 2.51–3.0kgs in our study.

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Two live born babies with antenatally detected increased NT were more than 2.5 Kgs.

SUMMARY

In this prospective clinical study, 100 pregnant women were enrolled between 11 – 14 weeks of gestation after informed consent, over a period of one year who attended the antenatal clinic at the department of Obstetrics and Gynaecology at Govt. Kasturba Gandhi Hospital, Chennai.

Observations in the study includes:

- Total Study population is 100
- Nuchal translucency was assessed in women between 11–14 weeks. 42% were done between 12-12.6 weeks.
- Incidence of increased NT > 95th percentile was 5%.
- All the fetuses with increased NT had adverse pregnancy outcome.
- 2 among 5 (40%) had chromosomal anomalies.
- Isolated cardiac defects were identified among 2(40%) fetuses with increased NT.
- 1(20%) had spontaneous miscarriage.
- Chromosomal anomalies and cardiac defects were commoner association in women with increased NT.
- Increased NT is very sensitive and specific to detect cardiac defects.
- Incidence of increased NT is higher in women more than 30 years (28.6%) against women less than 30 years (3.1%), indicating higher risk for fetal abnormalities in the former group.
- Fetuses with increased NT are at higher risk of developing an anomalous fetus than with fetuses with normal NT and this difference is statistically significant.
- No significant association was noted with increased NT and past obstetric risk factors and the present pregnancy risk.

CONCLUSION OF THE STUDY

- Fetal nuchal translucency is a noninvasive, reliable, screening tool in the first trimester to predict fetal abnormalities including fetal aneuploidies.
- Routine first trimester screening for fetal nuchal translucency should be done in all pregnant women irrespective of the age.
- Increased fetal nuchal translucency is seen in 5% of our study Population.
- Incidence of increased NT is higher among those women more than 30 Years indicating that these women at a higher risk for having anomalous fetus.
- Measurement of increased nuchal translucency provides the women with an early termination option in women with an anomalous fetus.
- Increased nuchal translucency with normal karyotyping does not warrant termination of pregnancy.

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