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Nuchal Translucency, A Marker of Fetal Genetic Health

KEYWORDS: Real Estate Sector,Foreign Direct Investment, Corporatisation,Gross Domestic Product,Government Policy

Dr. Mithil Patil	Dr. Savita Mehendale	Dr. T. M. Panchanadikar
Assistant Professor, Bharati Vidyapeeth University Medical College, Pune.	Professor, Bharati Vidyapeeth University Medical College, Pune	Professor, Bharati Vidyapeeth University Medical College.

Dr. Girija Wagh	Dr. Vandana Nimbargi
Professor & HOD. Bharati Vidyapeeth University Medical College.	Assistant Professor, Bharati Vidyapeeth University Medical College.

ABSTRACT *Nuchal Translucency (NT) is the sonographic appearance of a collection of fluid under the skin behind the fetal neck in the first trimester of pregnancy. In our routine practice anomaly scan is done at the gestational age of 20-21 wks & MTP is allowed only upto the gestational age of 20 wks. So if we found any congenital anomaly or suspicious of it , we can't terminate the pregnancy beyond 20 wks. Patient suffers from tremendous psychological upset knowing her baby is not well inside. So it has become the need of time to diagnose chromosomal & congenital anomalies as early as we can so safely terminate the pregnancies in the first trimester before the patient is `showing`. ACOG 2007 has recommended the same – All pregnant patients in the first trimester should be screened for chromosomal & congenital anomalies of the baby irrespective of the age of the mother. Aim: Screening of chromosomal & congenital anomalies in the fetus with the measurement of nuchal translucency in the first trimester of pregnancy(11 wks – 13.5 wks). Material & Methods : Prospective study is conducted at Bharati Hospital & Research Centre , Pune where almost 176 patients are included in the trial. Nuchal Translucency measurement is done at 11- 13.5 wks of pregnancy on a criteria laid down by the Fetal Medicine Foundation , UK. Observations : Most of the patients (80 %) Nuchal Thickness value falls between 1 to 1.5 mms. While 10 % of the patients have values below 1 mm & 10 % above 2 mm. We have kept 2.5 mms as a cut off between normal & abnormal value .we have come across 5 patients having NT measurement more than 2.5 mms out of which 2 patients were found to have downs fetus' on further diagnostic invasive testing. Conclusion : Nuchal Translucency measurement in the fetus at 11- 13.5 wks of pregnancy is a simple, cheap & less time consuming investigation . If NT measurement is done on a strict criteria laid down by the Fetal Medicine Foundation ,UK , it can certainly give us a first clue regarding the fetal genetic health. So every pregnant women regardless of the age & parity should be screened for chromosomal and structural anomalies by doing Nuchal Translucency Measurement at 11-13.5 wks of pregnancy.*

Introduction :
Nuchal Translucency (NT) is the sonographic appearance of a collection of fluid under the skin behind the fetal neck in the first trimester of pregnancy.

The term translucency is used, irrespective of whether it is septated or not and whether it is confined to the neck or envelopes the whole fetus . The incidence of chromosomal and other abnormalities is related to the size, rather than the appearance of NT . During the second trimester, the translucency usually resolves and, in a few cases, it evolves into either nuchal edema or cystic hygromas with or without generalized hydrops .

Every women is at risk of having a baby with chromosomal & structural anomalies. In our routine practice anomaly scan is done at the gestational age of 20-21 wks & MTP is allowed only upto the gestational age of 20 wks. So if we found any congenital anomaly or suspicious of it , we can't terminate the pregnancy beyond 20 wks. Patient suffers from tremendous psychological upset knowing her baby is not well inside.

So it has become the need of time to diagnose chromosomal & congenital anomalies as early as we can so safely terminate the pregnancies in the first trimester before the patient is `showing`. In most of the western countries, all pregnant patient in 11-13.6 Wks, screened for chromosomal & congenital anomalies. ACOG 2007 has recommended the same – All pregnant patients in the first trimester should be screened for chromosomal & congenital anomalies of the baby irrespective of the age of the mother.

Aim:
Screening of chromosomal & congenital anomalies in the fetus with the measurement of nuchal translucency in the first trimester of pregnancy(11 wks – 13.5 wks)

Material & Methods :
This prospective study was started in June 2011 in which we have screened 176 antenatal patients in their first trimester of pregnancy (age group 18 – 44 years).When the patients came with missed date & pregnancy is confirmed then every one of them is counseled regarding the need of NT Measurement . NT Measurement is done in the 11-13.5 weeks of pregnancy by the criteria laid down by Fetal Medicine Foundation,UK

Nuchal translucency is measured by the FMF,UK certified sonologist on the guidelines as mentioned below

1. CRL should be 45 – 84 mms.
2. Mid-sagital plane should be obtained with the fetus in a neutral position, and the magnification was such that the fetus occupied three-quarters of the image and fetal skin and amnion were distinguished.
3. Only head & thorax should be visualized.
4. There should not be over flexion or extension of the head.
5. Calipers should be placed on the inside of skin margins & maximum width of the Nuchal Translucency should be measured.

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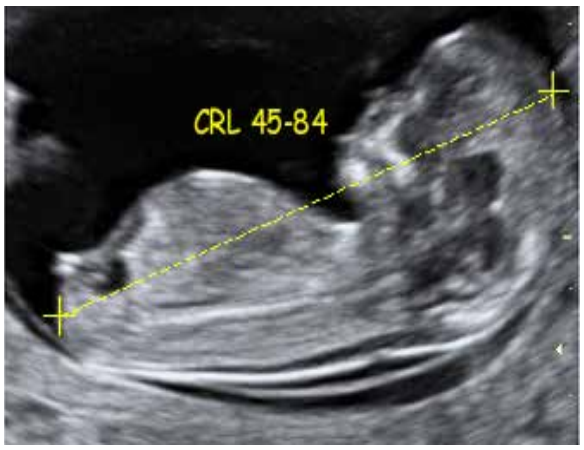
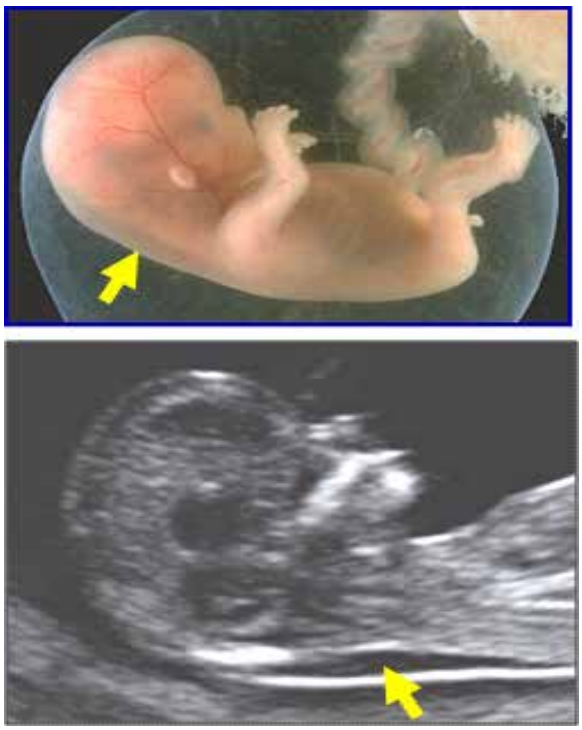


Ultrasound Image of a 12 weeks fetus in sagital view clearly depicting Nuchal Translucency.

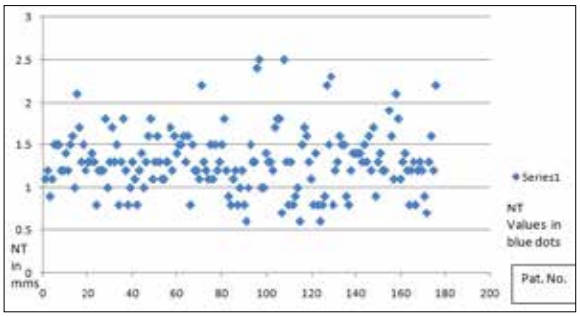


Nuchal Translucency as seen in a 12 weeks fetus.

Observations:
NT measurement is done at 11 – 13.5 wks of pregnancy on a strict guidelines implemented by Fetal Medicine Foundation , UK. Almost 176 patients are screened till date and we have found only 9 patients having NT measurement value more than 2 mms. Most of the patients Nuchal Translucency measurement lies between 1 to 1.5 mms . In our study we could diagnose 2 patients with Downs syndrome , having NT measurement 2.5 mm & 2.6 mms respectively.

Though alone NT measurement is having the sensitivity of 69 % in screening for the chromosomal anomalies, it can certainly point us regarding the high risk pregnancies . Those patients can be further evaluated by more specific & sensitive tests such as First Trimester Screening OR Contingent screening.

In our study we have screened all pregnancies with NT Measurement as a initial screening test and those patients found to have NT > 2.5 mms were subjected to further more specific screening test. In the study 2 patients were having raised NT > 2.5 mms , were screened by FTS , found to have risk ratios > 1/250 .On doing invasive diagnostic test(Amniocentesis) both these patients were found to have triosomy 21 (Downs Syndrome) in the fetus.



(Nuchal Translucency measurement values in millimeter as blue dots)

Discussion :
Screening for chromosomal abnormalities in an obstetric setting has traditionally meant screening for trisomy 211. It is the single most common cause of mental retardation of school-age children. The majority of chromosome abnormalities identified in prenatal samples are trisomy for chromosomes 13, 18, 21 and sex chromosome aneuploidies. These are associated with the newborn phenotypes, Patau, Edwards and Down syndromes (trisomy 13, 18 and 21 respectively), and the less severe Turner (monosomy X) and Klinefelter (XXY) syndromes. Down's syndrome, with an incidence rate of 1 in 800 pregnancies, is the predominant reason for women seeking prenatal diagnosis2, 3.

The distinction between screening and diagnostic tests is often spurious since most tests will be used in both ways in different patients. At present accurate prenatal diagnosis of chromosomal anomalies is only available by obtaining fetal cells through an invasive procedure, such as amniocentesis or chorionic villous sampling. Karyotype analysis of cells by culture is usually available in more than in one week. In order to reduce anxiety and improve pregnancy management, more rapid aneuploidy testing are used. The most widely established method is interphase-fluorescence in situ hybridization (FISH)4,5, in which a set of chromosome- specific fluorescence-labelled probes are hybridized to interphase nuclei of uncultured prenatal cells. The number of fluorescent signals in each nucleus represents chromosome copy number. Between 50 and 100 cells are usually analyzed to allow for low-level background and signal overlay that can occur during FISH procedures6. A quantitative fluorescence-PCR (QF-PCR) is a more recent addition to aneuploidy diagnosis7,8. The technique involves the relative quantification of microsatellite alleles to determine sequence copy number; amplification using fluorescence-labelled primers is followed by size separation and allele peak measurement on a semi-automated genetic analyzer.

Due to the fact that both amniocentesis and CVS are associated with a risk of miscarriage9 these procedures are currently applied only to small group of women who are in a higher risk of having an offspring with a chromosomal defect in comparison to the general population. The aim of the currently available screening tests is actually to identify, with the highest possible sensitivity and specificity, those women who should be offered the invasive procedure. The risk for many of the chromosomal defects increases with maternal age. Additionally, because fetuses with chromosomal defects are more likely to die in utero than normal fetuses, the risk decreases with gestational age. Maternal age of 35 years at delivery has been the medicoligal standard in USA and Europe for more than 20 years (Table 1). This high risk group constituted 5% of the pregnant population10. It was estimated that approximately 30% of trisomy 21 occurred to mothers >35 years old. It is a fact that although the risk of any individual 36 years old is higher than in a 26 years old woman for example, there are so many more pregnancies in the 26 years old group that from a population perspective, most abnormalities (approximately 70%) occur in the 'low risk' population11.

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Nowadays, such screening for prenatal diagnosis is provided by using the family history, the maternal serum screening and the ultrasonography. Every time a test is carried out the background risk is multiplied by the test factor to calculate a new risk, which then becomes the backround risk for the next test 12. This process is called sequential screening. Although screening tests are not diagnostic they can indeed alter the odds 13.

Conclusion :

Nuchal Translucency measurement in the fetus at 11- 13.5 wks of pregnancy is a simple, cheap & less time consuming investigation . If NT measurement is done on a strict criteria laid down by the Fetal Medicine Foundation ,UK , it can certainly give us a first clue regarding the fetal genetic health. So every pregnant women regardless of the age & parity should be screened for chromosomal and structural anomalies by doing Nuchal Translucency Measurement at 11-13.5 wks of pregnancy.

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