Ultrasonographic screening of major structural anomalies during the NT examinations in infertile patients

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Purpose

Nuchal translucency (NT) measurement in the first trimester screening have been recognized as a clear marker for chromosomal and non chromosomal abnormalities, but a true association between Increased NT and congenital abnormalities has only been identified in a few cases (1,2).

NT screening using sagital views can also screen for fetal structural anomalies for patients that need an early fetal anatomy scan (3). First trimester screening can lead to early detection and appropriate management which can improve outcome (4). Management includes karyotyping and detailed ultrasound performed between 18 and 22 weeks of gestation paying particular attention to the heart (5). If required, parents should be offered genetic counseling and a full neonatal examination by a pediatrician.

The need for prenatal screening is extra important in the case of assisted conception due to more concern over specific anomalies, multiplicity and poor outcome pregnancies. Therefore, every effort should be made to provide pregnant women particularly ART mothers with the most accurate screening tests. The purpose of this study was to evaluate the ability to screen for major structural fetal anomalies during the NT examinations in infertile patients.
Methods and Materials

In a prospective study during one year period 876 NT examinations were performed for 657 women (mean maternal age 30 years, range 19-50) who were conceived after Assisted Conception irrespective of its multiplicity at the Royan Institute between 11 to 14 weeks' gestation.

This study approved by research ethics committee and institutional review board in Royan Institute and informed written consent is obtained from the patients. Transvaginal and transabdominal scans were conducted in the first trimesters in order to detect any fetal anomalies during NT examinations. The sonographic findings were compared with pediatric examinations after birth or pathology reports in the case of terminated pregnancies. All the infants were examined by a pediatrician and followed for the first two weeks after birth for any anomalies. Patients with suspected anomalies were followed throughout the pregnancy and all cases with an increased NT (>95th percentile) referred to amniocentesis and karyotyping, genetic counseling and fetal echocardiography.

All the ultrasounds were performed by one experienced radiologist with ten years of experience in abdominal sonography and 7 years experience in transvaginal sonography and the ultrasound records were evaluated by independent observer experienced in obstetric ultrasonography.

Each patient was initially examined transabdominally (using Aloka # 10-probe 3.5-5 MHz-Japan), before undergoing transvaginal ultrasounds (Aloka # 10-probe 6-7.5MHz-Japan) during further analysis.

Throughout this study, NT was only measured when the fetus was in a neutral position, with the head in line with the spine. The ultrasound machine had a zoom option and was set so that only the fetus's head and thorax could be seen in the ultrasound image. At that point the thickest translucency behind the fetus's neck was measured according to the Fetal Medicine Foundation (FMF) guidelines. NT was measured as the maximal thickness (inner to inner) of sonolucent zone. During the scan more than one measurement was taken and the maximum one that meets the criteria was recorded.

In addition to NT measurement, in this study transabdominal scans were used to determine the gestational age of a pregnancy, evaluate fetal positioning; examine fetal heart beat and morphology and nasal bone (NB) and other ultrasound "soft markers" used in fetal genetic screening. However, more detailed anatomical features of the fetus including the fetal head, spinal cord, stomach, kidneys and bladder were thoroughly examined using transvaginal sonography. Any cardiac defect suspected on routine prenatal sonography referred to a detailed routine fetal echocardiography.

Statistics:
Statistical analysis was done by SPSS software. P<0.05 was considered as statistically significant. McNemar's test was used for data analysis.
Results

In the present study 10 fetuses (1.09%) with structural anomalies diagnosed including: hydrops fetalis, Cystic Hygroma (Fig1), Jejunal obstruction (Fig2), hydronephrose, hydrocephaly, trisomy 18, 2 cases of multiple anomalies and 2 cases of extremity anomalies (Fig 3). There were no cases of missed diagnosis of structural anomalies.

Anomalies were detected in only 6 out of 856 fetuses (0.70%) with normal NT, whereas from 20 fetuses with an abnormal NT (>95th percentile), 16 fetuses were available for follow-up, 3/16 (18.75%) diagnosed with at least one major congenital anomaly (2 extremity anomalies, 1 multiple anomalies) resulted in elective termination of Pregnancy, 2/20 neonatal death due to preterm delivery with normal karyotype, 2/20 intrauterine fetal demise (IUFD) and 4/20 reduction due to multiplicity were recorded.

According to McNemar’s analysis, a significant relationship exists between the NT and detected anomalies. However in this study no significant relationship could be derived between NT and chromosomal anomalies (P-value >0.99) probably due to small sample size.
**Fig. 1**: Ultrasound scan indicated an increased NT measurement of 7 mm. Note multiple thin septa (cystic hygroma).

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Fig. 2: Multiple dilated proximal bowel loops (15mm) due to small bowel atresia observed at 33 weeks gestation.

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Fig. 3: A 28-year-old, client is admitted at 12.5 weeks gestation with twin pregnancy. NT measured at (5mm) in one fetus. The diagnosis of Lower-extremity deformities (Bilateral club foot) was confirmed postnatally.

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Fig. 4: A 28-year-old, client is admitted at 12.5 weeks gestation with twin pregnancy. NT measured at (5mm) in one fetus. The diagnosis of Lower-extremity deformities (Bilateral club foot) was confirmed postnatally.

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Conclusion

The benefits of prenatal diagnosis of congenital anomalies particularly in fetal conceived by assisted reproductive technology (ART) methods are widely known. The need for more accurate and noninvasive prenatal screening such as NT measurement is extra important in the case of assisted conception due to more concern over associated risks of invasive test such as amniocentesis and chorion villus sampling (CVS). Furthermore the some noninvasive test such as triple test is not the effective screening test in multiple pregnancies that is common between ART mothers.

In the mid 1960s, amniocentesis was offered as pregnancy screening for fetal aneuploidy. Maternal age was used as the criterion for screening. New approaches in maternal serum and ultrasound screening has made it possible to assess the risk of having a fetus with chromosomal and non chromosomal anomalies even if conventional karyotyping is normal. Several studies have been published, since the first report on the ability of nuchal translucency measurement to detect pregnancies affected by Down's syndrome by Nicolaides et al in 1941 (6). In the last 10 years, several studies have been declared that the positive predictive value of the NT measurement is high enough in both high-risk and low-risk groups, that patients with increased NT should be offered a detailed fetal evaluation (genetic sonography) at the end of the first trimester and also at 18-22 weeks of gestation. Prenatal diagnostic testing such as triple test, amniocentesis, fetal echocardiography and follow-up after birth is necessary. Measurement of the NT thickness combined with biochemical markers has a false-positive rate of 5% (7) and it is regarded as a basic screening test with high sensitivity for identifying fetuses at risk for aneuploidy (2,8,9). However, NT is increased in 4.4% of euploid fetuses who are at risk for fetal anomalies and an adverse pregnancy outcome (9).

The risk of perinatal outcome increases in an exponential fashion as the NT measurement increases (10). The chance of delivering a healthy baby decreases with NT thickness from approximately 70% for an NT of 3.5-4.4 mm to about 15% for an NT of 6.5 mm or more (10).

Using NT screening to identify fetuses at risk for congenital anomalies at 11 to 14 weeks' gestation offer an option of early termination which is safer, and less traumatic in the expectant mother.

In this study there is only one case of anaploidy (trisomy 18) with increased NT and in contrast to previous studies no significant relationship could be derived between NT and chromosomal anomalies (P-value >0.99) probably due to small sample size.

According to the results of this study a significant relationship was found between a high NT and occurrence of malformations.
References


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