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Outcome of fetuses with NT above the 95th percentile in an resource limited population to enable effective counselling of couples

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Objective

To assess the presence of fetal aneuploidies, structural defects, fetal loss and delivery of a normal live baby in fetuses with NT above the 95th percentile at the 11^{+0} – 13^{+6} weeks' scan.

Methods

This is a retrospective study of prospectively collected data from a single tertiary fetal care centre in South India from August 2009 to December 2019. NT was measured at a gestational age of 11^{+0} – 13^{+6} weeks with CRL of 45mm - 84mm as per FMF guidelines. All data is stored on the FMF certified Astraia fetal database software. Fetuses with NT more than the 95th percentile were identified and analysed for the presence of aneuploidies, structural defects, intrauterine fetal demise or miscarriage and delivery of a normal alive baby.

Results

8,609 singleton pregnancies with known outcomes had 93 (1.1%) fetuses with aneuploidies, of which 83 (1%) were clinically significant. 652/ 8609 (7.5%) fetuses had NT > 95th centile. 397/652 (60.8%) fetuses with known karyotype were identified and divided into 5 groups of increasing NT measurements. 82/397 (20.6%), 208/397 (52.3%), 38/397 (9.5%), 21/397(5.2%), 48/397(12%) fetuses had NT >95th- 99th centile, 3-3.9mm, 4-4.9mm, 5 -5.9mm and >/= 6mm respectively. 7/82(8.5%), 20/208(9.6%), 12/38(31.5%), 7/21(33%), 26/48(54.1%), fetuses with NT >95th- 99th centile, 3-3.9mm, 4-4.9mm,5 -5.9mm and >= 6mm respectively had an abnormal karyotype. 75/82(91.4%), 188/208(90.3%), 26/38(68.4%), 14/21(66.6%), 22/48(45.8%) fetuses had normal karyotype in above mentioned groups respectively. There were 325/397(81.8%) euploid fetuses with increased NT. Out of 325 fetuses, 49/325(15%) had structural defects. 2/75(2.6%), 16/188(8.5%), 5/26(19.2%), 9/14(64.2%), 16/22(72%) structural defects seen in euploid fetuses with NT >95th- 99th centile, 3-3.9mm, 4-4.9mm,5 -5.9mm and >= 6mm respectively. 276/325(84.9%) fetuses had no structural defect, out of which 4/276(1.4%) structural defects were detected postnatal. (1 cardiac, 1 Noonan syndrome (pulmonary stenosis), 1 undescended testis, 1 Ambiguous genitalia) Among the euploid fetuses with no associated structural defects, 71/72(98.6%), 169/172(98.2%), 20/21(95.2%), 4/5(80%),5/6(83.3%) fetuses had live birth in the following groups respectively- >95th- 99th centile, 3-3.9mm, 4-4.9mm,5 -5.9mm and >= 6mm.

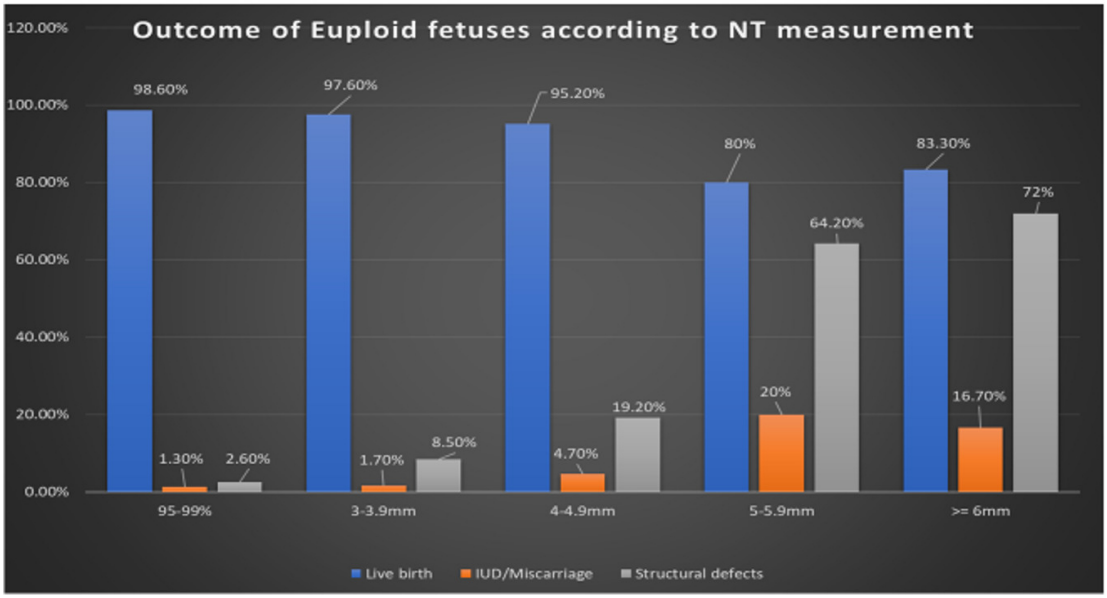
Conclusion

Increased NT defined as NT above the 95th centile for CRL is known to be associated with chromosomal anomalies, structural defects, especially cardiac and skeletal defects, and intrauterine fetal demise. The higher the NT, the higher is the risk of an adverse outcome. However, majority of the babies with increased NT will be normal. The pregnancy must go through a series of systematic examinations to exclude the anomalies associated with increased NT to prove that the fetus is normal. In a country like India where the healthcare services are rather skewed towards the rich and affording, many pregnancies are terminated without investigations in a "fear of abnormality" rather than proving it to be abnormal owing to the increased cost of prenatal investigations. In our study nearly 40% of fetuses had no prenatal tests and hence would have contributed to many normal fetuses being terminated. We believe our study is important in this set up as even with limited number of fetuses with increased NT (=397) that have been investigated, 18.1% had a chromosomal anomaly, 15% had structural defects, 2.2% intrauterine demise and 97.5% had a healthy live birth. This translates to 1: 5 with a chromosomal anomaly and 1: 6 with a structural defect. This information would be helpful in prompting the parents to investigate the pregnancy after the detection of increased NT rather than terminate without any checks. The strength of our study is the good sample size and correlation of NT with adverse fetal outcomes in a resource limited setting. The limitation of our study is the high rate of terminations in pregnancies with abnormal findings without investigations.

ID:4210 **Outcome in terms of associated chromosomal abnormalities , structural defects in fetuses with increased Nuchal translucency (NT) according to the assigned groups**

Total cohort- 8609 Fetuses with NT>95th centile- 652/8609(7.5%) NT>95th centile with known KT n-397/652- (60.8%)	n	Chromosomal abnormalities	Normal Karyotype	Structural defects in Euploid fetuses	No defects in Euploid fetuses
95-99th centile	82/397 (20.6%)	7/82 (8.5%)	75/82 (91.4%)	2/75 (2.6%)	73/75 (97.3%)
3 – 3.9mm	208/397 (52.3%)	20/208 (9.6%)	188/208 (90.3%)	16/188 (8.5%)	172/188 (91.4%)
4 - 4.9mm	38/397 (9.5%)	12/38 (31.5%)	26/38 (68.4%)	5/26 (19.2%)	21/26 (80.7%)
5 - 5.9mm	21/397 (5.2%)	7/21 (33%)	14/21 (66.6%)	9/14 (64.2%)	5/14 (35.7%)
≥6 mm	48/397 (12%)	26/48 (54.1%)	22/48 (45.8%)	16/2 2(72%)	6/22 (27.2%)

ID:4210 Fig 1 Pregnancy outcome in euploid fetuses with increased NT



(ID:4210) Images showing increased Nuchal Translucency(NT)