Prospective and retrospective study of congenital anomalies in a developing country: screening and diagnostic capabilities of first trimester NT evaluation

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Objective

To evaluate the ultrasound measurment of nuchal translucency (NT) at 11-13 weeks gestation and to correlate increase NT with a wide range of fetal defects, genetic syndromes and an overall increase in morbidity and mortality.

Methods

The study was carried out in pregnant women with gestational age of 11 weeks and 13 weeks 6 days at our institute hospital for a period of 3 years. Pregnant women undergone screening will be followed up until the pregnancy outcome and clinical assessment of newborn for congenital anomalies were complete. Exclusion criteria: 1) Pregnant women with gestational age less than 11 weeks or more than and equal to 14 weeks. 2) Crown-rump length less than 45mm or more than 84 mm. The data are entered in study proforma and statistical analysis will be done by using Chi-Square test. P<0. 05 is considered to be significant.

Results

1) Increased NT is 551. 41 times likely to have anomalies compared to normal NT. 2) Significant association of increased NT with Down's syndrome (15. 72%). 3) Increased NT thickness is associated with following distribution of congenital anomalies: Genito-urinary tract (30%), Facial Anomalies (15. 7%), Central Nervous System (14. 2%), Muskulo-skeletal system (14. 2%), Cardiovascular system (11. 4%), Respiratory system (10%) and Gastro-intestinal Tract (4%). 4) Multiple congenital defects seen in 84. 28% and single congenital defect seen in 15. 72% in association with thickened NT.

Conclusion

1) Nuchal translucency is a strong predictor of congenital anomalies and Downs syndrome. 2) First trimester scanning for NT helps in determining likelihood of fetal anomalies.

Distribution of normal and high NT among study group, (n=1,122).

NT	No anomaly	Congenital anomaly	Total
Normal	935 (83.33%)	1 (0.08%)	936 (83.4%)
High	117 (10.4%)	69 (6.2%)	186 (16.6%)
Total	1052 (93.8%)	70 (6.2%)	1122 (100%)

Proportion and distribution of congenital anomalies of the babies born to the study group (n=1.122).

Congenital Anomalies	Number (%)
Cardiovascular system	8 (0.7%)
Central Nervous System	11 (1.0%)
Gastro-intestinal Tract	3 (0.2%)
Genito-urinary tract	21 (1.8%)
Muskulo-skeletal system	10 (0.8%)
Facial Anomalies	11 (1.0%)
Respiratory system	7 (0.6%)
Trisomies	11 (1.0%)

Increased risk of congenital anomalies with high NT value (n=1122).

Congenital Anomalies	With low NT	With high NT value	Total
Cardiovascular system	0	8 (0.7%)	8 (0.7%)
Central Nervous System	1 (0.09%)	10(0.89%)	11 (0.98%)
Gastro-intestinal Tract	0	3 (0.26%)	3 (0.26%)
Genito-urinary tract	0	21 (1.87%)	21 (1.87%)
Muskulo-skeletal system	0	10(0.89%)	10(0.89%)
Facial Anomalies	0	11 (0.98%)	11 (0.98%)
Respiratory system	0	7 (0.6%)	7 (0.6%)
Trisomies	0	11 (0.98%)	11 (0.98%)