

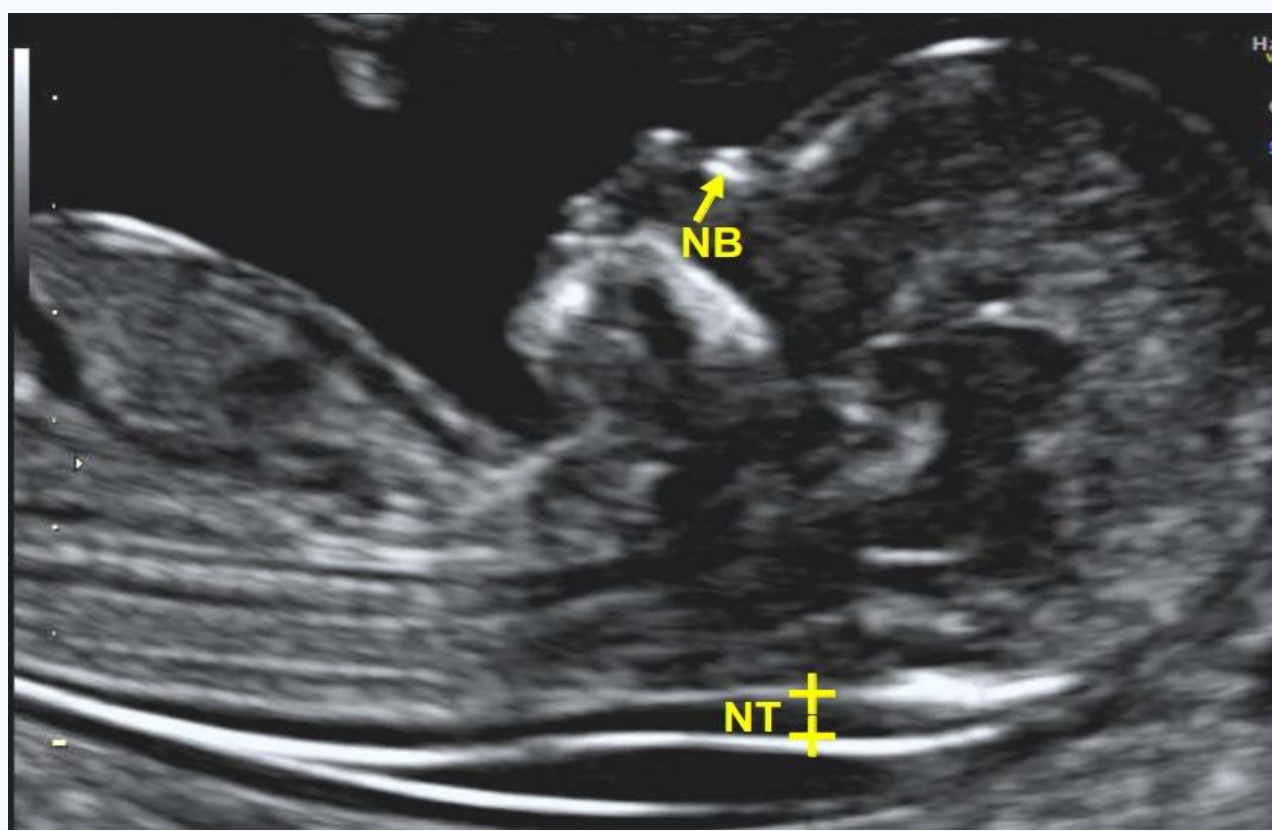
OUTCOME OF FETUSES WITH NUCHAL TRANSLUCENCY ≥ 99TH PERCENTILE - A PROSPECTIVE OBSERVATIONAL STUDY

Amala Jose, Vidyalekshmy R, Brinda Sabu, Rafeekha P

Department Of Obstetrics and Gynaecology & Maternal and Fetal Medicine KIMSHEALTH , Trivandrum, Kerala, India

INTRODUCTION

- Nuchal translucency (NT) is an important ultrasound tool for screening of fetuses at 11-13+6 weeks gestation, for aneuploidies, structural anomalies and other genetic defects.
- Popular belief is that increased NT invariably means bad outcome
- Studies conducted in fetuses with raised NT but a normal karyotype shows that the outcome is often favourable
- A cut-off of 99th centile for a particular CRL to define increased NT gives better sensitivity and lower false positive rate
- Study to obtain data on the outcome of fetuses with raised NT in Indian population, which will guide further evaluation and counselling of the parents.



METHODOLOGY

- Prospective observational study
- Tertiary health care centre in South India
- Study period -2 years
- Sixty singleton pregnancies were included in the study (2.7%, 60/2210) out off the 2210 first trimester scans
- Two were lost to follow up; remaining 58 pregnancies were followed up
- To find out the proportion of aneuploidy, and other adverse perinatal outcomes among the foetuses with increased NT
- Favourable outcome : live birth of a baby with no structural abnormalities and no obvious genetic markers reported after clinical examination by a neonatologist, immediately after delivery

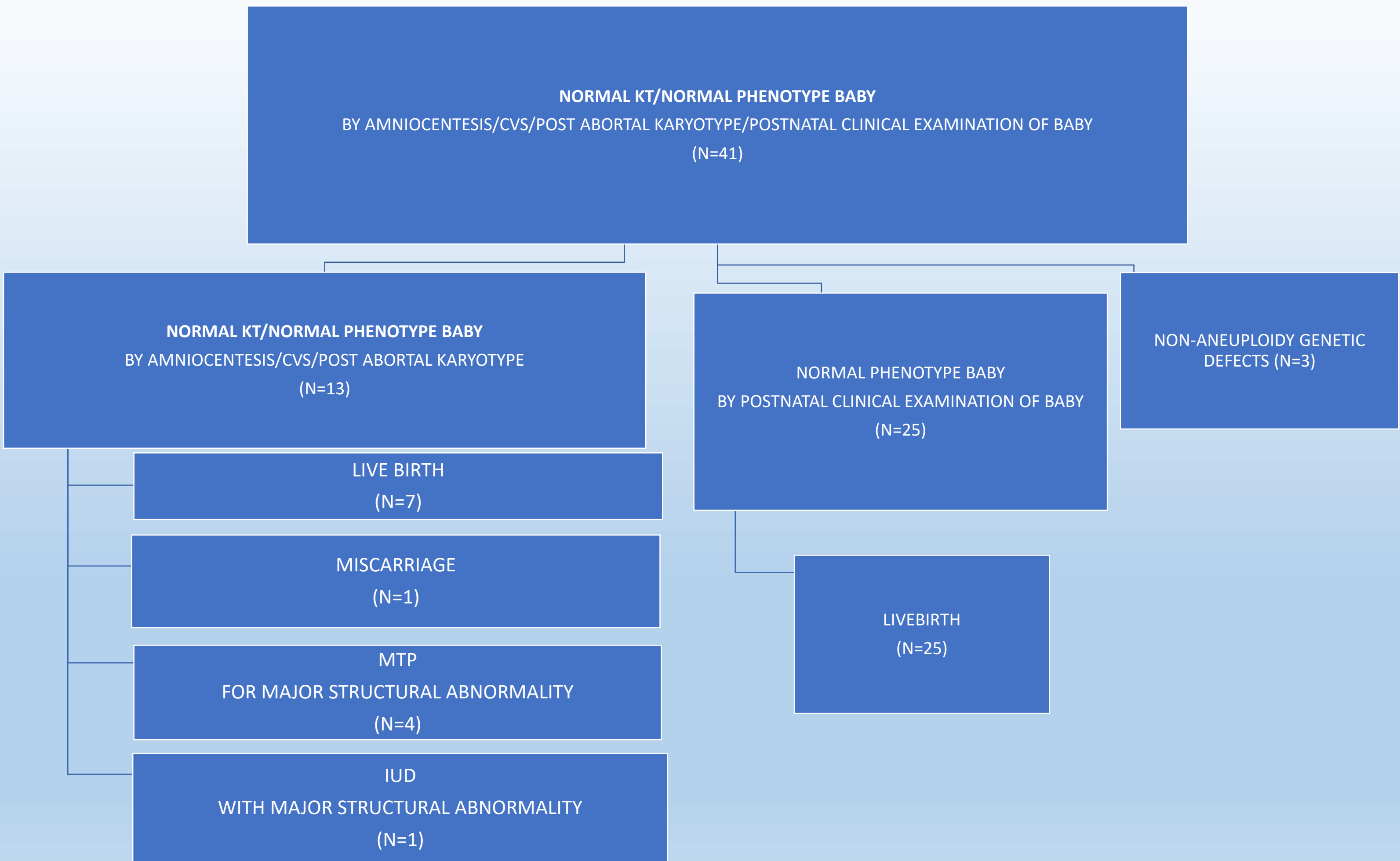
RESULTS

- The mean maternal age was 28.7 ± 4.6 years
- The mean CRL was 65.27 mm.
- The mean nuchal translucency thickness was 3±0.7 mm.
- The nuchal translucency thickness of fetuses with NT ≥ 99th centile in our study : 2.10 mm at a CRL of 54.4 mm to 4.6mm at a CRL of 57 mm

- The incidence of aneuploidy in our study was 12.1% (7/58)
- The incidence of normal karyotype was 70.7% (41/58)
- The percentage of live birth among them was 80.5% (33/41)- including one case of FGR.
- Major structural abnormality were found in 17% (7/41)
- Non aneuploidy genetic defects were found in 7.3% (3/41) of cases with normal karyotype

Type of defect	Count	Structural abnormality
Cardiac defect	1	Congenitally corrected
	1	TGA
		Single outflow tract
Cardiac defect + skeletal defect	1	Complex cardiac defect , caudal agenesis
Skeletal defect	1	Bilateral CTEV
Genitourinary	1	Megacystis
Body stalk anomaly	1	Abdominal wall defect
Encephalocele + skeletal defect + facial defect	1	Occipital encephalocele, club hand, facial dysmorphism

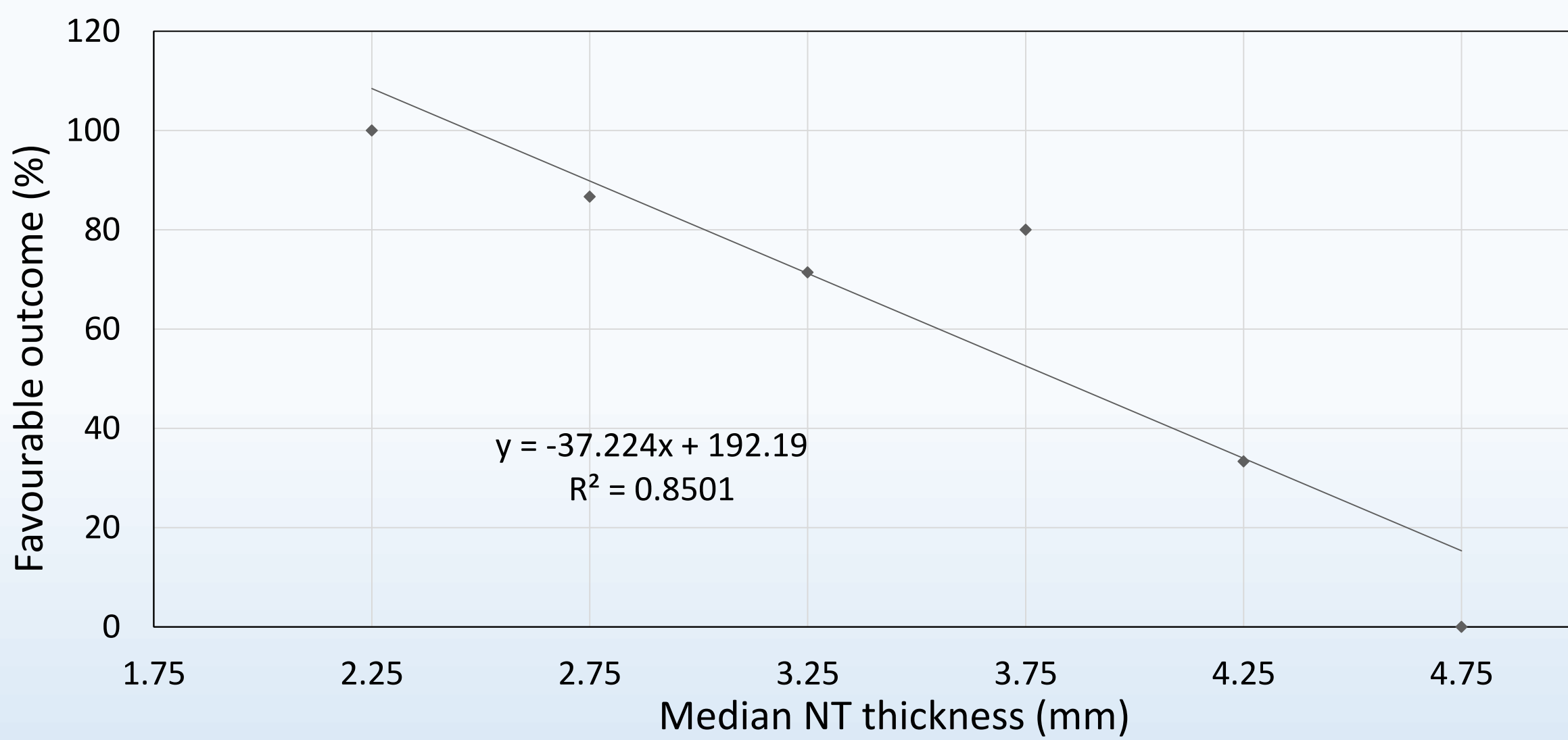
Table Percentage distribution of the sample according to outcome				
Outcome		Count		Percent
Favourable outcome		32		55.1
	Aneuploidy	7		
	Major anomaly	12		
	Other genetic defects	2		
	MTP Others	2		
		23		39.7
Miscarriage		1		1.7
IUD		1		1.7
FGR		1		1.7



- The presence of unossified nasal bone (p<0.01), markers of aneuploidy (p=0.037), or any other structural abnormality noted in the anomaly scan at 16 weeks (p=0.001) was associated with adverse outcome
- A low risk report in non-invasive prenatal testing was associated with favourable outcome (P=0.036)
- A positive association was found between incidence of aneuploidy and UNB (p=0.007)
- Majority of the cases with favourable outcome, delivered near term (75.6%; 31/41)
- The mean birth weight of the fetuses were -3.1 +/- 0.3 kg

DISCUSSION AND CONCLUSION

- Our study concluded that Increased NT resulted in adverse outcome like aneuploidy, structural abnormalities, non-aneuploidy genetic defects, miscarriage, FGR and IUFD.
- A favourable outcome was obtained in, 78% of fetuses with normal karyotype and 97% among those with, a normal karyotype and without structural abnormalities
- 62 % of the fetuses with NT ≥ 99th centile, with an initial NT value < 4 mm resulted in live birth of a normal baby
- We observed that, the chance of a favourable outcome depends on the degree of the initial nuchal translucency thickness
- A favourable outcome was found to depend on the initial nuchal translucency thickness, a normal genetic test and normal morphology on ultrasonography follow-up



The relationship between initial NT thickness and favourable outcome

- On detection of NT ≥ 99th centile in a fetus, it is imperative to refer the couple to a fetal medicine specialist for a detailed first-trimester ultrasound to look for anomalies and markers, offer genetic testing and follow-up the pregnancies closely.

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