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Evaluation of nuchal translucency as a sonographic marker for screening congenital heart defects in high risk cases

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Abstract
Introduction: Congenital heart defects (CHDs) are commonest structural birth defects and leading cause of infant morbidity & mortality. Present study aims to evaluate nuchal translucency (NT) as a sonographic marker for screening CHDs.
Methodology: 142 high risk mothers for CHDs, who attended antenatal clinic of hospital between July 2014 to June 2016 underwent NT scan between 10-14 weeks.
Results: Heart defects were diagnosed in six newborns (2 hypoplastic left heart syndrome, 1 tricuspid atresia, 2 ventricular septal defect, 1 atrial septal defect). For major cardiac anomalies sensitivity of NT scan was 66.7% & specificity 88.4% at 95th centile cut-off (LR+5.75 & LR-0.376), and at 99th centile cut off sensitivity remained same but it was 99.2% specific (LR +83.3, LR- 0.335). NT scan had poor screening potential for right heart lesions.
Conclusion: Increased nuchal translucency thickness was associated with higher risk of major congenital heart defects in chromosomally normal pregnancies.

Keywords: Congenital heart disease, nuchal translucency, echocardiography, karyotyping

Introduction
Congenital heart defects (CHDs) are structural birth defects regarded as important cause of infant morbidity and mortality. World-wide incidence varies between 4/1000 to 50/1000 live births [1]. While in developing countries, estimated prevalence is around 8 per 1000 live births [2]. Due to very high birth rate, burden of CHD is very high in India, with over 180,000 children born with CHD every year [3]. Thus arising the need of early screening and detection of CHDs. Nuchal translucency signifies the transient subcutaneous fluid collection behind the fetal neck as seen on ultrasonography at 10-14 weeks gestation. Increased nuchal translucency is associated with increased risk of fetal anomalies in karyotypically normal foetuses [4]. Cardiac failure secondary to CHD also leads to fluid accumulation causing the nuchal edema. Thus, emphasising on the fact that NT measurement at this gestation can be used as a screening tool for CHDs. The aim of this study was to evaluate the measurement of NT as a sonographic marker between 10 to 14 weeks gestation for screening CHDs in high risk cases.

Methods
The present study was a prospective observational study performed in High risk group for fetal CHD. All antenatal women attending the antenatal clinic of Department of Obstetrics & Gynaecology of hospital with any maternal indication (risk) of CHD [i.e. Maternal metabolic disorder, Bad obstetric history, Maternal/fetal sibling with CHD, Collagen disorder, first trimester teratogen exposure, TORCH infection, IVF conceived pregnancy] were recruited for the study from July 2014 to June 2016. Written informed consent was obtained from all the participants prior to enrolment.
Nuchal translucency was measured between 10 to 14 weeks gestation in a sagittal plane as the maximal son lucent zone between the inner aspect of the fetal skin and the outer aspect of the cervical spine. Nuchal translucency thickness normally increases with crown-rump length, and the 95th centile is 2.2 mm for a crown-rump length of 38 mm and 2.8 mm for a crown-rump length of 84 mm; the 99th centile does not change significantly with crown-rump length and is about 3.5 mm.

Echocardiogram was performed in all cases after delivery. Two dimensional echocardiographic identification of cardiac chambers was performed initially to look for structural cardiac anomalies. The views seen were 4 chambered view, 3 vessel view, 3 vessel tracheal view, outflow tract view, short axis view, ductal and aortic arches. Heart defects were divided in major (i.e. requiring immediate intervention) and minor categories. Karyotyping was done in all new-borns to exclude aneuploidy.
The data were collected from 142 high risk cases for CHD, and collected data was statistically analysed by using IBM SPSS 25.0 version to draw valid conclusions.

Results

Among 142 high risk cases recruited previous issue with any congenital anomaly was the major risk factor (38%). In 19% cases mother had congenital cardiac disease, 14.2% mothers were diabetic (pre-gestational), 18% mothers had TORCH infection, 7.6% were IVF conceived pregnancies. Majority of high risk cases were between 25-29 years age group (45.1%), and the mean gestation age at the time of screening was 12.4 weeks. 15 foetuses lost to follow up while 3 new-borns were found aneuploidy on karyotyping. Thus, our study group included 124 cases.
Major CHDs are the ones posing risk to infants life and requiring surgery during the first year of life [5, 6]. Major heart defects were identified in 3 cases in postnatal echocardiogram (2 hypoplastic left heart syndrome, 1 tricuspid atresia) and 3 newborns were diagnosed with minor septal defects (2 ventricular septal defect, 1 atrial septal defect). Left heart lesions (i.e. Hypoplastic left heart syndrome) and septal defects were identified by increased NT, but right heart lesion (i.e. tricuspid atresia) was not identified as shown in table 1

Table 1: Detection rate of specific heart defects using increased nuchal thickness at different cut-offs

Cardiac defect	No. (%) with NT>= 95th centile	No. (%) with NT>= 99th centile
Left heart lesion	2 (100)	2(100)
Right heart lesion	0 (0.00)	0 (0.00)
Septal Defects	1 (33.3)	0 (0.00)

At 95th centile cut-off correlation of NT thickness with CHDs was statistically significant but was statistically highly significant on using 99th centile as cut off, as shown in table no. 2. Mean NT thickness was higher in foetuses with major CHD (i.e. 3.6mm) than those with normal heart (i.e. 1.7mm).

Table 2: Correlation of nuchal translucency with cardiac anomaly (n=124)

Nuchal translucency	Cardiac anomaly		Major CHD	
	Not detected (%)	Present (%)	Not detected (%)	Present (%)
≤ 95th centile> 95th centile	105 (89.0) 13 (11.0)	3 (50) 3 (50)	107 (88.4) 14 (11.6)	1 (33.3) 2 (66.7)
p- value	0.028		0.044	
≤ 99th centile	117 (99.1)	4 (66.7)	120 (99.1)	1 (33.3)
>99th centile	1 (0.9)	2 (33.3)	1 (0.9)	2 (66.7)
p-value	0.006		0.001	

The sensitivity & specificity of NT scan for major CHD was 66.7% & 88.4% respectively on using 95th centile cut off (LR+5.75 & LR-0.376) while on using 99th centile cut off sensitivity remained same but it was 99.2% specific (LR +83.3, LR- 0.335), as shown in table 3

Table 3: Sensitivity and specificity of fetal nuchal translucency thickness in screening major CHDs

Nuchal Translucency	sensitivity	specificity	95% confidence interval		Area under curve
			lower	upper	
Using 95th centile (%)	66.7	88.4	0.457	1.094	0.775
Using 99th centile (%)	66.7	99.2	0.502	1.156	0.829

Discussion

Our study shows that only 50% of cardiac abnormalities are associated with increased fetal nuchal translucency thickness (>95th centile for CRL) at 10-14 weeks of gestation. Showing that increased NT is not a good screening tool for CHDs as such. But for major CHDs at 95th centile NT scan sensitivity was 66.7% and specificity was 88.4%, at 99th centile sensitivity remained same but it was 99.2% specific. Concluding that cases with raised Nuchal translucency are at higher risk for major congenital cardiac defects and it constitutes an indication for specialist fetal echocardiography**.** Jon Hyett *et al.*, 1999 [7] A review of 29 154 singleton pregnancies screened for congenital defects by measuring NT thickness at 11–14 weeks' gestation found that NT was above the 95th centile in 56% of fetuses with major cardiac abnormalities. Reporting its sensitivity at 95th centile cut-off to be 56.0% (42.0 to 70.0, 95%CI) and specificity 93.8% (93.6 to 94.1. 95% CI). While at 99th centile cut off sensitivity was 40.0% (26.0 to 54.0, 95%CI) and specificity 99% (98.9 to 99.1, 95%CI). Similarily, In a meta-analysis including 58492 fetuses, Makrydimas *et al.,* (2003) [8] found that a NT measurement > 99th percentile had a sensitivity of 31% and specificity of 98.7% for the diagnosis of major CHD. A sensitivity of 37% and specificity of 96.6% was found using the 95th percentile cut-off. A. Sotiriadis *et al.,* (2013) [9] reported the pooled sensitivity and specificity of NT > 95th centile for diagnosis of major CHDs to be 44.4% (95% CI, 39.5-49.5) and 94.5% (95% CI, 94.4-94.6), respectively. The pooled sensitivity and specificity of NT > 99th centile was 19.5% (95% CI, 15.9-23.5) and 99.1% (95% CI, 99.1-99.2), respectively.
We found that increased NT thickness was associated mainly with left heart lesions and septal defects, an observation similar to previous studies [10, 11]. While it had poor screening potential for right heart lesions.
Thus, routine measurement of the nuchal translucency shows

great promise in helping to identify fetuses at increased risk of major congenital heart disease, and therefore may help direct resource-efficient use of fetal echocardiography. However, by itself, increased nuchal translucency does not predict the diagnosis of a "specific type" of cardiac defect in the fetus.

Conclusion

Measurement of fetal nuchal translucency thickness at 10-14 weeks of gestation traditionally used to identify fetuses at high risk of aneuploidy, also identifies the majority of pregnancies with major heart defects. NT is not a very good screening tool for CHDs, but cases with raised Nuchal translucency are at higher risk for major congenital cardiac defects and it constitutes an indication for specialist fetal echocardiography. Every centre should have nuchal translucency nomogram, and specialist fetal echocardiogram should be done in every case with increased NT.

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