Original Article

Prenatal and postnatal echocardiography in NT fetuses with normal karyotype

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Abstract

Introduction: Targeted fetus echocardiography at midpregnancy can detect major defects in major cardiovascular organs. The present study aimed to evaluate prenatal and postnatal echocardiography in fetuses with increased nuchal translucency (NT) with normal karyotype. **Methods:** In this retrospective study, data on the screening of fetuses in pregnant women between 2014 and 2015 were evaluated. The fetuses at the gestational age were 14–11 weeks, and NT ≥ 95 percentile (or 3 mm). For all fetuses with increased NT, follow-up anomaly scan was performed at 18–22 weeks of pregnancy, while fetal echocardiography was performed at weeks 16–19 of pregnancy. The results were analyzed by Statistical Package for the Social Sciences (version 22) and the level of significance was less than 0.05. **Results:** A total of 26.27% of the fetuses were diagnosed with prenatal heart defects and confirmed after birth. The strongest relationship was observed between increased NT and the diagnosis of prenatal heart defects at 2.5–5.3 mm. The increased NT was higher in younger mothers. Moreover, increased NT was higher in mothers with less body mass index. **Conclusion:** By measuring NT in the 11–13 weeks of pregnancy and considering the risk factors, it is possible to evaluate the probability of cardiac abnormalities in the fetus and perform the necessary diagnostic evaluations for high-risk cases.

Keywords: Cardiac abnormalities, echocardiography, fetus, NT

Introduction

Congenital heart disease is the most recurrent congenital malformations that affects four to eight infants per 100 births. Congenital heart malformations are responsible for most infant deaths in the first year of life. This continued until the late 1970s, when ultrasound imaging resolution reforms began to allow the professionals to evaluate the anatomical structures of heart and associated malformations. The ability to observe the normal anatomical structures and subsequent recognition of cardiac malformations has improved rapidly since then.[1] There is increasing evidence that, in definite cases of cardiac abnormalities and other structural abnormalities, prenatal diagnosis is likely to be beneficial and even life-saving.[2] Neck transparency refers to the normal subcutaneous space between the cervical spine and skin that is seen in the first trimester ultrasonography.[3-5] It is possible to measure the thickness of the fetal NT in the 10–14 week of pregnancy via transabdominal and transvaginal methods. These are sensitive and precise methods for screening chromosomal defects and cardiac defects in both normal and abnormal chromosomal fetuses.[6-11]

Since the 1990s, extensive studies have shown that fetuses with increased nuchal translucency (NT) have a higher risk for a wide range of fetal structural defects, especially congenital heart defects and specific genetic syndromes.[3] Since NT is associated with chromosomal abnormalities and many other defects, parents

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with increased NT are first recommended to use karyotypes. If the karyotype is normal, they will be presented with subsequent ultrasound examinations during pregnancy.[12] The increased NT thickness in the first trimester should provide an indication for follow-up scans with details and even more genetic tests that may detect many of the above-mentioned abnormalities.[4] The number of NT-related abnormalities is not fully understood, but there is a large group of fetuses with NT that ultimately become healthy babies. Therefore, NT alone is not a fetal anomaly. Accordingly, approximately 70%, 50%, 30%, and 15% of fetuses with NTs up to 3.5–4.4 mm, 4.5–5.4 mm, 5.5–6.4 mm, and more and equal to 6.5 mm, respectively, were born healthy after the chromosomal defects were rejected. The prevalence of fetal abnormalities and undesirable pregnancy outcomes increases exponentially with NT thickness, especially when it reaches a thickness of 3.5 mm. Quantitative studies have focused on live-born babies with an increased NT, normal karyotype, and normal ultrasound scans.[13] Despite several studies, there is still uncertainty and concern about the implications of increased NT in fetuses. This causes anxiety in parents.[7] Therefore, the present study aimed to evaluate prenatal and postnatal echocardiography in fetuses with increased NT with normal karyotype in 2014–15.

Materials and Methods

This retrospective study aimed to evaluate the outcomes of the increased NT in terms of chromosomes.

In order to observe ethical principles, the researcher obtained permission and getting the code of ethics (IR.AJUMS.REC.1396.448) from Ahvaz University of Medical Sciences to conduct the research. In this study, the information about the fetuses of 34 women (aged 20–38 years old) referring to the Narges Laboratory of Ahvaz for the first trimester (gestational age of 11–14 weeks, NT ≥ 95 percentile (or mm^3) was extracted. Fetuses with NT were recommended to undergo karyotype by cytogenetic analysis of the substance with chorionic villus sampling (CVS), amniocentesis, or cell-free DNA. All fetuses of with NT underwent follow-up scan at weeks 18–22 of pregnancy and gestational echocardiography at weeks 16–19 of pregnancy. Physical examinations and postnatal echocardiography were performed in babies born alive to confirm the diagnosis of congenital heart disease. This information was completed by checklists and files.

Data analysis

The data were analyzed by Statistical Package for the Social Sciences (version 22) and the significance level was less than 0.05.

Results

In this study, the effect of NT in the first trimester on prenatal and postnatal echocardiography findings was investigated. Moreover, the prenatal and postnatal echocardiographic findings were compared. The relationship between NT and congenital heart abnormalities was investigated. The results are shown in Table 1.

In Table 1, NT was divided into six categories and the prenatal and postnatal echocardiography response was based on normal and abnormal NT.

The effect of maternal body mass index (BMI) on pre-and postbirth echocardiography findings was evaluated, and the results of this assessment can be seen in Table 2.

BMI of mothers was divided into three groups: <24.99, 25.-29.99, >30, respectively. The prenatal and postnatal echocardiography response was based on normal and abnormal BMI.

The effect of maternal age on prenatal and postnatal echocardiography was examined. The mothers' age group was divided into four categories, the results of which are shown in Table 3.

According to Table 4, ten fetuses (26.32%) were identified with prenatal heart defects, which were confirmed after birth.

Discussion

The results have shown that there is a strong relationship between NT and abnormalities of the heart. This is found in fetuses with normal and abnormal chromosomes. The present study aimed

Table 1: Comparison of the prenatal and postnatal echocardiographic findings based on NT

NT echocardiography	1.5-2.4 mm (*n*=15)	2.5-3.4 mm (*n*=16)	3.5-4.4 mm (*n*=3)	4.5-5.4 mm (*n*=3)	5.5-6.4 mm (*n*=0)	≥6.5 mm (*n*=1)
Before and after normal birth	5 (45.45%)	3 (27.27%)	1 (9.09%)	2 (18.18%)	-	0
P			0.027*			
Before and after abnormal birth	3 (30%)	6 (60%)	0	1 (10%)	-	0
P			0.040*			
Before normal birth; after abnormal birth	2 (66.67%)	1 (33.33%)	0	0	-	0
P			0.287			
Before abnormal birth; after normal birth	5 (35.71%)	6 (42.86%)	2 (14.29%)	0	-	1 (7.14%)
P			0.007*			

Table 2: Comparison of the results of prenatal and postnatal echocardiography based on BMI of mothers

BMI Echocardiography	< 24.99 (*n*=7)	25-29.99 (overweight) (*n*=24)	≥30 (Obesity) (*n*=7)
Before and after normal birth	2 (18.18%)	7 (63.64%)	2 (18.18%)
P		0.223	
Before and after abnormal birth	1 (10%)	7 (70%)	2 (20%)
P		0.189	
Before normal birth; after abnormal birth	0	1 (33.33%)	2 (66.67%)
P		0.199	
Before abnormal birth; after normal birth	4 (28.57%)	9 (64.29%)	1 (7.14%)
P		0.089	
NT (mean±SD)	2.87±0.83	3.13±1.22	2.3±0.4

Table 3: Comparison the results of prenatal and postnatal echocardiography based on the age of mothers

Age Echocardiography	19<age ≤25 (*n*=7)	25<age ≤30 (*n*=11)	30<age ≤35 (*n*=15)	> 35 (*n*=5)
Before and after normal birth	3 (27.27%)	3 (27.27%)	4 (36.36%)	1 (9.09%)
P		0.238		
Before and after abnormal birth	2 (20%)	3 (30%)	3 (30%)	2 (20%)
P		0.261		
Before normal birth; after abnormal birth	1 (33.33%)	0	1 (33.33%)	1 (33.33%)
P		0.261		
Before abnormal birth; after normal birth	1 (7.14%)	5 (35.72%)	7 (50%)	1 (7.14%)
P		0.238		
NT (mean±SD)	2.66±0.81	3.68±1.4	2.72±0.86	2.35±0.35

Table 4: The distribution of echocardiography (ECO) diagnosis before and after normal/abnormal birth

Echocardiography	*n*	Percentage	*P*
Before and after normal birth	11	28.95%	0.213
Before and after abnormal birth	10	26.32%	
Before normal birth; after abnormal birth	3	7.89%	
Before abnormal birth; after normal birth	14	36.84%	

to investigate the prenatal and postnatal echocardiography in fetuses with normal karyotype. Accordingly, 34 women aged 20–38 years (mean age 30.24 ± 5.464 years old) referring to the clinics in the first trimester were selected, and the necessary information was extracted. Of 34 pregnant women, 4 had twin pregnancies. A total of 38 fetuses were examined. The mean gestational age was 20.95 ± 3.304 weeks and the mean BMI was 27.45 ± 3.168. The fetuses with NT were recommended to undergo karyotype via amniocentesis, CVS, or cell-free DNA. Amniocentesis (60.53%) was used more than other methods in karyotype studies. The parameters studied include determining the effect of NT during the first trimester on the prenatal and postnatal echocardiography findings, comparing the prenatal and postnatal echocardiographic findings, determining the relationship between NT and congenital heart abnormalities, determining the relationship between the age of the mother and an increased incidence of congenital heart defects. In the past, several other studies have also examined the relationship between NT and congenital anomalies. In this section, the results of the present study are compared with those of previous studies.

According to the results, 26.27% of the fetuses were diagnosed with prenatal heart defects, all of which were confirmed after birth. Besides, the results showed that the diagnosis of prenatal heart defects at a thickness of 2.5–4.5 mm should be paid much attention. Meanwhile, at a thickness of more than 6.5 mm, further examinations are needed.

The results of this study were in good agreement with those of Orosz *et al.* (2009), in which it was concluded that the prevalence of heart defects increased with increasing NT.[3] Moreover, the results of this study were in good agreement with those of Bilardo *et al.* (1998), in which there was a strong relationship between NT measurements and congenital anomalies.[11] Saldanha *et al.* (2009) showed that pre-natal and post-natal defects and NT-related abnormalities were associated with NT thickness. This was in agreement with the results of our study.[13]

Barker *et al.* (2007) carried out a study on 967 people and showed that congenital heart disease increased only in 4.9% of newborns with NT. The normal karyotype was also reported.[14] Mayon *et al.* (2000) argued that the prevalence of abnormalities in infants with NT was very low.[15]

In this study, postnatal extracorporeal anomalies were studied in all neonates, all of which were reported as normal. This conclusion was not consistent with the results of Orosz *et al.* (2009), in which other large anomalies were elevated with increasing NT.[3] Bilardo *et al.* (2007) reported that one out of five fetuses with NT had adverse pregnancy outcomes and prenatal abnormalities, which was not consistent with our study.[12]

The results showed that NT was the lowest in mothers aged 35≤, suggesting that NT is higher in younger mothers. The results

of our study were in good agreement with those of Maymon *et al.* (2000), which concluded that the prognosis of fetuses with large NT (>5 mm) was more reliable in younger women.[15]

Conclusion

According to the results, it can be said that by measuring NT in the 11–13 weeks of pregnancy and considering the risk factors, it is possible to investigate the probability of cardiac abnormalities in the fetus and to carry out the necessary diagnostic evaluations for high risk cases. The results also showed that an increase in BMI and age of mothers decreases the possibility of an accurate diagnosis of cardiac anomalies in the fetus.

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Conflicts of interest

There are no conflicts of interest.

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