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ASSESSMENT OF THE VALUE OF DUCTUS VENOSUS DOPPLER STUDY IN THE DETECTION OF CONGENITAL MALFORMATIONS IN PREGNANCIES WITH NORMAL NUCHAL TRANSLUCENCY AND NASAL BONE

Radiology

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ABSTRACT

To assess the value of the first trimester Ductus Venosus doppler study in the detection of congenital malformations in pregnancies with normal nuchal translucency and nasal bone. The study population included 100 women between 20 to 30 years of age who attended ASRAM hospital, Eluru, from December 2018 to March 2019. Nuchal translucency(NT) was measured, and the presence or absence of nasal bone was determined. The fetal Ductus Venosus pulsatility index for veins (PIV) and the lowest velocity during atrial contraction (A·wave) were evaluated in 100 singleton pregnancies at 11 to 14 weeks in fetuses with normal NT and the presence of nasal bone. One hundred antenatal women between the age of 20-30 years at 11 -14 weeks of gestation were included in the study. The NT measurement in the sample population ranged from 0.9 mm to 1.8 mm, with a mean range of 1.2mm. The presence of nasal bone was documented in all the fetuses. Ductus venosus waveform was found to be normal in all the fetuses with the PIV ranging from 0.9- 3 and an average of 1.5+/- 0.2. The pregnancies were followed up with TIFFA and into the postnatal period and were found to be without any congenital anomalies. Fetuses with normal NT and nasal bone had normal ductus venosus blood flow in the first trimester and showed no congenital malformations on followup. Despite some reports showing adverse fetal outcomes such as congenital heart diseases and fetal growth restriction in fetuses with abnormal DV flow in the presence of normal NT, evaluating such a small vessel (~2 mm) with Doppler sonography in the first trimester may preclude its practical use in screening for fetal abnormalities especially in fetuses with normal NT and nasal bone. However, the study has to be conducted on a larger population to draw a final inference.

KEYWORDS

Ductus Venosus(DV), nuchal translucency (NT), Nasal bone

1.INTRODUCTION

Major fetal malformations are associated with fetal chromosomal anomalies, particularly central nervous system anomalies, facial abnormalities, cystic hygroma, diaphragmatic hernia, gastrointestinal, and genitourinary anomalies, non-immune hydrops fetalis, and abnormalities involving extremities. The nuchal translucency (NT) measurement in combination with maternal serum analytes, pregnancy-associated plasma protein-A (PAPP-A), and free b HCG (human chorionic gonadotropin), is the most common aneuploidy screening paradigm in the first trimester of pregnancy.

The documentation of nasal bone, along with NT and serum screening, can increase the detection rate of trisomy 21 without significantly altering the false positive rate.

The Ductus Venosus is a vessel that transfers the oxygen-and nutrient-rich blood from the umbilical vein to vital organs in the fetus. It has a low-velocity flow that is forward throughout the cardiac cycle. The potential changes in these velocities based on underlying pathology can be different and represent changes in cardiac status.

Several studies have proved an association between Down syndrome and abnormal flow in the DV at 11-14 weeks. Recent data shows that an increased nuchal translucency (NT) coupled with an abnormal flow in the DV, can help improve the predictive capacity of detecting major congenital heart defects (CHD) in chromosomally normal fetuses.

2.MATERIAL AND METHODS

This is a prospective study of 100 singleton pregnancies at 11–14 weeks gestation. The study population comprised women between 20-30 years of age who attended the antenatal clinic for regular checkups between December 2018 and March 2019. Routine fetal biometry, measurement of NT, assessment of nasal bone, and Doppler study of Ductus Venosus was performed according to a standardized protocol. They were followed up for the entire period of pregnancy until delivery. Ultrasound examinations were performed trans-abdominally using a GE Voluson S8 ultrasound machine with a 2-6 MHz curvilinear probe.

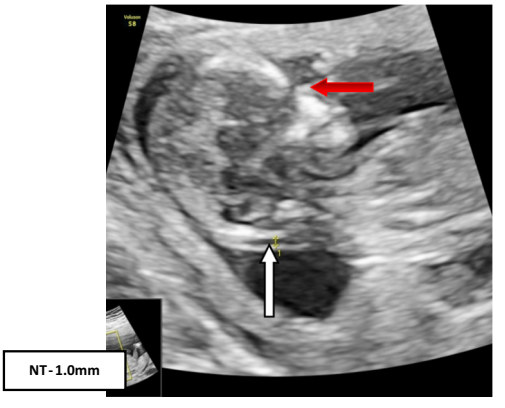

Fig 1: Representative image showing measurement of normal nasal bone(red arrow) and nuchal translucency(white arrow).

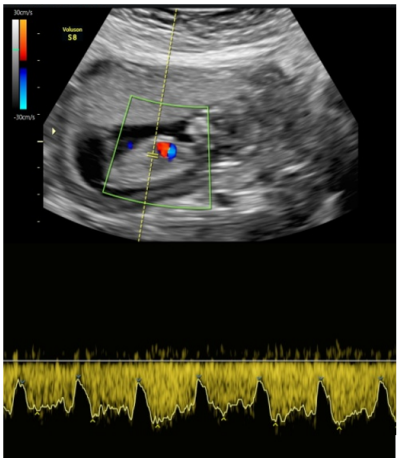

Fig 2: Representative image showing the normal ductus venous Doppler tracing

3.RESULTS

One hundred antenatal women between 11 -14 weeks of gestation who visited our hospital for regular antenatal care were included in the study. All the women were between the age of 20-30 years, with a median age of 25 years. There was no maternal risk or family history of chromosomal abnormalities. The NT measurement in the sample population ranged from 0.9 mm to 1.8 mm, with a mean range of 1.2mm. The presence of nasal bone was documented in all the fetuses. Ductus venosus waveform was found to be normal in all the fetuses with the PIV ranging from 0.9- 3 and an average of 1.5+/- 0.2. The pregnancies were followed up with TIFFA and into the postnatal period and were found to be without any congenital anomalies.

4. DISCUSSION

In the population, 2% to 3% of the newborns have a congenital malformation or genetic disease identified at birth. Despite advances in genetics, the cause of more than half of the human congenital abnormalities remains unknown. The presence of chromosomal abnormalities is documented in about 0.9% of the newborns and include abnormalities of chromosome number as well as structural abnormalities. With the latest developments in medical science, there are multiple tools available for aneuploidy screening, such as maternal serum marker analysis, ultrasound evaluation of nuchal translucency and nasal bone, and prenatal genetic screening with chorionic villus sampling(CVS) and amniocentesis.

NT, which is a crucial sample to these markers, identifies fetuses with trisomy 21 or other major aneuploidies, skeletal dysplasia, and cardiac defects in the first trimester. An increase in NT is recognized as an early soft marker of a broad range of chromosomal, genetic, and structural abnormalities. Therefore, first-trimester US to assess NT has evolved as a screening tool that is widely available to the general obstetric population. NT is a powerful prenatal marker for Down syndrome, with detection rates ranging from 63% to 77% with a 5% false-positive rate. Identification of increased NT alerts the practitioner to the risk of aneuploidy and should prompt a thorough anatomic survey for structural abnormalities and a detailed fetal echocardiogram, even in the setting of a normal karyotype.

Incorporation of nasal bone evaluation in the first-trimester screening protocol has been attempted by researchers to improve the accuracy of aneuploidy diagnosis. The nasal bone measurement improved the detection rate of Down syndrome to 90% and reduced the false positive rate to 2.5%.

The DV is a fetal blood vessel that shunts highly oxygenated blood from the umbilical vein to the right atrium, after which it is shunted to the foramen ovale to provide oxygenated blood to the fetal circulation. Doppler study of DV typically shows triphasic, pulsatile forward flow. Absent a- wave or reversed a- wave during atrial systole has been recognized in cases of fetal cardiac malformations and aneuploidy**.**

Maiz et al. [1] found that in fetuses with increased NT and normal genome following invasive testing, an absent or reversed a-wave in the DV is associated with a three-fold increase in the likelihood of major cardiac defects; however, there is a 50% reduction of the risk for such defects in fetuses with normal DV flow. Martínez et al. .[2] reported that of congenital heart disease (CHD) detection improved from 28.9% to 40.0% in chromosomally normal fetuses with the incorporation of increased NT and abnormal Doppler flow of DV in the first trimester.

Recently, Borrell et al. .[3] proposed that the best method was to combine the NT and DV Doppler for the detection of major CHD in euploid fetuses and demonstrated different detection. They showed that an abnormal DV Doppler could detect approximately 70% of major CHD in the first trimester and is an indication for referral for specialized fetal echocardiography, even when the NT is normal

A recent study reported that an abnormal DV a-wave could detect 83% of CHD cases in association with an increased NT and 19% when the NT was normal[4]. Another study of 40,000 singleton pregnancies at 11-14 weeks with normal chromosomes showed that using an abnormal NT and the reversed a-wave of the DV as a screening tool could detect 47.1% of major CHD[5]. Although the exact mechanism of the abnormal DV flow in fetuses with CHD is still unclear, it is postulated that an abnormal DV flow may reflect impaired atrial contraction and reduced myocardial contractility resulting in subcutaneous edema. Particularly, atrioventricular septal defects with valve regurgitation and hypoplastic left heart syndrome suggest that right atrial volume and pressure overload can reflect the DV and may be responsible for the reversal of the DV a-wave and increased DVPIV.

In about 90% of fetuses with chromosomal abnormalities and increased NT, there is evidence of cardiac dysfunction during the first trimester, with or without structural abnormalities, as documented by associated abnormal ductus flow abnormalities. Nonetheless, since it is also associated with cardiac anomalies in chromosomally normal fetuses, an increased NT thickness may constitute the most effective method of screening for major cardiac and great-vessel abnormalities According to the data collected in our study, which included a relatively small sample size of 100 antenatal patients without any maternal risk factors, we documented the presence of nasal bone in all the fetuses and nuchal translucency recorded was found to be within normal limits. The ductus venosus Doppler study demonstrates a typical waveform without any absence or reversal of A wave. The DVPI also was recorded and found to be within normal limits. The fetuses were followed up with a TIFFA scan and also postnatally to rule out congenital malformations. All the pregnancies had a healthy outcome.

We could conclude from our study that, in antenatal women without any risk factors, in the presence of nasal bone and a normal nuchal translucency, the ductus venosus Doppler study was found to be within normal limits. As these fetuses on a followup up did not show any demonstrable developmental abnormalities, it will be justifiable to say that the presence of a normal NT and a nasal bone along with a normal DV Doppler in the first trimester rules out fetal aneuploids with considerable precision. However, our study population being limited, and our data not including any chromosomal malformations; further studies have to be done to draw a final inference.

5.CONCLUSION

Fetuses with normal NT, presence of nasal bone, and normal ductus venosus blood flow in the first trimester, wherein the mothers had no identifiable risk maternal risk factors, had no congenital malformations on followup. Despite some reports showing adverse fetal outcomes such as congenital heart diseases and fetal growth restriction in fetuses with abnormal DV flow in the presence of normal NT, evaluating such a small vessel (~2 mm) with Doppler sonography in the first trimester may preclude its practical use in screening for fetal abnormalities especially in fetuses with normal NT and nasal bone.

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