Fetal Heart Assessment during the 13-week Scan

Alaa Ebrashy¹, Sherif Elsirgany², Sara H El-Dessouky³

Abstract

Congenital heart defects (CHD) affect nearly between 6 and 12 per 1,000 live births in the general population. The 13-week scan was mainly constructed to perform screening for chromosomal anomalies. Recently there was a strong relation between wide NT and negative ductus venosus (DV) with congenital heart later direct heart examination was achievable at acceptable degree of success and at satisfactory levels during the 13-week scan as part of fetal anatomy check at this age of pregnancy. Early diagnosis of major heart anomalies was done with the application of same principles of examining the heart at 2nd trimester. Most of the literature published recently dealing with fetal anatomy at 13-weeks is mentioning the details of heart examination and how to perform it using all methods available including color Doppler and the results were very satisfactory. The higher the crown-rump length (CRL) the better the ability to visualize the fetal heart in details.

Keywords: Ductus venosus, Fetal heart examination, Fetal scan, Nuchal translucency.

Introduction

Fetal structural malformations affect nearly 2–3% of all pregnancies;¹ among them congenital heart defects (CHDs) affect nearly between 6 and 12 per 1,000 live births in the general population. At least half of these are major cardiac anomalies that require corrective or palliative surgery in the postnatal period.¹⁻³ First-trimester scan was mainly focusing on screening for chromosomal aneuploidies; however, postponing the anatomic assessment to the second trimester may result in delaying the detection of major structural malformations.⁴ It has been demonstrated that ultrasound examination at 11–13 weeks’ gestation leads to the diagnosis of a large set of fetal nonchromosomal abnormalities.⁴⁻⁶ Screening for fetal CHDs was usually performed during the fetal anomaly scan at 18–24 weeks’ gestation; however, following the recognition of early markers for these disorders and the development of better ultrasound resolution, interest has turned toward performing the screening of the fetus, including the heart, at the same time during first-trimester scan especially in high-risk pregnancies for CHD and in families with previous history of cardiac structural malformations.⁷⁻¹⁰ Congenital heart defects represent fetal nonchromosomal abnormalities potentially detectable during the first trimester based on a standardized protocol and the presence of detectable markers for an underlying abnormality, such as increased nuchal translucency (NT) thickness, ductus venosus (DV) abnormal flow, and tricuspid regurgitation (TR).¹¹⁻¹⁹

Increased Nuchal Translucency

Nuchal translucency is defined as the sonographic appearance of a subcutaneous collection of fluid behind the fetal neck; the optimal gestational age for the measurement of fetal NT is 11 weeks of gestation to 13 weeks 6 days of gestation. The minimum fetal crown-rump length (CRL) should be 45 mm, and the maximum length should be 84 mm.²⁰ Since the first description of the association between an increased NT and Down syndrome in 1987, several studies have been performed to elucidate the potential association between increased NT and fetal structural malformations, chromosomal abnormalities, and genetic syndromes. Special attention has been given to its association with CHDs in euploid fetuses.²¹

¹Department of Obstetrics and Gynecology, Fetal Medicine Unit, Cairo University, Cairo, Egypt
²Reproductive Health Research Department, National Research Centre, Dokki, Cairo, Egypt
³Prenatal Diagnosis and Fetal Medicine Department, Human Genetics and Genome Research Division, National Research Centre, Cairo, Egypt

Corresponding Author: Alaa Ebrashy, Department of Obstetrics and Gynecology, Fetal Medicine Unit, Cairo University, Cairo, Egypt, Phone: +201222172441, e-mail: ebrashy63@gmail.com

How to cite this article: Ebrashy A, Elsirgany S, El-Dessouky SH. Fetal Heart Assessment during the 13-week Scan. Donald School J Ultrasound Obstet Gynecol 2020;14(3):231–236.

Source of support: Nil

Conflict of interest: None

Hyett et al.¹¹ initiated the idea of using the NT measurement as a screening tool to increase the still disappointingly low, prenatal detection of CHD. They reported the finding of an increased NT in 56% of the fetuses with CHDs, in a study of 29,154 pregnancies and recognized that by referring the 1% of fetuses with a NT measurement ≥99th percentile for fetal echocardiography in a specialized center, 40% of major CHDs (catheter or surgical intervention required) could potentially be identified.

Since then several studies were performed reporting on the association between increased NT and major heart defects varied in the definition of increased NT, which included NT >95th percentile, ≥95th percentile, >2.5 mm or ≥2.5 mm, and of very increased NT, which included NT >99th percentile, ≥99th percentile, >3.5 mm, ≥3.5 mm, or ≥2.5 multiples of the median.¹⁶⁻¹⁸,¹⁹,²²⁻³⁴

Makrydimas et al.²⁵ in a meta-analysis of eight studies (including 58,492 fetuses) found that a NT > 99th percentile had a sensitivity of 31% and specificity of 98.7%, with a positive predictive value of 24 for the identification of major CHDs, whereas a sensitivity of 37% and specificity of 96.6% were found when using the 95th percentile of NT measurement cutoff. Wald et al.³⁵ in a later meta-analysis of seven studies defined the relationship between an enlarged NT [cutoff 1.7 multiples of the median at a false-positive rate (FPR) of
Fetal Heart Assessment during the 13-week Scan

Abnormal DV Flow

The DV is a small vessel acting as an important regulator of the fetal hemodynamic system; it carries oxygenated blood in a high-velocity jet, its waveform being characterized by arterial-like peak velocities. Flow velocity waveforms in the DV are described as having a peak during ventricular systole (S-wave), another peak during ventricular diastole (D-wave), and a nadir during atrial contraction in late diastole (A-wave). Reversed A-wave was observed in 2% of the normal fetuses with a positive likelihood ratio of 30.5. Most recently, Minnella et al. found that the detection rate (DR) and FPR of NT ≥ 95th percentile were about 37 and 6%, respectively, and that those of NT ≥ 99th percentile were 21 and 1%, respectively, which are consistent with the results of most previous studies. Studies have also shown that the risk for CHD increases with increasing NT measurement without a particular preference for one CHD above another. In counseling couples about the risk of finding a CHD in a chromosomally normal fetus with an enlarged NT, the results of a pooled analysis of 11 studies may be helpful (Fig. 1).

Tricuspid Regurgitation

It has been reported that the assessment of tricuspid flow can improve the performance of first-trimester screening for aneuploidies and CHD. However, the performance of TR as a screening marker for CHD in euploid fetuses is yet to be established. Tricuspid regurgitation was diagnosed if it was found during at least half of systole and with a velocity of over 60 cm/second, since aortic or pulmonary arterial blood flow at this gestation can produce a maximum velocity of 50 cm/s. Tricuspid regurgitation was determined by pulsed wave Doppler. Two studies reported on the association between TR and major heart defects. Pereira et al. examined 40,990 pregnancies, including 85 (0.21%) with a major heart defect and reported that the DR and FPR of TR were 32.9 and 1.3%, respectively; the combination of NT ≥ 95th percentile, TR, or abnormal DV flow had a DR of 57.6% and FPR of 8.0%. Very high velocities (≥ 3.5 mm) were associated with a threefold increased risk of CHDs; the finding of a normal DV flow pattern in this setting halved the CHD risk. A study of 40,000 singleton pregnancies with normal chromosomes at 11–14 weeks showed that screening for CHD using an increased NT and the reversed A-wave of the DV can detect 47.1% of major CHD with an FPR of 6.7%.

Several studies reported on the association between abnormal flow in the DV, defined as reversed or absent/reversed A-wave, and major heart defects. The number of pregnancies examined varied from 1,066 to 40,990, the incidence of heart defects varied from 0.21 to 0.74%, the DR varied from 20 to 39%, and the FPR varied from 1.7 to 5.5%. Chelemen et al. underwent a prospective first-trimester screening for aneuploidies, including measurement of fetal NT and assessment of DV flow. They examined 85 cases with major cardiac defects and 40,905 without cardiac defects. The fetal NT was above the 95th and above the 99th centile in 30 (35.3%) and 18 (21.2%) of the fetuses with cardiac defects, respectively, and in 1,956 (4.8%) and 290 (0.7%) of those without cardiac defects, respectively. Reversed A-wave was observed in 24 (28.2%) of the fetuses with cardiac defects and in 856 (2.1%) of those with no cardiac defects. Specialist fetal echocardiography for cases with NT above the 99th centile concluded that assessment of DV flow improves the performance of NT screening for cardiac defects.

Maiz et al. in a meta-analysis of seven studies, including 600 chromosomally normal fetuses with an NT ≥ 95th percentile, found that an abnormal DV flow pattern at 11–14 weeks’ gestation in the presence of an NT ≥ 3.5 mm was associated with a threefold increased risk of CHDs; the finding of a normal DV flow pattern in this setting halved the CHD risk. A study of 40,000 singleton pregnancies with normal chromosomes at 11–14 weeks showed that screening for CHD using an increased NT and the reversed A-wave of the DV can detect 47.1% of major CHD with an FPR of 6.7%.

...
similar results were obtained by Volpe et al., who examined 4,445 pregnancies, including 18 (0.40%) with a major heart defect, and reported that the DR and FPR of TR were 33.3 and 1.7%, respectively; the combination of NT > 95th percentile, TR, or abnormal DV flow had a DR of 55.6% and FPR of 10.1%.

**EARLY FETAL ECHOCARDIOGRAPHY**

Early detailed assessment of the fetal heart can be used to screen for fetal cardiac anomalies prior to that routinely performed in the second trimester. Nowadays there is supporting evidence that the first-trimester ultrasound is a safe procedure provided that the thermal and mechanical indices are taken into account. Additionally, cardiac abnormalities may be an indication for the presence of other subtler anomalies, syndromes, chromosomal abnormalities, or even rarer conditions, such as infections or metabolic disorders. Despite the relatively small size of cardiac anatomical structures at this early stage, combing high-resolution transducers for transabdominal (TA) and transvaginal (TV) imaging, color-flow mapping, and operator experience has led to the identification of many cardiac malformations at this stage. The standard protocol: At the 11–13-week scan, the protocol included a transverse section of the thorax and use of color Doppler to assess the four-chamber view of the heart and outflow tracts and blood flow across the tricuspid valve and in the DV. At the routine second- and third-trimester scans, the protocol included a sweep through the heart in the transverse plane to assess the four-chamber view, outflow tracts, and three-vessel view. Presence or absence of TR was determined by pulsed-wave Doppler during fetal quiescence. A Doppler sample volume of 3.0 mm was positioned across the tricuspid valve and partially in the right atrium in an apical four-chamber view of the fetal heart such that the angle with the direction of flow was less than 30°. The tricuspid valve could be insufficient in one or more of its three cusps and therefore the sample volume was placed across the valve at least three times in an attempt to interrogate the complete valve. The diagnosis of TR was made if it was found during at least half of the systole and with a velocity of >60 cm/second. Care was taken not to misinterpret with the aortic flow overlap, which has a much slower (30–50 cm/second) flow pattern at this early gestational age.

Several studies have compared the accuracy of the early fetal echocardiography with the more conventional 18-week to 22-week fetal echo. Haak et al. underwent a study to explore the possibilities of fetal echocardiography in 85 women with uncomplicated singleton pregnancies; three transvaginal ultrasound examinations between 11 + 0 and 13 + 6 weeks’ gestation were performed and they concluded that the ability to perform a full cardiac examination increased from 20% in week 11 to 92% in week 13. Zidere et al. found that a high degree of accuracy in the identification of CHD can be achieved by early fetal echocardiography (sensitivity 84.8% (95% CI, 75.0–91.9), specificity 95.3% (95% CI, 93.9–96.4)), although the identification of every case of tetralogy of Fallot and small AVSDs presents particular diagnostic challenges at this gestational age. A small but significant group showed progression of findings during this stage of rapid fetal heart growth, particularly in obstructive lesions.

Orlandi et al. performed early fetal heart examination to 4,820 singleton pregnant women aiming to assess the accuracy of a simplified fetal cardiac study, inclusive of four-chamber view (4CV) and ventricular outflow tracts, performed during the 11–14-week screening by well-trained obstetricians to detect CHDs. Among the 4,030 included cases, 32 CHD cases were detected (20 major and 12 minor); 18 of the major (90%) and 5 of the minor (42%) were detected or suspected in the first trimester, 1 major and 6 minor in the second trimester, and 1 major and 1 minor only after birth. Their conclusion was that the simplified protocol is an effective tool to screen for CHD at 11–14 weeks. Ebrashy et al. examined fetal heart in 3,240 pregnant women during the first trimester between 11 weeks and 13 + 6 weeks. Grey scale and color mapping were used in the determination of four chambers and outflow tracts and showed that the success rate was 97% in visualization of both...
Fetal Heart Assessment during the 13-week Scan

Donald School Journal of Ultrasound in Obstetrics and Gynecology, Volume 14 Issue 3 (July–September 2020)

the pulmonary trunk and the three-vessel view while it was 94% in the long-axis aorta and crossing arteries. The conclusion was a high degree of accuracy in the identification of CHD can be achieved by a first-trimester fetal echocardiography and it was recommended that the best time for early fetal echocardiography is between 13 and 13 + 6 weeks.

Figures 2 and 3 show a normal and abnormal fetal heart during first trimester.

CONCLUSION AND RECOMMENDATIONS

Fetal heart assessment done as a part of fetal anatomy scan as a part of the 13-week scan is now considered to be integral part of such examination. The NT and DV as well as TR are considered to be screening methods for heart anomalies at this age of pregnancy with variable degree of sensitivity and specificity. Early fetal echocardiography between 12 weeks and 14 completed weeks' gestation is very useful and is promising for detection of CHD. Well-trained obstetrician or fetal medicine specialist can perform full heart examination within this time frame of the 13 weeks' scan. The higher the crown-rump length (CRL) the better visualization of the fetal heart. We recommend that the best timing to perform a first-trimester anatomy scan including fetal cardiac scan is between 13 and 13 + 6 weeks. Cases with small CRL (less than 45 mm) are recommended to be booked later for the first-trimester scan and fetal heart examination specially now where the fetal anatomy scan at 13 weeks is gaining major attention in the era of cell-free DNA testing of maternal blood.49

REFERENCES

1. Arslan E, Büyükkurt S, Sucu M, et al. Detection of major anomalies during the first and early second trimester: single-center results of six years. J Turk Ger Gynecol Assoc 2018;19(3):142–145. DOI: 10.4274/jtggaa.2017.0125.
2. Lai S, Lau L, Leung C, et al. Is ultrasound alone enough for prenatal screening of trisomy 18? A single centre experience in 69 cases over 10 years. Prenat Diagn 2010;30(11):1094–1099. DOI: 10.1002/pd.2623.
3. Yun W. Congenital heart disease in the newborn requiring early intervention. Korean J Pediatr 2011;54(5):183–191. DOI: 10.3345/kjp.2011.54.5.183.
4. Souka AP, Snijders RJ, Novakov A, et al. Defects and syndromes in chromosomally normal fetuses with increased nuchal translucency thickness at 10–14 weeks of gestation. Ultrasound Obstet Gynecol 1998;11(6):391–400. DOI: 10.1046/j.1469-0705.1998.11060391.x.
5. Syngelaki A, Chelemen T, Dagklis T, et al. Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11–13 weeks. Prenat Diagn 2011;31(1):90–102. DOI: 10.1002/pd.2642.
6. Syngelaki A, Hammami A, Bower S, et al. Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks’ gestation. Ultrasound Obstet Gynecol 2019;54(4):468–476. DOI: 10.1002/uog.20844.
7. Rossi AC, Prefumo F. Accuracy of ultrasonography at 11–14 weeks of gestation for detection of fetal structural anomalies, a systematic review. Obstet Gynecol 2013;122(6):1160–1167. DOI: 10.1097/AOG.0000000000000015.
8. Johnson B, Simpson L. Screening for congenital heart disease: A move toward earlier echocardiography. Am J Perinatol 2007;24(8):449–456. DOI: 10.1055/s-2007-986681.
9. Sairam S, Carvalho JS. Early fetal echocardiography and anomaly scan in fetuses with increased nuchal translucency. Early Hum Dev 2012;88(5):269–272. DOI: 10.1016/j.earlhumdev.2012.02.008.
10. Rogers L, Li J, Liu L, et al. Advances in fetal echocardiography: Early imaging, three/four dimensional imaging, and role of fetal echocardiography in guiding early postnatal management of congenital heart disease. Echocardiography 2013;30(4):428–438. DOI: 10.1111/echo.12211.
23. Chelemen T, Syngelaki A, Maiz N, et al. Contribution of ductus venosus Doppler in first trimester screening for major cardiac defects. Fetal Diagn Ther 2011;29(2):127–134. DOI: 10.1159/000322138.

24. Pereira S, Ganapathy R, Syngelaki A, et al. Contribution of fetal tricuspid regurgitation in first-trimester screening for major cardiac defects. Obstet Gynecol 2011;117(6):1384–1391. DOI: 10.1097/AOG.0b013e3182ee1a720.

25. Borrell A, Grande M, Bennasar M, et al. First-trimester detection of major cardiac defects with the use of ductus venosus blood flow. Ultrasound Obstet Gynecol 2013;42(1):51–57. DOI: 10.1002/uog.12349.

26. Shanizadeh M, Adibi A, Kazemi K, et al. Normal reference range of fetal nuchal translucency thickness in pregnant women in the first trimester, one center study. J Res Med Sci 2015;20(10):969–973. DOI: 10.4103/1735-1995.172786.

27. Clur S-AB, Bilardo CM. Early detection of fetal cardiac abnormalities: How effective is it and how should we manage these patients. Prenatal Diagnosis 2014;34(13):1235–1245. DOI: 10.1002/pd.4466.

28. Michalidis GD, Economides DL. Nuchal translucency measurement and pregnancy outcome in karyotypically normal fetuses. Ultrasound Obstet Gynecol 2001;17(2):102–105. DOI: 10.1046/j.1469-0705.2001.00341.x.

29. Mavrides E, Cobian-Sanchez F, Tekay A, et al. Limitations of using first-trimester nuchal translucency measurement in routine screening for major congenital heart defects. Ultrasound Obstet Gynecol 2001;17(2):106–110. DOI: 10.1046/j.1469-0705.2001.00342.x.

30. Hafner E, Schuller T, Metzenbauer M, et al. Increased nuchal translucency and congenital heart defects in a low-risk population. Prenat Diagn 2003;23(12):985–989. DOI: 10.1002/pd.721.

31. Makrydima G, Sotiридios A, Ioannidis JP. Screening performance of first-trimester nuchal translucency for major cardiac defects: a metaanalysis. Ultrasound Obstet Gynecol 2003;18(5):1330–1335. DOI: 10.1067/mso.2003.97803.00645-8.

32. Bruns RF, Moron AF, Murta CG, et al. The role of nuchal translucency in the screening for congenital heart defects. Arq Bras Cardiol 2006;87(3):307–314. DOI: 10.1590/s0066-782x2006000600013.

33. Westin M, Saltvedt S, Bergman G, et al. Is measurement of nuchal translucency thickness a useful screening tool for heart defects? A study of 16 383 fetuses. Ultrasound Obstet Gynecol 2006;27(6):632–639. DOI: 10.1002/uog.2792.

34. Müller MA, Clur SA, Timmerman E, et al. Nuchal translucency measurement and congenital heart defects: modest association in low-risk pregnancies. Prenat Diagn 2007;27(2):164–169. DOI: 10.1002/pd.1643.

35. Wald NJ, Morris JK, Walker K, et al. Nuchal translucency and cystic hygroma in screening for fetal major congenital heart defects in a series of 12 910 euploid pregnancies. Ultrasound Obstet Gynecol 2010;35(3):273–279. DOI: 10.1002/uog.7534.

36. Chelemen T, Syngelaki A, Maiz N, et al. Contribution of ductus venosus Doppler in first trimester screening for major cardiac defects. Ultrasound Obstet Gynecol 2004;23(4):341–345. DOI: 10.1002/uog.1025.

37. Wieheec M, Nocun A, Matyszkiwicz A, et al. First trimester severe ductus venosus flow abnormalities in isolation or combination with other markers of aneuploidy and fetal anomalies. J Perinat Med 2016;44(2):201–209. DOI: 10.1515/jpm-2014-0323.

38. Volpe P, Ubaldo P, Volpe N, et al. Fetal cardiac evaluation at 11–14 weeks by experienced obstetricians in a low-risk population. Prenat Diagn 2011;31(11):1054–1061. DOI: 10.1002/pd.2831.

39. Wald NJ, Morris JK, walker K, et al. Prenatal screening for congenital heart defects using nuchal: Translucency: a metaanalysis. Prenat Diagn 2008;28(12):1094–1104. DOI: 10.1002/pd.2124.

40. Minnella GP, Crupano FM, Syngelaki A, et al. Diagnosis of major heart defects by routine first-trimester ultrasound examination: association with increased nuchal translucency, tricuspid regurgitation and abnormal flow in ductus venosus. Ultrasound Obstet Gynecol 2020;55(5):637–644. DOI: 10.1002/uog.21956.

41. Sotiriadis A, Papateodorou S, Eleftheriades M, et al. Nuchal translucency and major congenital heart defects in fetuses with normal karyotype: a meta-analysis. Ultrasound Obstet Gynecol 2013;42(4):383–389. DOI: 10.1002/uog.12488.

42. Clur SA, Ottenkamp J, Bilardo CM. The nuchal transculency and the fetal heart: a literature review. Prenat Diagn 2009;29(8):739–748. DOI: 10.1002/pd.2281.

43. Bahmann F, Wellek S, Reinhardt J, et al. Reference of ductus venosus flow velocities and calculated waveform indices. Prenat Diagn 2009;29(8):739–748. DOI: 10.1002/pd.2281.
47. Zidere V, Bellsham-Revell H, Persico N, et al. Comparison of echocardiographic findings in fetuses at less than 15 weeks’ gestation with later cardiac evaluation. Ultrasound Obstet Gynecol 2013;42(6):679–686. DOI: 10.1002/uog.12517.

48. Orlandi E, Rossi C, Perino A, et al. Simplified first-trimester fetal cardiac screening (four chamber view and ventricular outflow tracts) in a low-risk population. Prenat Diagn 2014;34(6):558–563. DOI: 10.1002/pd.4348.

49. Ebrashy A, Aboulghar M, Elhodiby M, et al. Fetal heart examination at the time of 13 weeks scan: A 5 years’ prospective study. Perinat Med 2019;47(8):871–878. DOI: 10.1515/jpm-2019-0222.