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Fetal heart defects: Potential and pitfalls of first-trimester detection

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SUMMARY

Congenital heart defects (CHDs) are the leading cause of infant mortality due to birth defects. In the last 15 years, with the shift in screening for aneuploidies to the first trimester, extensive research has concentrated on early screening and detection of CHDs. Early detailed assessment of the fetal heart requires a high level of expertise in early anomaly scanning and fetal echocardiography. However, the detection of major CHDs at 11–13 weeks is influenced by their association with easily detectable markers, such as the nuchal translucency, ductus venosus blood flow and tricuspid regurgitation, and a policy decision as to the objectives of this scan and the allocation of resources necessary to achieve them. The use of transvaginal ultrasound and newer techniques are likely to improve the detection rate. However, the limitations of fetal echocardiography in the first trimester must be borne in mind, and follow-up at mid-gestational echocardiography is prudent in some cases.

1. Introduction

Congenital heart defects (CHDs) account for one-third of all congenital anomalies and are the leading cause of infant mortality due to birth defects.[1] They are commonly associated with fetal aneuploidy and genetic syndromes. In the last 30 years extensive studies have reported the prenatal diagnosis of cardiac defects during the second trimester of pregnancy.[2] However, in the last 15 years, with the shift in screening for aneuploidies to the first trimester, extensive research has concentrated on early screening and detection of CHDs.[3–8] Although the primary aims of the early ultrasound scan, which takes place at 11–13 weeks of gestation, are dating of the pregnancy, detection of multiple pregnancies and screening for aneuploidies there is increasing emphasis on the early detection of major defects. The advantages of early detection of major fetal defects include the possibility of scheduling additional assessment well before the limits for legal termination, the option for an earlier and safer pregnancy termination, and, in cases with a normal scan, earlier reassurance that a major defect is unlikely to be present.[9,10]

This article reviews the detection of major cardiac defects during the first trimester of pregnancy including description of the markers which could help identify the high-risk group requiring specialist fetal echocardiography and the techniques which could improve the detection of these defects.

2. Detection rate of congenital cardiac defects in the first trimester

The results of screening studies providing data on the prevalence of cardiac abnormalities and the proportion detected in the first-trimester scan are summarised in Table 1.[3–8,11–30] In most of these studies, all abnormalities were classified by the authors as being major. Most studies included only euploid fetuses but four included fetuses with aneuploidies. The combined data on specific groups of cardiac abnormalities and their early detection in euploid fetuses from 14 studies that provided such details are presented in Table 2. The early detection rate for the most common cardiac abnormalities varied from around 51% for hypoplastic left heart to 16% for coarctation of the aorta, 18% for tetralogy of Fallot and transposition of the great arteries.[3–8,11–28]

The largest study, involving 44 859 singleton pregnancies undergoing a first-trimester ultrasound scan as part of routine screening for aneuploidies, reported that the detection rate of major CHDs was 34%.[3] The study reported that this scan led to the diagnosis of around half of the cases of double outlet right ventricle, hypoplastic left heart and transposition of the great arteries, around one-third of the cases of atrio-ventricular septal defect, coarctation of the aorta, tetralogy of Fallot and pulmonary atresia,

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Table 1
Screening studies reporting on the effectiveness of the first-trimester scan in the diagnosis of major fetal cardiac abnormalities.

Study	Total	Scan route	GA (weeks)	Minor defects excluded	Prevalence	Aneuploidies	Early detection	Increased NT	
								Cut-off	Prevalence
Hernádi and Töröcsik[11]	3991	TA, TV	11–14	2	1 (0.02%)	–	–	–	Not stated
D'Ottavio et al.[4]	4078	TV	13–14	2	12 (0.29%)	–	3 (25.0%)	–	Not stated
Bilardo et al.[13]	1690	TA, TV	10–14	–	4 (0.23%)	–	–	3.0 mm	2 (50.0%)
Hafner et al.[14]	4233	TA	10–14	5	14 (0.33%)	–	1 (7.1%)	2.5 mm	4 (28.6%)
Hyett et al.[12]	29 154	TA	10–14	7	43 (0.15%)	–	1 (2.3%)	95th centile	25 (58.1%)
Schwarzler et al.[19]	4523	TA	10–14	2	9 (0.20%)	–	–	2.5 mm	1 (11.1%)
Mavrides et al.[17,a]	7339	TA	10–14	2	24 (0.33%)	–	4 (16.7%)	2.5 mm	4 (16.7%)
Michailidis and Economides[18]	6650	TA, TV	10–14	2	9 (0.14%)	–	2 (22.2%)	95th centile	2 (22.2%)
Orvos et al.[20]	4309	TV	10–13	7	32 (0.74%)	–	–	3.0 mm	16 (53.3%)[b]
Taipale et al.[5]	4789	TV	10–16[c]	7	18 (0.38%)	–	1 (5.6%)	3.0 mm	4 (22.2%)
Chen et al.[6]	1609	TA, TV	12–14	5	7 (0.44%)	4 (57.1%)	4 (57.1%)	–	Not stated
Bahado-Singh et al.[21]	8167	TA	10–14	15	6 (0.07%)	–	–	2.5 mm	3 (50.0%)
Bruns et al.[22]	3664	?	11–14	11	9 (0.25%)	–	–	95th centile	2 (22.2%)
Becker and Wegner[23]	3094	TA, TV	11–14	–	11 (0.36%)	–	6 (54.5%)	2.5 mm	6 (54.5%)
Cedergren and Selbing[7]	2708	TA	11–14	6	3 (0.11%)	–	–	–	Not stated
Dane et al.[16]	1290	TA	11–14	–	1 (0.08%)	–	–	–	Not stated
Westin et al.[24,d]	16 260	TA	12–14	–	29 (0.18%)	–	–	3.0 mm	2 (6.9%)
Muller et al.[25]	4144	TA	10–14	–	13 (0.31%)	–	–	99th centile	2 (15.4%)
Chen et al.[15]	7642	TA	10–14	13	19 (0.25%)	10 (52.6%)	7 (36.8%)	–	Not stated
Oztekin et al.[26]	1805	TA	11–14	1	2 (0.11%)	–	–	95th centile	0 (0.0%)
Hildebrand et al.[27]	21 189	?	11–14	–	62 (0.29%)	–	0	–	Not stated
Syngelaki et al.[3]	44 859	TA, TV[e]	11–13	–	106 (0.24%)	–	36 (34%)	95th centile	30 (28.3%)
Volpe et al.[8]	4445	TA, TV	11–14	11	28 (0.63%)	10 (35.7%)	23 (82.1%)	95th centile	14 (50%)
Grande et al.[28]	13 723	TA, TV	11–14	80	44 (0.32%)	312 (2.2%)	25 (56.8%)	97.5th centile	16 (36.4%)

GA, gestational age; TA, transabdominal; TV, transvaginal; NT, nuchal translucency., In some of the studies, the authors included minor defects (atrial or ventricular septal defect) and functional abnormalities (tricuspid or aortic regurgitation) and in this analysis we have excluded these abnormalities.
Adapted from Syngelaki et al.[11]
[a] Includes the data published by Carvalho et al.[29] (not shown) and 25% of data of Schwarzler et al.[19]
[b] NT available in 30 of the 32 fetuses with cardiac defects.
[c] 10% of the population were above 14 weeks.
[d] Includes all data published by Westin et al.[30]
[e] TA mainly, only TV if inadequate views.

but none of the cases of ventricular septal defect, Ebstein anomaly, aortic or pulmonary stenosis, tricuspid atresia or cardiac tumours.[3]

A recent review of the published series with more than 1000 cases from 1993 to 2008, which included data from 36 237 pregnancies generated by eight centres, suggests that the overall detection rate of major congenital anomalies at 11–13 weeks is 29% (95% confidence interval: 25–33).[27] The pooled detection rate of cardiac defects was 17% (10–25%). The authors suggested that the detection rate could be improved if the ultrasound assessment at the first trimester follows well-delineated protocols.[31]

3. Approach to ultrasound examination of the heart in the first trimester

The basic principles are the same as ultrasound examination of the heart in the second or third trimester but colour flow mapping

Table 2
Studies providing details on the early diagnosis of specific cardiac abnormalities in euploid fetuses.

Cardiac abnormality	Screening study[a]														Total
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	
Coarctation of the aorta		0/1		0/3		0/1		0/1	2/5	0/2	0/8	4/15	0/1		6/37 (16.2%)
Tetralogy of Fallot		1/2	0/1	0/2		0/1	0/1		0/3	0/2	0/9	3/10	2/3		6/34 (17.6%)
Hypoplastic left heart		1/2	0/2	1/3				0/1	1/2	1/3	0/6	5/10	2/2	10/10	21/41 (51.2%)
Transposition of the great arteries				0/2	0/1			0/3	0/3	0/1	0/8	2/5	2/2	2/8	6/33 (18.2%)
Atrioventricular septal defect		0/3		0/7							0/2	3/9	8/9	4/5	15/35 (42.9%)
Pulmonary stenosis			0/1	0/1		0/1		0/1	0/4		0/1	0/5	1/1		1/15 (6.7%)
Aortic stenosis		1/3							0/2			0/1	2/3		3/9 (33.3%)
Tricuspid atresia	0/1								0/2			1/1	1/1		2/5 (40.0%)
Ebstein's anomaly								0/1			0/2	0/5			0/8 (0.0%)
Double outlet right ventricle					0/2							4/7			4/9 (44.4%)
Anomalous pulmonary venous return		0/1									0/1				0/2 (0.0%)
Mitral atresia											0/1		1/1		1/2 (50.0%)
Interrupted aortic arch											0/1		1/1		1/2 (50.0%)
Pulmonary atresia											0/1	1/3			1/4 (25.0%)
Double inlet left ventricle									0/1						0/1 (0.0%)
Common truncus arteriosus											0/1				0/1 (0.0%)
Ventricular septal defect	1/2			0/7	1/1	0/4		0/8	0/1	0/2	0/7	0/16	5/8		7/56 (12.5%)
Total	1/3	3/12	0/4	1/25	1/4	0/7	0/1	0/15	3/23	1/10	0/48	23/87	25/32	16/23	74/294 (25.1%)

Adapted from Syngelaki et al.[11]
[a] 1, Hernandi and Torocsic[11]; 2, D'Ottavio et al.[4]; 3, Bilardo et al.[13]; 4, Taipale et al.[5]; 5, Chen et al.[6]; 6, Cedergren and Selbing[7]; 7, Dane et al.[16]; 8, Chen et al.[15]; 9, Mavrides et al.[17]; 10, Michailidis and Economides[18]; 11, Hyett et al.[12]; 12, Syngelaki et al.[3]; 13, Volpe et al.[8]; 14, Grande et al.[28].