International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Elhabashy A. Int J Reprod Contracept Obstet Gynecol. 2024 Sep;13(9):2262-2268
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: https://dx.doi.org/10.18203/2320-1770.ijrcog20242471

Original Research Article

Pattern recognition for prenatal sonographic diagnosis of most congenital heart disease: the "7×2" CHD algorithm

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Received: 26 May 2024
Revised: 12 July 2024
Accepted: 15 July 2024

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ABSTRACT

Background: Congenital heart defects (CHD) are common anomalies and are commonly missed prenatally because of the difficult learning curve of fetal echocardiography. The aim study was to establish an algorithm to review most of the fetal CHD using a simple pattern recognition that will provide easy approach for their diagnosis. To our best knowledge this is the first algorithm that gather the salient sonographic features of most CHD.
Methods: We had reviewed 66 cases of CHD that were gathered by one tertiary center from January 2020 till December 2023. We used an algorithmic path to allow easy deferential diagnosis of these anomalies by using certain sonographic features (we call them: altered key points). This work reviewed 14 fetal CHD. After exclusion of isolated ventricular septal defect (VSD).
Results: Each 2 of these CHD has a common altered key point during echocardiography. These 7 key points are: right dominance, left dominance, absent crossover of great arteries, aorta override VSD, abnormal crux, abnormal area behind the heart and abnormal three vessels trachea view. These 14 anomalies were grouped in tables with their salient sonographic markers for easy mind mapping during echocardiography.
Conclusions: The "7×2" CHD algorithm is a simple way that allow easy recognition of 14 fetal CHD by focusing on 7 altered key points that can be recognized on the common sonographic views where each of them has 2 main differential diagnosis. These 14 anomalies represents most of the CHD (78%) after exclusion of isolated VSD.

Keywords: fetal echocardiography, CHD, differential diagnosis, pattern recognition, the "7×2" algorithm

INTRODUCTION

Congenital heart defects (CHD) are common as they represent about third of fetal anomalies and about third of them has an associated extra-cardiac anomaly or chromosomal aberration. Unfortunately, the overall prenatal detection of CHD worldwide not exceeds 50% and this could be explained by the facts that less than third of CHD had cardiomegaly and about third of them had near normal four-chamber view (4CV). Most of CHD occurred in cases without an identifiable prenatal risk.[1,2]

Many international scientific societies had published several guidelines to define the midtrimester sonographic planes for both basic (screening) routine scanning and additional ones for advanced (detailed) fetal echocardiography. The commonly adopted in practice are those published by the international society of ultrasound in obstetrics and gynecology (ISUOG) and the American institute of ultrasound in medicine (AIUM).[3,4]

Both ISUOG and AIUM agreed about the use of Yagel sweep for getting basic fetal heart views during routine second trimester anomaly scan. It encompasses 5 planes: situs, four chamber view (4CV), left ventricular outflow tract (LVOT), right ventricular outflow tract (RVOT), 3 vessels view (3VV) and 3vessles tracheal view (3VTV). The 20+2 plane approach provided by ISUOG for routine

mid-trimester fetal sonographic scanning hand included 4CV, LVOT, RVOT and 3VTV and named them plane: 7, 8, 9 and 10 respectively.[5]

The advancement in sonographic technology with high frequency probes with the use of harmonics and dynamic control had refined the resolution of ultrasound images and made the acquisition of fetal heart planes easies than before. Moreover, the presence of fetal cardiac pre-set in almost all current ultrasound machines enabled further refinement in fetal echocardiography by automatically decreasing the field of view (sector width) and the depth of penetration in order to increase the frame rate that will enhance the resolution and decrease the artifacts.[3,4]

Ultrasound companies had released various technical advancement tools specific to fetal echocardiography during the last decade. Examples of these tools are: STIC, V-CAD heart, 5D heart, tissue Doppler and HQ. Several studies had shown their benefits in recognizing the fetal cardiac structure and function but their limited availability due to high cost make their share to increase the detection rate of CHD worldwide is nil.[6]

The main aim of formal guidelines for fetal echocardiography that had released from various official societies in the last decade is to increase the detection rate of most cases with clinically significant CHD prenatally. Nevertheless, the detection rate of congenital heart defects remains problematic worldwide even in areas with good resources and expertise in fetal scanning. We think about the cause of this disappointing discrepancy and found two main reasons for that. First is the degraded acoustic window (DAW) caused by maternal obesity, abdominal surgery or fetal lie. This factor can be overcome in many cases by either maternal repositioning or rescheduling. The second cause in our opinion is guidelines-real life dissociation.[7,8]

METHODS

Our study was a retrospective cross sectional descriptive study. We have reviewed retrospectively cases of congenital heart diseases that were gathered by one tertiary center which is Habashy 4D scan (Alexandria, Egypt) from January 2020 till December 2023. Ethical approval was taken from the ethical committee of medical research, Faculty of Medicine, Alexandria University, Egypt.

We had two objectives; a) the main one was to establish an algorithm to review most of the CHD using a simple pattern recognition that will provide easy approach for their diagnosis; b) the secondary one was to define which are the specific ultrasound signs that we are going to include in the algorithm. All official societies provide guidelines for normal fetal cardiac sonographic view but not provide a standard protocol for diagnosis of anomalies in these views. To our best knowledge this is the first algorithm that gather the salient sonographic features of most CHD. We hope that our algorithm used as a supplement to guidelines in their update as this will facilitate the prenatal diagnosis of congenital heart disease

We had used the Yagel sweep in assessment of fetal heart as recommended by the ISUOG and the AIUM. Cases included in the study were scanned at 17 weeks of gestation onwards, singleton or twin. There was no restriction of enrolment according to maternal age, parity nor method of conception. Yagel sweep entailed assessment of the fetal heart at 5 planes; namely: situs, four chamber view (4CV), left ventricular outflow tract (LVOT), right ventricular outflow tract (RVOT) and three vessels tracheal view (3VTV).[3,4]

We used an algorithmic path to allow easy deferential diagnosis of CHD by using certain sonographic features (we call them: altered key points). We had studied 14 CHD that represent about 78% of the CHD (after exclusion of isolated VSD). Each 2 of them has a common altered key point during fetal cardiac scanning.

The total number of reviewed CHD cases was 66. Although the number of cases diagnosed with CHD during the study period was larger than that, we selected only cases that fulfil all the sonographic key points and salient features that we discuseds. Also we included cases that were confirmed with our diagnosis by either prenatal sonography at other centre or postnatal neonatal echocardiography.

RESULTS

Our 66 cases, there were 9 cases mitral atresia, 3 cases critical aortic stenosis, 4 cases tricuspid atresia, 3 cases critical pulmonary stenosis, 5 cases dextro-transposition of the great arteries (D-TGA), 3 cases double outlet right ventricle, 3 cases common arterial trunk, 14 cases complete atrioventricular septal defect, 3 cases Ebstein's anomaly, 4 cases interrupted inferior vena cava, 3 cases total anomalous pulmonary venous return, 3 cases right aortic arch and 5 cases aberrant right subclavian artery.

Table 1 shows demographic data of cases.

These 7 key points are: 1) right dominance: which mean that right ventricle larger than the left; 2) left dominance: which means that the left ventricle is larger than the right; 3) absent crossover of great arteries: which means that the great arteries are parallel instead of being at a right angle; 4) aorta override a ventricular septal defect (VSD): which means septoaortic discontinuity; 5) abnormal crux (fetal heart crux is formed by the meeting of atrioventricular valves with the ventricular septum and septum primum); 6) abnormal area behind the heart (ABTH): means behind the left atrium; 7) abnormal three vessels trachea view (3VTV). We found that most CHD has one of the following 7 key points. And each key point has 2 important causes. We tabulated the main features of the 7 key points of the 14 congenital heart diseases. Each altered key point has 2 main differential diagnosis.

Table 1: Demographic data of the studied cases.

Variables	Description
Gravidity	Range: 1-5; mean: 3
Parity	Range: 0-4; mean: 2
Method of conception	57 cases (86.4%): spontaneous. 9 cases (13.6%): ICSI
Number of fetuses	64 cases (97%): singleton. 2 cases (3%): twin
Cause of referral (risk factors)	44 cases (66.7%): routine midtrimester scan (no risk factors) 9 cases (13.6%): suspected CHD 7 cases (10.6%): thick NT in 1st trimester scan 6 cases (9.1%): previous child with CHD
Gestational age at the time of diagnosis	Range: 17-40 w, Mean: 28w

Table 2: Salient features of the 7 key points for diagnosis of most CHD (the 7x2 algorithm).

Altered key point	Salient features		Diagnosis
Rt Dominance	↓ Mitral flow	Globular LV (American football)	Severe aortic stenosis AS
	No mitral flow	Very small LV	Mitral atresia MA - HLHS
Lt dominance	TR	RV hypertrophy	Severe pulmonary stenosis PS
	No tricuspid flow	Very small RV	Tricuspid atresia TA - HRHS
Absent crossover (parallel outflows) Shut gun barrel sign	Great arteries related to each ventricle		D- transposition of the great arteries D-TGA
	Both great arteries originate from RV		Double outlet right ventricle DORV (TGA type)
Aorta override VSD	RVOT present		Tetralogy of fallot TOF
	RVOT absent (no pulmonary valve)		Common arterial trunk CAT
Abnormal crux	No septum primum		Complete atrioventricular septal defect c-AVSD
	↑ atrioventricular septum (apical displacement of TV)		Ebstein's anomaly EA
Abnormal Area behind the heart ABTH	Double barrel sign (large azyogos beside aorta behind LA)		IVC interruption
	↑ ABTH (behind the LA) Twig sign = confluent vein behind LA		Total anomalous pulmonary venous return TAPVR
Abnormal 3VT view	Trachea between Ao and Pul. a		Right aortic arch RAA
	Artery coursing behind the trachea		Aberrant right subclavian artery ARSA

Table 2 shows the salient features of the seven key points that we had used for diagnosis of most CHD.

First key point: right dominance

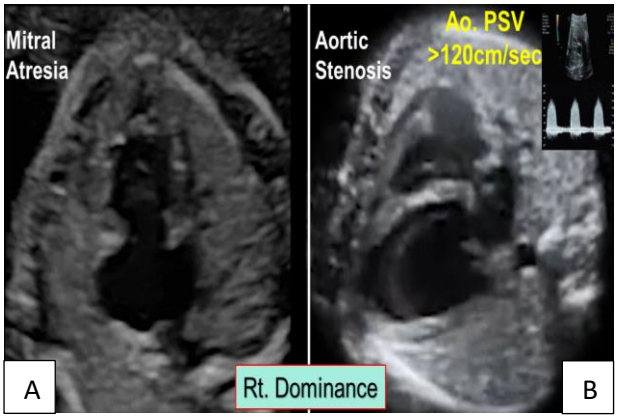

Figure 1 (A and B): Right dominance caused by mitral atresia and aortic stenosis.

Second key point: left dominance

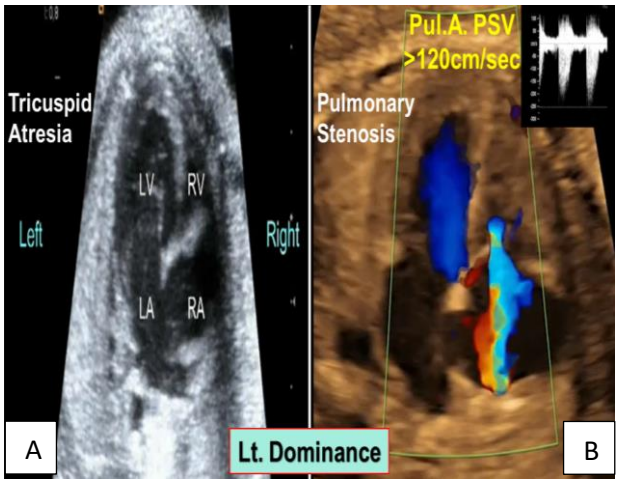

Figure 2 (A and B): Left dominance caused by tricuspid atresia and severe pulmonary stenosis.

Third key point: absent crossover of the great arteries

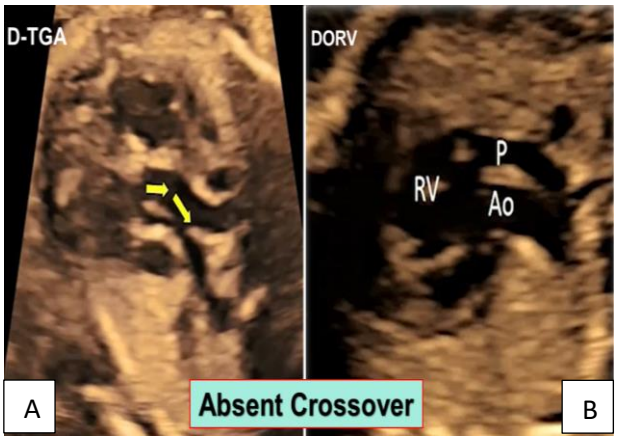

Figure 3 (A and B): Absent crossover caused by transposition of great arteries and double outlet right ventricle.

Fourth key point: aorta override a ventricular septal defect

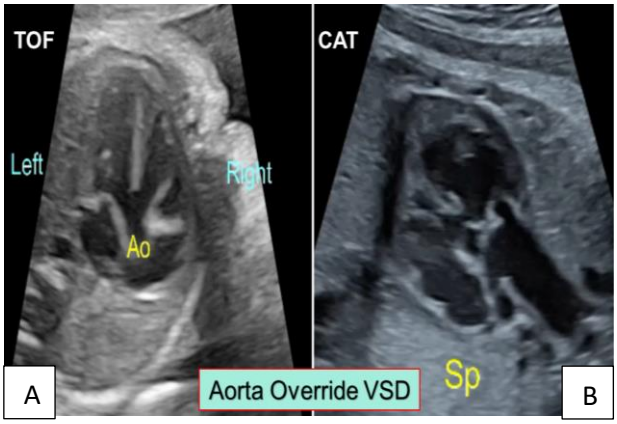

Figure 4 (A and B): Aorta override VSD caused by tetralogy of Fallot and common arterial trunk.

Fifth key point: abnormal crux

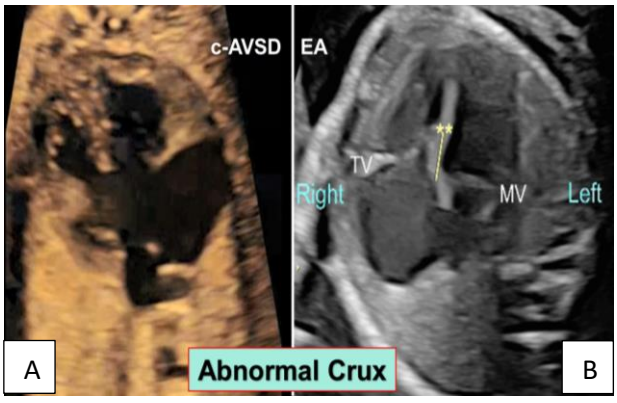

Figure 5 (A and B): Abnormal crux caused by atrioventricular septal defect and Ebstein's anomaly.

Sixth key point: abnormal area behind the heart (ABTH)

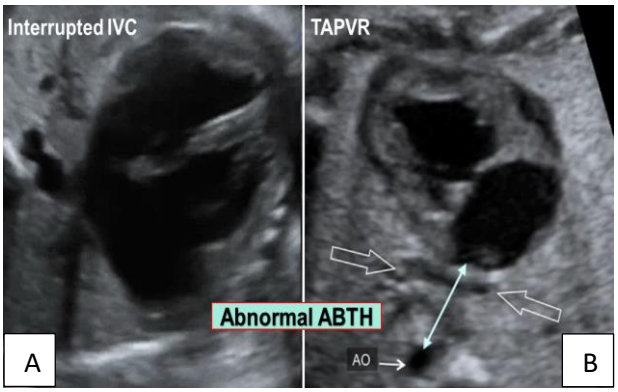

Figure 6 (A and B): Abnormal area behind the heart (ABTH) caused by IVC interruption and total anomalous pulmonary venous return.

Seventh key point: abnormal three vessels trachea (3VT) view

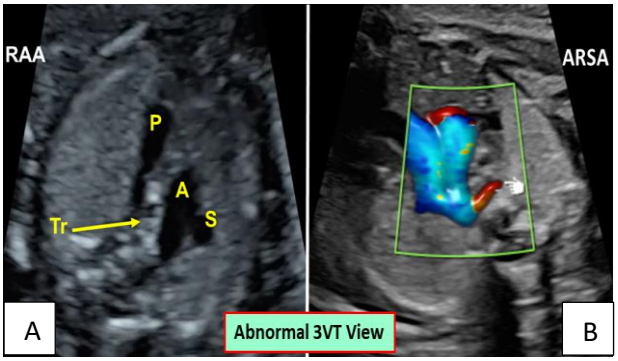

Figure 7 (A and B): Abnormal 3VTV caused by right aortic arch and aberrant right subclavian artery.

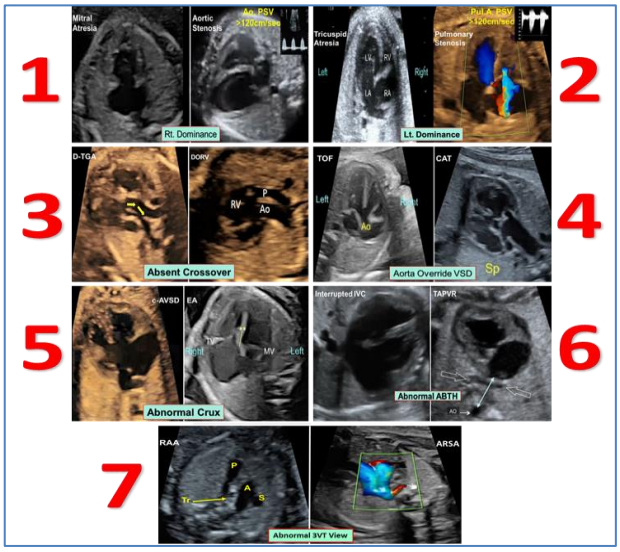

Figure 8: The 7×2 algorithm for diagnosis of 14 CHD. 7 altered key points, each has 2 important causes.

Table 3: Paladini et al algorithm for differential diagnosis of congenital heart diseases.[10]

Description		Diagnosis
CHD detectable on the four-chamber view	Right arteriomegaly with tricuspid insufficiency and normally located tricuspid valve	Tricuspid dysplasia
	Right arteriomegaly with apical displacement of the insertion of the tricuspid valve	Ebstein's anomaly
	Defect in the muscular interventricular septum	VSD
	Common atrioventricular valve	AVSD
	Left ventricular hypoplasia and mitral atresia	Hypoplastic left heart syndrome
	Right ventricular hypoplasia with VSD	Tricuspid atresia
	2 atrioventricular valves without interventricular septum	Double inlet single ventricle
	Ventricular disproportion with moderate prevalence of the right ventricle	Indirect sign for aortic coarctation
	Biventricular hypertrophy	Cardiomyopathy
	Tumors	Rhabdomyoma
CHD detectable on the left ventricular out flow tract	Narrow LVOT with endocardial fibroelastosis	Critical aortic stenosis
	Lack of crossover and each vessel connected to the contralateral ventricle	Transposition of great arteries
	Lack of crossover and both great vessels are connected with the right ventricle	Double-outlet right ventricle
	VSD in which the interventricular septum is normally aligned with anterior aortic wall	Simple outlet VSD
Right ventricular outflow tract assessment for differential diagnosis of malalignment VSD (aorta overrides the VSD)	Normal RVOT	Isolated malalignment VSD
	RVOT is reduced in size	Classic tetralogy of Fallot
	RVOT is absent (atretic)	Pulmonary atresia with VSD
	Pulmonary trunk and branches are severely dilated	Tetralogy of Fallot with functionally absent pulmonary valve
	Pulmonary artery arises from the aorta instead of the RV	Common arterial trunk

DISCUSSION

We had summarized our findings in Table 2 that can be used for easy mind-mapping for diagnosis for most CHD. Beside that table, we add a photo that summarized the salient features of each key point of the 14 CHD that we had addressed. These photos can be used as pattern recognition by eyeballing for most of CHD.

As regard to the right dominance; Axt-Fliedner et al described that cases with aortic stenosis have turbulent flow at the aortic valve with increased aortic blood velocity during spectral Doppler analysis.[9] Paladini et al described that cases with critical aortic stenosis have narrow LVOT with endocardial fibroelastosis. Their findings were comparable to our findings.[10] Simpson et al and Paladini et al described cases with hypoplastic left heart syndrome as having a slit-like left ventricle with no communication between the left atrium and the left ventricle (i.e. no mitral flow). Their description was similar to ours.[10,11]

As regard to the left dominance; Todros et al had enumerated criteria of severe pulmonary stenosis as the following: tricuspid regurgitation, right ventricular hypertrophy, post stenotic dilatation, marked increase in the flow velocity through the pulmonary valve and reversed flow in the ductus arteriosus. Our results were similar to their criteria.[12] Michael et al and Paladini et al had described the following findings in cases with tricuspid atresia: hypoplastic right ventricle with VSD and thick linear echodense membrane in place of the tricuspid valve (i.e. no tricuspid flow). Their findings were comparable to our results.[10,13]

As regard the absent great arteries' crossover; Bravo-Valenzuela et al had enumerated three findings for diagnosis of D-TGA which were: parallel outflow tracts, ventriculoarterial discordance, i.e. left ventricle give rise to the pulmonary artery that had early branching that was described as baby bird's beak sign and two vessels instead of three in the 3VTV that was described as I-shaped aorta or the boomerang sign.[14] Their findings were matched with our results. Gottschalk et al had described double outlet right ventricle by the presence of three criteria which were: parallel outflow tracts, both pulmonary artery and aorta originating from the morphological right ventricle and presence of a ventricular septal defect.[15] Our results matched with their findings.

As regards to the septoaortic discontinuity; Poon et al and Paladini et al had described cases of classic tetralogy of

Fallot as having two findings which were: the aorta arises astride an outlet VSD (i.e. aorta override a VSD) with reduced size of the right ventricular outflow tract.[10,16] Their description was similar to our results. Valope et al and Paladini et al had described cases of common arterial trunk as having malalignment VSD with single great artery arising from the base of the heart (i.e. RVOT is absent and the pulmonary artery arises from the aorta instead of the RVOT).[10,17] Our results were matched with their findings.

As regards to the crux abnormalities; Huggon et al and Paladini et al had described cases of atrioventricular septa defect as having common atrioventricular junction and central septal deficiency that is more apparent in diastole.[10,18] Their description was comparable to our findings. Zimmer et al had described cases with Ebstein's anomaly as having downward displacement of the tricuspid valve below the annulus that will lead to atrialization of the proximal part of the right ventricle with enlargement of the right atrium and small distal functional right ventricle.[19] It is associated with tricuspid regurgitation. Their description for criteria of Ebstein's anomaly was comparable to our results.

As regards the abnormalities of the area behind the heart (ABTH); Yoo et al had described cases of interrupted inferior vena cava as having dilated azygos vein beside the aorta both in the abdominal circumference plane and in the posterior mediastinum behind the heart at the four-chamber view.[20] Their description was matched with our findings. Mao et al had described sonographic features of fetuses with total anomalous pulmonary venous connection.[21] They found a wide separation between the left atrium and descending aorta (left atrium-descending aorta distance: LDD). Our results were comparable to their finding.

As regards the abnormalities of the 3VTV; Yoo et al had described the sonographic features of right aortic arch in which a sausage-shaped aortic arch is located to the right of trachea (instead of the left).[22] Their description was matched with our finding. Rembouskos et al had described the sonographic finding in cases with aberrant right subclavian artery as retrotracheal artery originating from the junction between aortic arch and ductus arteriosus and has a straight course toward the fetal right clavicle.[23] Their findings were comparable to our results.

We did a literature review and found that the most structured algorithm for prenatal differential diagnosis of congenital heart diseases was that described by Paladini et al.[10] Table 3 summarizes the algorithmic path that Paladini et al had used then we have compared his results with ours.

After exclusion of isolated VSD; Paladini et al had enrolled 15 CHD in his algorithm and our algorithm was entailed 14 CHD. There were 5 anomalies that we had listed in our algorithm for CHD that was not listed in Paladini et al algorithm; which are: pulmonary stenosis, inferior vena cava interruption, total anomalous pulmonary venous return, right aortic arch and aberrant right subclavian artery. There were 6 anomalies that Paladini et al algorithm had listed and were not listed in our algorithm; which are: double inlet single ventricle, aortic coarctation, cardiomyopathy, rhabdomyoma, pulmonary atresia with VSD, TOF with absent pulmonary valve.[10]

We think that our algorithm; i.e. the "7x2" algorithm for CHD has three advantages over Paladini's algorithm. First, instead of describing 15 anomaly using 3 cardiac planes to allocate them, we had described definite 7 altered key points for checking during fetal cardiac sonography and each altered key point has 2 differential diagnosis and put that in clear side by side photos. We called this way pattern recognition and we think it is better for mind-mapping especially for those who are beginners in the field of fetal echocardiography. Second advantage of our algorithm is that we added to the salient features some points to refine diagnosis. The last advantage of our algorithm is that we had estimated the proportion of CHD that could be diagnosed with our algorithm; which was about 78%, while Paladini et al did not mentioned the proportion of CHD that could be diagnosed with his algorithm.

We think that our algorithm has three limitations. First; it excludes ventricular septal defect. Second limitation is that it not covers all the congenital heart diseases. The last limitation is that not categorize congenital heart diseases according to the ductal dependence which is crucial in the prenatal counselling and the postnatal management and prognosis

CONCLUSION

The "7×2" CHD algorithm is a simple way that allow easy recognition of 14 fetal congenital heart defects by focusing on 7 altered key points that can be recognized on the common sonographic views where each of them has 2 main differential diagnosis. These 14 anomalies represent most of the CHD (78%) after exclusion of VSD. We hope that our algorithm used as a supplement to guidelines in their update as this will facilitate the prenatal diagnosis of congenital heart diseases.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: The study was approved by the Institutional Ethics Committee

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Cite this article as: Elhabashy A. Pattern recognition for prenatal sonographic diagnosis of most congenital heart disease: the "7×2" CHD algorithm. Int J Reprod Contracept Obstet Gynecol 2024;13:2262-8.