Prenatal Diagnosis by Fetal Echocardiography

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Congenital heart disease (CHD) is one of the most common congenital anomalies found at the rate of 4.3-8:1,000 live births and is a leading cause of neonatal and infant mortality.[1-3] Prenatal diagnosis of CHD has benefits on parental counseling, decision making during pregnancy, prenatal interventions, site and mode of delivery and postnatal management. Previous studies showed improvement of neonatal morbidity and mortality with prenatal diagnosis of certain CHD e.g. transposition of great arteries, hypoplastic left heart syndrome and coarctation of aorta.[4-6] Recent reports have shown successful outcomes of intrauterine therapy e.g. balloon dilation of severe aortic stenosis which can restore the left ventricular growth and biventricular circulation and prevent hypoplastic left heart syndrome.[7,8]

Despite the clear benefits of prenatal diagnosis, CHD is still the most commonly overlooked lesion during antenatal ultrasound evaluation with the detection rate of 4.5%-75% depending on the method and experience of examiners.[3,9-17] Combination of 4-chamber view (4CV) and outflow tracts examination has increased the detection rate substantially. The American Institute of Ultrasound in Medicine (AIUM) and International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) recommend this practice in routine fetal cardiac examination.[18,19] Some conditions, e.g. maternal diabetes, infections during pregnancy, maternal CHD, some medications, predispose the fetuses for CHD but fetal cardiac screening should be performed in a universal fashion because the majority of CHDs occur in low risk population.[20-23]

Currently, 2-dimensional (2D) ultrasound has been the gold standard for fetal cardiac examination. Viewing different planes of fetal cardiac structures in cross section and sagittal section can help the obstetricians to diagnose normalcy of fetal cardiac structures and various CHD. Those planes are demonstrated in Fig 1 (normal) and Fig 2 (common CHD).

Doppler ultrasound offers further informations about the direction, amount and velocity of fetal blood in the cardiovascular system and fetal cardiac rhythm by showing the relation of atrial and ventricular contraction. Many CHD can be confirmed using this technique at 3 planes i.e. 4CV, 5-chamber view (5CV) and 3-vessel trachea view (3VT) views. For example, tricuspid regurgitation which is one of the soft markers of Down syndrome can be demonstrated by observing Doppler gate volume overlap tricuspid valves at 4CV and 1:1 relationship of atrio-ventricular (A-V) contraction can be demonstrated by observing Doppler gate volume overlap mitral valves at 4CV or 5CV. (Fig 3, 4)

M-mode ultrasound has been used mainly for the diagnosis of fetal cardiac arrhythmias and fetal cardiac function assessment. Placing the M-mode line over the atrial and ventricular chambers, the signal of wall motion will be demonstrated and the relation of atrial and ventricular contraction can be evaluated. To assess the fetal cardiac function, the M-mode line should be placed perpendicular to the interventricular septum and both ventricular walls and diameters of each ventricular chamber during systole and diastole can be measured (Fig 5). Shortening fraction, a percentage difference between diastolic and systolic ventricular dimension, is a useful parameter for monitoring of fetal cardiac condition and obstetricians can provide proper managements accordingly.

3-Dimensional (3D) and 4-dimensional (4D) ultrasound are novel modalities that have gained more attention lately. Spatio-Temporal Image Correlation (STIC) is a new function in 3D/4D ultrasound which helps to collect volume data of a beating heart and analyze and render the pictures of fetal cardiac structures in various different planes. Using some special modes, e.g. surface mode, inversion mode, tomographic ultrasound imaging (TUI) mode, and so on, fetal cardiac examination is easy and clearer than ever before (Fig 6). Many reports showed comparable efficacy when compared to 2D ultrasound and obstetricians, with or without fetal echocardiography skills, can diagnose normalcy of fetal cardiac structures and various CHDs.[24-26] Fetal cardiac function assessment using inversion mode and specialized software so called VOCAL (Virtual Organ Computer-aided Analysis) is claimed to be more superior to 2D ultrasound owing to the ability to calculate virtual cardiac volume that is not a geometric form instead of using mathematical calculations used in 2D technique (Fig 7).[27] Virtual view of some structures can only be achieved by 3D/4D technique, e.g. viewing of interventricular septum or A-V valves (Fig 8).

CHD can be associated with other malformations or chromosomal abnormalities. Approximately 20% of

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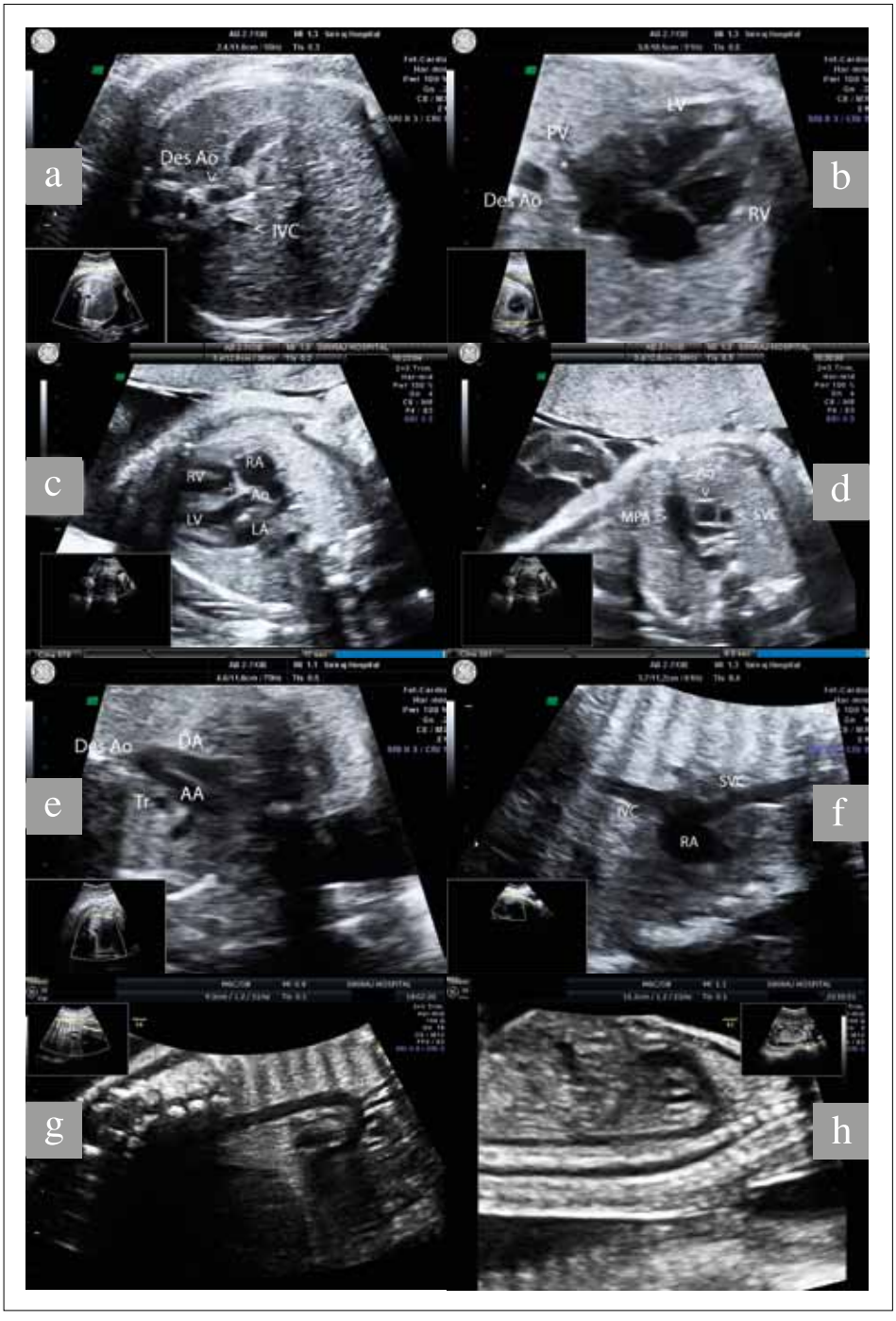

Fig 1. Different planes of normal fetal heart.
a = abdominal plane shows descending aorta on the left side of abdomen, inferior vena cava on the right side and slightly anterior to the aorta.
b = 4-chamber view (4CV) shows 2 equal atrial chambers and 2 equal ventricular chambers with intact interventricular septum. Two AV valves, 2 pulmonary veins and only one vessel behind the heart are seen.
c = 5-chamber view (5CV) shows aortic root, membranous interventricular septum (arrow) which is well aligned to the anterior wall of the aorta.
d = 3-vessel view (3VV) shows main pulmonary artery on the left side bifurcating into right and left pulmonary artery, the cross section of ascending aorta and superior vena cava on the far right. The size of MPA is slightly bigger than that of Ao.
e = 3-vessel and trachea view (3VT) shows ductus arteriosus and transverse aortic arch of comparable size joining together at descending aorta. Trachea is seen to the right of aortic arch.
f = caval view shows inferior vena cava and superior vena cava drain into the right atrium.
g = aortic arch view shows candy caine appearance of aortic arch with 3 neck vessels (arrow).
h = ductal arch view shows wider curve of ductal arch (hockey stick appearance) rising more anterior in the chest wall and without any branches.
(Des Ao=descending aorta, IVC=inferior vena cava, RV=right ventricle, LV=left ventricle, PV=pulmonary veins, RA=right atrium, LA=left atrium, MPA=main pulmonary artery, SVC=superior vena cava, DA=ductus arteriosus, AA=aortic arch, Tr=trachea)

Fig 2. Examples of common CHD.
a = Ventricular septal defect (VSD); the 4CV shows defect of muscular part of the interventricular septum (IVS).
b = Atrioventricular septal defect (AVSD); the 4CV shows absence of atrial septum primum, crux of the heart and upper part of interventricular septum (asterisk).
c = Overriding of the aorta diagnosed by 5-chamber view.
d = The 3VT view showed remarkable difference in the size of 2 vessel arches seen in hypoplastic left heart syndrome.

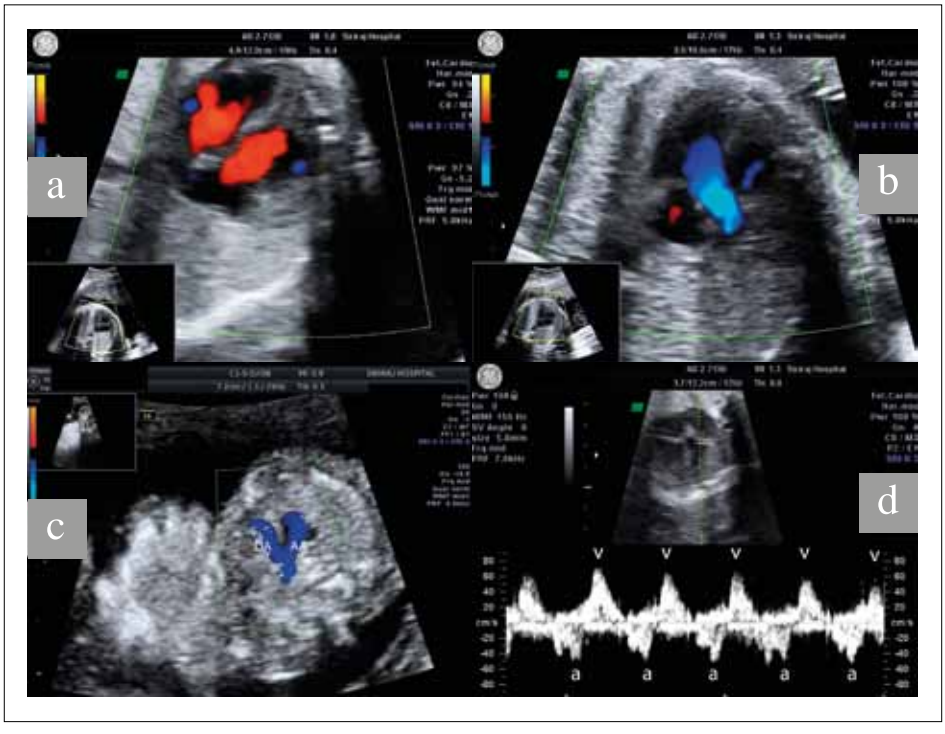

Fig 3. Normal Doppler ultrasound.
a = At 4CV, 2 strips of homogenous color flow of blood filling the ventricles from atriums are demonstrated.
b = At 5CV, homogeneous color flow of aortic root is seen without aliasing appearance.
c = At 3VT view, the same color seen in both vessels reflects the same direction of blood flow in the 2 arches in normal condition.
d = Pulsed wave Doppler ultrasound at 5CV showed 1:1 relation of inflow waveform (below the baseline) and outflow waveform (above the baseline). (a=atrial contraction, v=ventricular contraction)

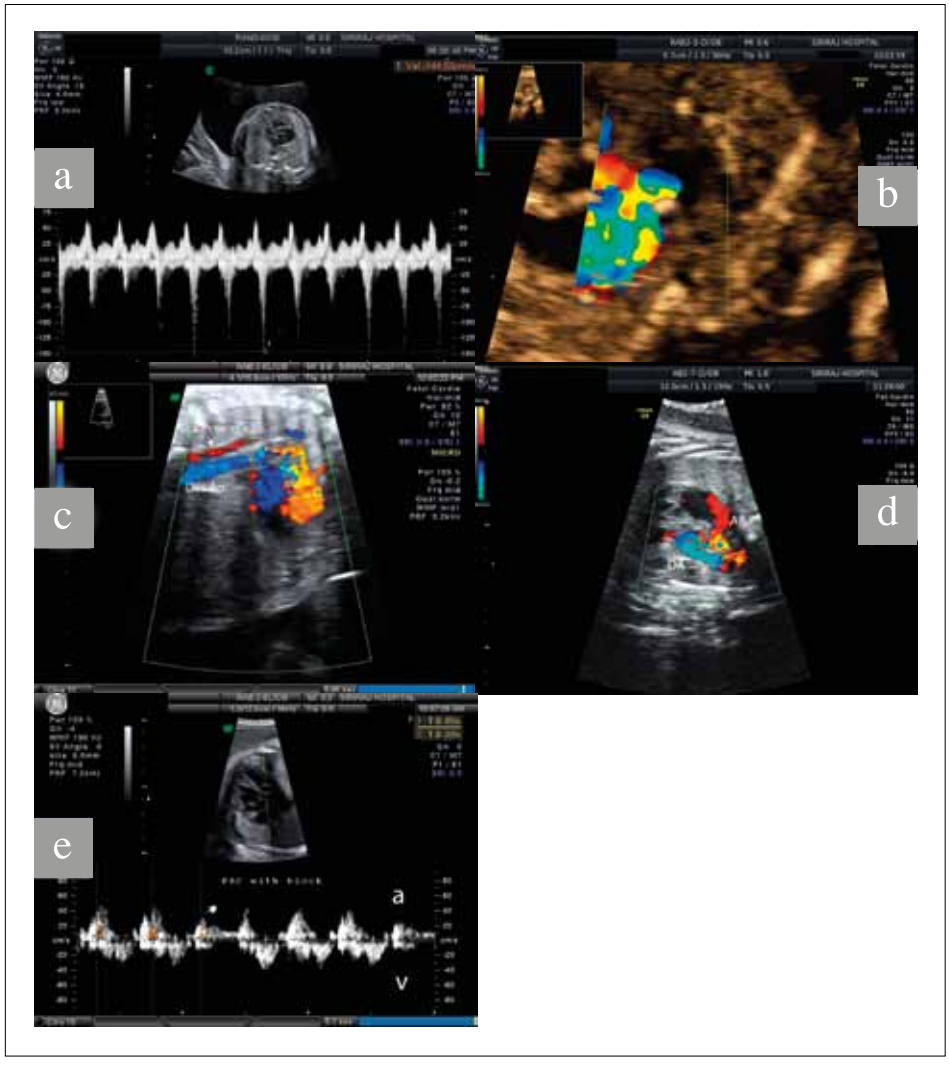

Fig 4. Examples of abnormal Doppler ultrasound findings.
a = Tricuspid regurgitation; pulsed wave Doppler ultrasound shows abnormally high velocity reversed flow (below the baseline) from the right ventricle back into the right atrium during systole.
b = Aliasing color flow in main pulmonary artery due to turbulent flow in pulmonary stenosis. Post-stenotic dilation is also seen.
c = Two vertical vessels in fetal thorax are seen by color Doppler ultrasound. The direction of blood flow in the descending aorta (blue color) was opposite to that of the other (azygos vein; red color). The diagnosis is left isomerism.
d = At 3VT view, the opposite direction of blood flow in 2 arches is seen resulting from hypoplastic left heart syndrome.
e = Pulsed wave Doppler ultrasound shows a premature atrial contraction without ventricular response.

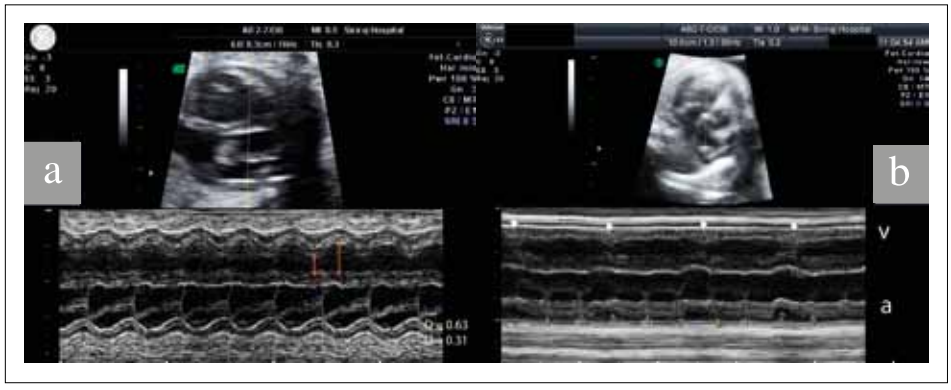

Fig 5. The use of M-mode ultrasound.
a = The maximal diameter of right ventricle at diastole (long orange line) and maximal diameter of right ventricle at systole (short orange line) are measured to calculate the shortening fraction of ventricles.
b = The M-mode line is placed over the right atrium and left ventricle and inconsistent relation between atrial and ventricular contraction is observed. The diagnosis is 3rd degree A-V block.

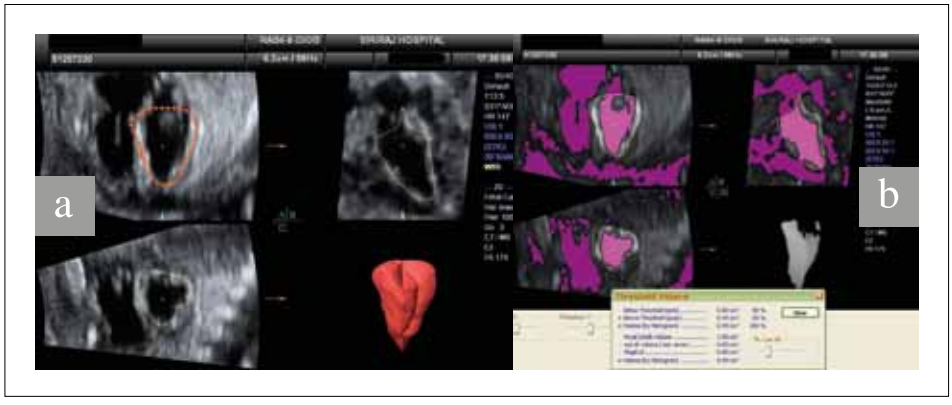

Fig 7. Assessment of fetal cardiac function by 3D ultrasound.
a = The use of VOCAL function illustrates the volume of the left ventricle in diastole.
b = The inversion mode shows the virtual volume of blood contained in the ventricular chamber (in pink) seperated from the total volume which included surrounding soft tissues. Stroke volume and cardiac output can be calculated by repeating the technique in systole.

the fetuses with CHD will have extra-cardiac anomalies while 10-20% of nonimmune hydropic fetuses will have CHD.[28] The frequency of chromosomal abnomalites in liveborns with CHD varies from 5-15% and is as high as 30-40% during the fetal period. This discrepancy is possibly a result of intrauterine fetal death of some fetuses with chromosomal abnormalities especially trisomy 18 and Turner syndrome.[29] Some forms of CHD are highly associated with chromosomal abnormalities, e.g. AVSD (68%), ASD (27%), VSD (18.2%), while some CHDs, e.g. conotruncal anomaly, do not increase the incidence. The incidence of CHD in fetuses with Downs syndrome has been reported to be 44-56% with AVSD being the most common (19.8%) followed by VSD (15.4%) and tetralogy of Fallot (1.8%).[30,31] With this association, many studies have suggested that ultrasound findings of the fetal heart could be used to increase sensitivity of detection and to adjust the risk of Downs syndrome e.g. right-to-left disproportion of the atrial and/or ventricular chambers (LR = 88.3), VSD (LR = 12.5), tricuspid regurgitation (LR = 5.9), and pericardial effusion (LR=10).[32] The risk of Down syndrome following a normal ultrasound study is decreased with LR of 0.11 when all the ultrasound markers including fetal cardiovascular markers are examined while the LR of

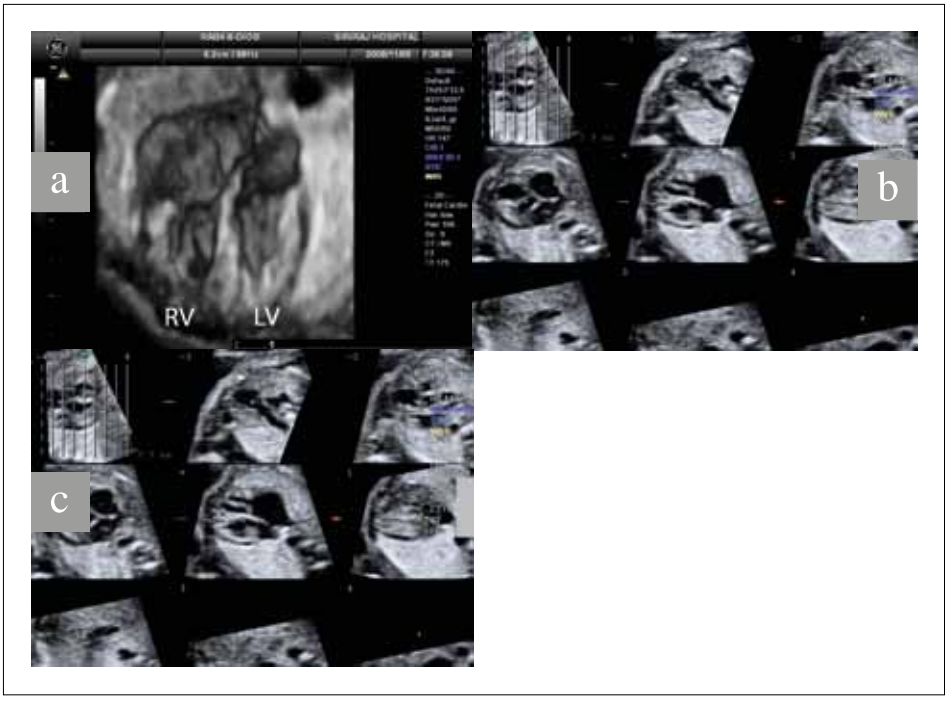

Fig 6. Examples of special modes of STIC.
a = The surface mode shows a 4-chamber view in 3D with different depth of intracardiac structures.
b = Nine different transverse planes of fetal cardiac structures are displayed by TUI. The distance between sections can be adjusted to display clear pictures of each plane of cardiac structures.
c = The outflow tracts are seen in criss-cross pattern by inversion mode which reverses the hypoechogenic appearance of fluid containing structures into hyperechogenic structures.

Fig 8. Virtual view of intracardiac structures.
a = Virtual view of A-V valves
b = Virtual view of intact interventricular septum
c = Virtual view of ventricular septal defects
(MV=mitral valve, TV=tricuspid valve)

0.42 is used to adjust the risk when only non-cardiovascular markers are evaluated.[32] This benefits in couselling the high risk women who refuse to have the invasive prenatal diagnosis.

In conclusion, prenatal diagnosis of CHDs enables better prenatal care and impoves pregnancy outcomes. The important challenges are low detection rate and insufficient skills of examiners. The continuous training and distribution of properly-equipped instruments along with universal screening in both high risk and low risk populations might yield a better future of pregnancy and child health.

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