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SYSTEMIC REVIEW

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Prenatal detection of congenital heart disease with fetal echocardiography in Saudi Arabia: Systemic Review

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Abstract

Objectives: *To evaluate the clinical value of color doppler echocardiography for the diagnosis of fetal congenital heart disease (CHD) included in routine obstetric care which provides relevant information for decision making, detailed anatomic survey performed at skilled, high-volume obstetric centers, and to investigate the technical limitations of fetal echocardiography in this patient population.*

Methods: *This was a retrospective descriptive review of fetal echocardiograms performed at Children's Hospital Riyadh from (2010-2017). All women referred during the second trimester for fetal echocardiography because of maternal diabetes were included. Those with severe heart disease suspected on obstetric ultrasound examination were excluded.*

Results: *There were 313 initial fetal echocardiograms. Forty three patients were diagnosed with multiple heart diseases, 32 patients were diagnosed with suspected mild heart disease (such as small ventricular septal defect), of which 8 patients had normal follow up study. Of 150 patients who had normal follow-up fetal evaluation, 33 patients had severe CHD, 2 patients had IDDM (diabetes). Most of these pregnancies did not have a postnatal cardiac evaluation. Forty-seven fetuses had benign cardiac findings. Fifty patients were asked to return for at least one follow-up visit, most due to the inability to complete the exam at the initial visit.*

Conclusions: *In an environment with access to high-volume, skilled comprehensive ultrasound services, fetal echocardiography by a pediatric cardiology program adds little to the care of women with diabetes and high risk women. Poor acoustic windows may indicate frequent multiple visits.*

Keywords: *ultrasound, IDDM, fetal echo, congenital herat disease*

Introduction

Congenital heart disease (CHD) occurs in approximately 0.8% of all live-born humans [1]. The causes are multifactorial and include genetic as well as environmental (in utero) factors. Maternal diabetes mellitus (MDM) is known to increase the likelihood of fetal CHD, with an estimated increased risk of up to 8.5% of live births [2]. Most types of cardiac structural lesions have been associated with MDM, ranging from small septal defects to duct-dependent heart disease [3-6]. As a separate concern, there is an increased risk for the development of hypertrophic cardiomyopathy in the third trimester in this population.

The purpose of prenatal cardiac screening is to identify those patients with defects that may require further evaluation and treatment, and to provide appropriate counseling to the family in a timely manner. In many cities, high-volume tertiary care centers treat the majority of pregnancies complicated by MDM. In some centers, it has also become standard practice to screen all pregnancies complicated by MDM with fetal echocardiography performed at a pediatric cardiology center. Though fetal echocardiography is mostly reserved for high risk pregnant women, its role as a routine prenatal screening tool still needs to be defined

We undertook this study in an effort to learn how often members of this population successfully undergo anatomic survey by the fetal cardiology group at our center and how often our evaluation changed the care of the pregnancy.

Methods

This was a retrospective study of all fetal echocardiograms, performed at the Children's Hospital /King Saud Medical City, This study enrolled consecutive pregnant women who attended antenatal clinic between 2010 and 2015 in a tertiary care hospital. These pregnant women were categorized into two groups: high risk group included pregnant women with traditional risk factors for CHD as laid down by Pediatric Council of the American Society of Echocardiography and low risk group. Detailed fetal 2 D echocardiography was doneour primary analysis, as we were only interested in assessing the utility of 'routine mid-trimester echocardiographic screening' in this population. Patients who had more than one pregnancy during this period were counted as separate studies, as were twin pregnancies.

All fetal echocardiograms during this time were carried out using either a Siemens Acuson 128X echocardiographic system, equipped with curvilinear fetal probes. All studies were performed according to a standard fetal echocardiographic protocol, which included: biometric data, four-chamber and outflow views, short-axis sweeps of the heart and great vessels, long-axis view including the aortic arch and Doppler flow interrogation of ventricular inflow and outflow Figure 1, foramen ovale and ductus arteriosus, pulmonary veins, umbilical vessels, ductus venosus and middle cerebral artery.

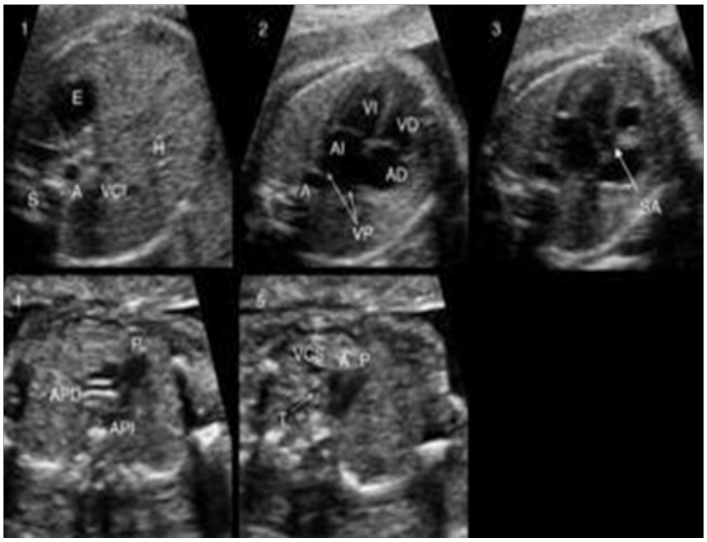

Figure 1. *Four-chamber view at a non-optimal magnification (a) and after application of zoom function and magnification of the image (b) Details are better visualized on the magnified image.*

Initial fetal echocardiogram findings	Total fetal studies (n) 156	Outcome of repeat study(ies)	History of IDDM	In insulin	Postnatal follow-up	Other risk factors
Cardiomyopathy	11fetus	Cardiomyopathy	+ IDDM	YES	No follow up	-
Mild VSD,ASD	32 fetus	VSD,ASD	+ IDDM(5)	DIET	Small VD, ASD in 21 patients	-
AV canal	22 fetus	-	No	No	8 follow up +AV canal	Down syndrome [5]
Tricuspid RE	8 fetus	-	No	No	No follow up	-
Aortic arch	18	-	No	No	No	-
Multiple conginetal anomalies	43	Single ventricels, hypoplastic	No	No	Single ventriceles [12] Hypoplastic with sever pulmonary stenosis [5]	+ve history of CHD
Complex heart	22	Complex heart, ASD	No	No	Complex H Disease (18 follow up)	Cleft palate in 1

Table 1. *Prevalence and rate of prenatal ultrasound diagnosis of major and minor congenital heart defects.*

Maternal indications
Family history of congenital heart disease Metabolic disorders (eg., diabetes, PKU) Exposure to teratogens Exposure to prostaglandin synthetase inhibitors (eg., ibuprofen, salicylic acid, indomethacin) Rubella infection Autoimmune disease (eg., SLE, Sjogren's) Familial inherited disorders (Ellis van Creveld, Marfan, Noonan's, etc) In Vitro fertilization
Fetal Indications
Abnormal obstetrical ultrasound scan Extracardiac abnormality Chromosomal abnormality Arrhythmia Hydrops Increased first trimester nuchal translucency Multiple gestation and suspicion of twin-twin transfusion syndrome

Table 2. *Indications for foetal echocardiography as laid down by Pediatric Council of the American Society of Echocardiography*

Cardiac defects were divided into three categories based on the echocardiographic diagnosis. Those that required significant counseling with the family, surgical intervention postnatally or mandatory cardiac follow-up postnatally were categorized as 'severe' CHD. Examples include common atrioventricular canal defects or duct-dependent lesions. Defects that were not likely to be hemodynamically significant but might require postnatal follow-up were categorized as 'mild' CHD, for example, small ventricular septal defect or minor valve abnormalities.

Data including maternal age, gestational age and fetal echocardiographic diagnosis were recorded. We reviewed the perceived technical quality of the images, based on comments in the fetal report, and the reasons documented for difficult imaging. We also reviewed the number, if any, of repeat fetal echocardiograms performed for each patient, whether the repeat studies were performed due to technically inadequate initial studies whether or additional clinically useful information. Postnatal echocardiogram results were reviewed, if available. In the design of the study, maternal records were to be reviewed for information about glycemic control for those fetuses with prenatally diagnosed severe CHD. As this was a descriptive study, no formal statistics were performed.

Results

In our study we review (retrospective cohort review) to all mother had been refer to do fetal echo either with high risk or low risk as screening according to the American society of Heart (Table 2) review was done for all these patients, The ultrasound had been repeated in 56 patients (17.3%).

In our review 43 (13.7%) patients had multiple congenital anomalies like (hypoplastic heart, sever pulmonary stenosis, single ventricles) and in 32 fetus they had mild congenital heart disease like VSD and ASD) repeated echo study done in 21(6%) patients did showed mild VSD, ASD, the rest had no show after delivery.

Interestingly 22(7%) fetus diagnosed with complex heart disease, and follow up done for 9 of them, 5 cases referred to higher center before delivery so, we lost their follow up, and the rest no clear information in the chart. There were 5 cases of chromosomal abnormalities (prevalence, 0.63%) and one patient had cleft palate. We were able to perform an extended basic fetal echocardiogram on (200)6% of the pregnant women There were 56 cases of congenital heart defects, with a prevalence of 0.2% , of which 9% (18 cases) were major heart defects (prevalence,5%). The most frequent major congenital heart defect was VSD larger than 3mm (19 cases) and ASD 13 patients in 5 of those patients IDDM was diagnosed, followed by endocardial cushion defect (eight cases). In 11 patients cardiomyopathy was suspected but no follow for those patients was mention interestingly all the women were diagnosed with IDDM (on insulin), we do not know is that related to diabetic history but no further follow up to confirm it.

In sever congenital heart disease no discussion for termination of pregnancy. The rest of patients (111, 35 %) there ECHO study was normal. The fetal Echo in 157 patients was normal study, and there were no follow up.

In our review both high risk mother and low risk mother had positive risk of CHD detection. The detection of CHD prenatally was 49.8 % positive in comparison to the normal fetal echo study with is really high and significant.

Discussion

Studies investigating the clinical impact of prenatal diagnosis on the outcome for individuals with CHD are conflicting [7-10]. In general, prenatal diagnosis provides the opportunity for counseling of the family, anticipatory care and discussions involving potential surgical intervention.

Detailed fetal anatomic surveys in the early second trimester are common practice and typically include examination of both four-chamber and outflow tract views of the fetal heart. Yet, although the use of these views has been shown to provide effective screening for cardiac defects [11-14], many centers continue to recommend referral for a focused cardiac prenatal evaluation. Our study echocardiograms performed in 313 patients, however, found that no patient was identified with previously undiagnosed CHD that necessitated a change in prenatal care or delivery plans. In addition, there were no patients with severe CHD.

We detected 32 (10.2%) patients with suspected mild CHD. None of these had suspected cardiac findings on the obstetric referral, and thus they represent cardiac defects that would otherwise have gone undetected in utero. The majority of these were small VSDs and mild aortic valve abnormalities with normal valve function. The sensitivity and specificity of identifying small VSDs on fetal echo is limited given fetal physiology and the anatomy of the ventricular septum [3].These infants would be expected to have relatively benign perinatal courses. If such defects are still present postpartum, they are usually detected by a routine physical exam within the first weeks to months of postnatal life, at which time the patient would be brought to the attention of a pediatric cardiologist. Defects not detected prenatally (false negatives) were not considered clinical errors in a prior study [15]. Some of the possible small defects in our group may have represented false positives or defects that would go on to close in utero.

In contrast to VSDs, postnatal detection of mild aortic valve disease by physical exam can be difficult and the lesion may go undetected for years or decades if there is no significant valvar stenosis or regurgitation. However, if valve function is normal, no treatment is necessary for a mild anatomical abnormality of the aortic valve, and so there is little clinical advantage in being aware of this diagnosis prior to birth. Because none of our patients with suspected mild valve abnormality had a postnatal echo at our institution, we do not know if any represented false positives. Of the remaining five patients with suspected mild disease, four were ultimately found to have normal cardiac findings either on a repeat prenatal study or on postnatal echocardiography.

A significant proportion (42.0%) of the initial studies performed were incomplete. In the majority of cases this was due to poor acoustic window, often attributed to maternal body habitus. Despite diagnostic improvement with the technological advancement of modern ultrasound machines, maternal BMI has been correlated with rates of suboptimal visualization of cardiac structures at second-trimester screening [12]. In our population, there was a high prevalence of poor acoustic windows which necessitated additional studies for many women. This can become a logistical burden for both the patient and the diagnostic center while providing little benefit.

The utility of routine fetal echocardiographic screening in diabetic patients has been considered in several studies. Muller et al. [13] reviewed routine fetal echocardiographic screening in all high-risk patients (including those with MDM) with normal

obstetric four-chamber and outflow tract views, and no cases were found among patients with isolated pre-existing diabetes mellitus. Our study, which evaluated all diabetic patients, both those with pre-existing and those with gestational diabetes, corroborates their conclusion. However when foetal echocardiography which involves a detailed, focused assessment of the foetal cardiovascular system is performed, the rate of detection of CHDs exceeds 85% to 90% [16].

The lack of comprehensive follow-up postnatal information was a limitation of this study. we did not have detailed information regarding the type of diabetes, level of glucose control or duration of illness in our patients. Despite this, the absence of significant positive cardiac studies is an important finding. Again the post natal follow up echocardiography of all the cases could not be done in our study which would have provided us with more critical information regarding sensitivity and specificity of fetal echocardiography and also about the outcome of the pregnancies

We may also have under-reported the number of incomplete studies as our data were ascertained from echo reports rather than from raw information. If the echocardiographer chose not to document these details on the report, then it could not be considered in our study. Finally, though not a limitation, it is important to reiterate that our cohort cannot be used to estimate the risk of CHD in the population of women with MDM.

This early detection of CHD in early pregnancy given adequate time for physician to discuss severity, and prognosis of the disease with the family, where they can still make a decision regarding the course of pregnancy. These cases are those which otherwise would have been missed on antenatal screening, or would have been diagnosed at late pregnancy where decision making would have been more difficult. While theoretical concerns exist regarding the harmful effects of ultrasound energy on the developing foetus, no confirmed effect has been detected till date [17].

Conclusion

Our study shows no difference in incidence of CHDs between pregnancies associated with high risk factors compared to low risk pregnancies. So we advocate that fetal echocardiography should be included as a part of routine antenatal screening.

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