Abstract

Objective: We present a case of diffuse endocardial fibroelastosis diagnosed at 22 weeks of gestation. The classification and clinical approach to fetal cardiomyopathies based on this syndrome will also be discussed.

Case: A healthy 22 year old primigravid woman had an uneventful pregnancy until the ultrasonographic examination at 22 weeks of gestation. Fetal echocardiographic evaluation revealed a dilated and hypotonic left ventricle with diffuse hyperechogenic endometrial lining. Left ventricular endocardium was thickened with accumulation of layers of collagen and elastin. Pathological confirmation of the diagnosis could not be made because parents refused autopsy examination.

Conclusion: Endocardial fibroelastosis is a rare disease and sporadically diagnosed during antenatal period. Sonographic criteria include a dilated left ventricle with poor contractility and hyperechogenic bright thickening of endocardial surface. When diagnosed, it should be wise to terminate the affected pregnancy since the prognosis is poor if it’s detected before fetus is viable.

Keywords: Endocardial fibroelastosis; cardiac; pregnancy termination.

Isolated Fetal Endocardial Fibroelastosis Diagnosed and Terminated at 22 Weeks of Gestation: A Case Report

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Yirmiikinci gebelik haftasında tanı konan ve termine edilen nadir bir izole endokardiyal fibroelastoz vakası: Bir olgu sunumu


Sonuç: Endokardiyal fibroelastoz antenatal dönemde sporadik olarak tanı konulan nadir bir hastalıkdır. Sonografik kriterler kötü kontraktülitel olursa ventriküler ve hipereköjen paralak kalınlaşmış endokardiyal yüzeyi içerir. Tanı konduğunda, hastalığın prognozu kötü olduğundan dolayı, fetüs viabilite kazanmadan etkilenmiş gebelikin termine edilmesi akılca olacaktır.

Anahtar Sözcükler: Endokardiyal fibroelastozis; kardiyak; gebelik terminasyonu.
Introduction

Endocardial fibroelastosis (EFE) is a rare cardiac disorder characterized by diffuse proliferation of elastin and collagen fibers within the endocardium which mainly affects the left ventricle. This mainly leads to decreased compliance and stroke volume. It has been classified as primary and secondary forms according to whether a structural cardiac anomaly is present such as aortic stenosis, coarctation or anomalies at the origin of left coronary artery or pulmonary trunk. In the absence of these anomalies, it's described as primary disease. But most authors consider EFE as a secondary reactive process set off in the endocardium by stress on the myocardium. We present a case of diffuse endocardial fibroelastosis diagnosed at 22 weeks of gestation. The classification and clinical approach to fetal cardiomyopathies based on this syndrome will also be discussed.

Case

A healthy 22 year old primigravid woman had an uneventful pregnancy until the ultrasonographic examination at 22 weeks of gestation. Her past obstetrical and gynecologic history was unremarkable. Fetal echocardiographic evaluation revealed a dilated and hypotonic left ventricle with diffuse hyperechogenic endocardial lining (Figure 1,2). Ultrasonographic examination of the fetus revealed normal aortic and mitral valve diameters and decreased aortic peak systolic velocity and mitral blood flow measurements were determined by doppler ultrasonography investigation. No associated cardiac or systemic anomalies were found on sonography. Parvovirus, coxackievirus infections and genetic or metabolic disorders were excluded. A presumptive diagnosis of EFE was made. Parents were told about the condition of the fetus and they elected to terminate the pregnancy. A male fetus compatible with 22 weeks of gestation with no other dysmorphic features was submitted to autopsy examination but parents refused it. Any other major abnormalities of the fetus was not found at postpartum examination.

Discussion

Cardiomyopathies (CM), account for 8% to 11% of the cardiovascular diagnoses detected prenatally. CM is diagnosed in 3% of newborns with cardiovascular disease. Single gen disorders (Noonan syndrome, metabolic disorders familial CM, congenital myotonic dystrophy, X-linked myotubular myopathy), mitochondrial disorders, chromosome abnormalities and α
thalassemia are intrinsic and familial causes of primary CM with recurrence risk. Extrinsic causes of primary CM necessitates the investigation of fetal myocardial dysfunction, maternal hematologic indices and serological workup, amniocentesis if needed and invasive fetal sampling to assess for anemia, thrombocytopenia, high specific IgM titers, viral cultures, and polymerase chain reaction for the specific infectious agent.6,7

Secondary CM includes cardiac causes, high output states, altered ventricular filling and altered ventricular afterload disorders. Diastolic dysfunction is associated with the greatest risk of mortality. Left and right ventricular end-diastolic diameters and wall thickness can be measured with M-mode tracings or 2-dimensional images (8,9). In normal fetuses semilunar and atrioventricular valve peak flow velocities gradually increases during pregnancy. Mitral and tricuspid valves have greater peak A velocity values than peak E velocity values throughout pregnancy. Diastolic dysfunction is considered when at least two of the following parameters are identified: Abnormal E/A ratio through mitral or tricuspid valve inflow (<2 SD below the mean for gestational age), increased duration of isovolumic relaxation time IVRT (>2 SD above the mean for gestational age), increased a-wave reversal in the inferior vena cava or hepatic vein (>20 cm/s) or a biphasic rather than triphasic flow pattern, and the presence of umbilical venous pulsations. Fetal echocardiography with a general fetal anatomic ultrasonographic scan and maternal laboratory investigations to establish the pathogenesis and exclude potentially treatable conditions should be evaluated during fetal CM investigation.10

The Tei index is a useful, new, noninvasive Doppler index of combined systolic and diastolic function calculated IVRT plus isovolumic contraction time (IVCT) divided by ejection time (ET). The Tei index readily provides early detection of diminished myocardial function, particularly ventricular dysfunction.11

Endocardial fibroelastosis is a rare disorder of newborns accounting for no more than 1-4% of total congenital heart diseases.5,12 Around 80% of patients present with congestive heart failure within the first year of life.13 Also one third of patients with clinically diagnosed EFE dies of congenital heart failure during the first 2 years of life. Late deaths occur in the group of patients with clinically resolved EFE.4

Classically, it has been classified into primary and secondary forms according to whether a structural cardiac anomaly is present since it was first proposed in 1960's. Recently it has been proposed that this was a nonspecific response to many stressors of myocardium such as congenital malformations of vessels and valves, viral agents affecting myocardium e.g. parvovirus, coxackievirus, or genetic disorders, mitochondrial cardiomyopathies and metabolic disorders. According to this thinking, diffuse intimal fibroelastic thickening of muscular arteries in response to chronic hypertension shares the same mechanism with EFE. When the heart is thought as a kind of modified vascular artery, its response to chronic stress will be the endocardial thickening which corresponds to intima of vessels.14 Endocardial smooth muscle cells which are normally few in number are seen to proliferate, transform into fibroblasts and produce both collagen and elastin under myocardial stress.

This fibroelastic reaction seems to occur during fetal development and growth, continuing after birth and throughout early infancy. The reason why this occurs more frequently in that life period is because of greater growth potential of cells at this period. Intestinal hyperechogenisities were also proposed as response to various fetal insults such as infection, hypoxia, vascular disease supporting the 'response to stress' theory.15

During the fetal development of EFE, echocardiographic appearance initially demonstrates left ventricular dilatation and hypococontractility with hyperechogenic thickening of endocardial surface as seen in this case. As the gestational age advances, the left ventricular cavity decreases in size, there's a progressive left ventricular wall hypertrophy and an increase in the hyperechogenicity of endocardial surface.16
A direct association between the thickness of endocardium and time of onset of myocardial stress was proposed.

Sonographic criteria include a dilated left ventricle with poor contractility and hyperechogenic bright thickening of endocardial surface. According to this, presented case fulfills the both criteria. In differential diagnosis, causes of intracardiac echogenic focus should be included, most importantly Trisomy 21 and 13 which usually have other morphologic abnormalities.

Since there are controversies related to causes and majority of cases are sporadic as the presented case, a risk population to screen has not been proposed. In the literature, cases reported so far were diagnosed in the second and third trimester, the earliest one being diagnosed at 14 weeks of gestation. Time of diagnosis may be related to the severity of insult and the response of insulted tissue besides the duration of insult meaning under a severe stress, reaction of the tissue can be more prominent leading to early diagnosis.

**Conclusion**

EFE is a rare disease which sporadically diagnosed during antenatal period. Serial ultrasonographic evaluation is needed since it’s a progressive condition which has a spectrum of findings. It should always be remembered that EFE is a response to a disease state rather than being a disease itself. Presence of hypococontractility and hyperechogenic endocardium necessitates excluding fetal echocardiography and congenital heart diseases. Since it’s a progressive lesion, it has aspectum of endocardial changes ranging from microscopic thickening to gross changes. When performing screening sonography, this should be kept in mind and special attention should be given to heart even if the previous scan is normal. When diagnosed, it should be wise to terminate the affected pregnancy since the prognosis is poor if it’s detected before fetus is viable. If not, remaining antenatal period should be under the control of a pediatric cardiologist and an experienced perinatologist.

**References**