CASE REPORT

Mitral and Aortic Atresia, Ductus Arteriosus Aneurysm, and Aortic Tortuosity: First- and Second-trimester HDlive Flow Features

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ABSTRACT

We present the first- and second-trimester HDlive Flow features of mitral and aortic atresia, ductus arteriosus aneurysm, and aortic tortuosity and kinking. Retrograde aortic flow was evident, and aortic tortuosity and kinking were suspected using HDlive Flow with spatiotemporal image correlation (STIC) at 13 weeks and 6 days of gestation. Mitral and aortic atresia and tricuspid regurgitation were recognized by conventional fetal echocardiography at 20 weeks of gestation. HDlive Flow clearly showed retrograde aortic flow, ductus arteriosus aneurysm, and helical aortic tortuosity and aortic kinking. Congestive heart failure (pericardial effusion, pleural effusion, and ascites) was also evident. Termination of pregnancy was conducted due to the parent's desire to discontinue the pregnancy. There were no apparent phenotypic features of connective-tissue disorder. The parents refused further work-up such as autopsy, chromosomal analysis, and genetic studies of the baby. To the best of our knowledge, this is the first report on aortic tortuosity and kinking of a fetus in the first trimester of pregnancy using HDlive Flow with STIC.

Keywords: Aortic tortuosity, Ductus arteriosus aneurysm, HDlive Flow, Mitral and aortic atresia, Prenatal diagnosis, STIC.

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INTRODUCTION

There have been only four reports on antenatal diagnosis of fetal aortic tortuosity.¹⁻⁴ These diagnoses were made after 20 weeks of gestation. Two cases were suspected as aortic tortuosity syndrome,^{1,2} and the other two were isolated aortic tortuosity cases.^{3,4} One of the isolated cases was associated with a ductus arteriosus aneurysm (DAA).⁴ To the best of our knowledge, there has been no report on antenatal diagnosis of fetal aortic tortuosity with DAA in the first trimester of pregnancy.

HDlive Flow with spatiotemporal image correlation (STIC) provided useful and additional information for the assessment of normal fetal cardiac anatomy and prenatal diagnosis of congenital heart disease (CHD),⁵⁻⁷ especially for the first- and early second-trimester diagnosis of fetal CHD.^{8,9} In this study, we present the first- and second-trimester HDlive Flow features of mitral and aortic atresia, DAA, and aortic tortuosity and kinking.

CASE DESCRIPTION

A 37-year-old pregnant Japanese woman, G (1), P (0), received routine obstetrical examination at 13 weeks and 6 days of gestation. Fetal biometry was consistent with the corresponding gestational age. However, retrograde aortic flow was evident with transabdominal STIC (Fig. 1), and aortic tortuosity and kinking were suspected using HDlive Flow with STIC (Fig. 2).

The second-trimester screening demonstrated mitral and aortic atresia and tricuspid regurgitation using conventional fetal echocardiography at 20 weeks of gestation (Fig. 3). Moreover, wide oval thoracic aorta was noted. HDlive Flow clearly showed retrograde aortic flow, DAA, and helical aortic tortuosity and aortic kinking (Fig. 4). Congestive heart failure (pericardial effusion, pleural effusion, and ascites) was evident. Moreover, a single umbilical

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artery was noted. However, fetal biometry was consistent with the corresponding gestational age.

The patient and her husband desired to discontinue the pregnancy, and abortion was induced at 21 weeks and 4 days of pregnancy, resulting in a male abortus weighing 387 gm, with a height of 24.7 cm. The parents refused further work-up such as autopsy, chromosomal analysis, and genetic studies of the baby.

DISCUSSION

Arterial tortuosity is considered to be caused by genetic arteriopathies, and the main affected artery is the aorta.¹⁰ The exact

Fig. 1: Multiplanar view with spatiotemporal image correlation of fetal aortic tortuosity (small arrow) and kinking (large arrow) at 13 weeks and 6 days of gestation (Ao, aorta; DAo, descending aorta; PA, pulmonary artery; RV, right ventricle)

cause and timing of arterial tortuosity are unclear. There have been only four reports on fetal aortic tortuosity, but all patients were diagnosed after 20 weeks of gestation.¹⁻⁴ In the present study, fetal aortic tortuosity was suspected in the first trimester of pregnancy, and was confirmed at 20 weeks of gestation. To the best of our knowledge, this is the first report on antenatal diagnosis of fetal aortic tortuosity. Therefore, aortic tortuosity may occur early in fetal life. Further studies are needed to confirm the exact timing of onset of fetal aortic tortuosity.

Karmegaraj et al.⁴ reported the association of fetal aortic tortuosity with DAA at 23 weeks of gestation. In their report, HDlive Flow clearly showed the fetal aortic tortuosity and DAA. In the present study, this technique suggested the aortic tortuosity and kinking late in the first trimester of pregnancy. Moreover, helical aortic tortuosity, aortic kinking, and DAA were clearly recognized using HDlive Flow at 20 weeks of gestation. The association of aortic tortuosity and DAA may be a new pathogenic concept of arteriopathies. Further studies using HDlive Flow with STIC are needed to test this hypothesis during the fetal period.

Figs 2A to D: HDlive Flow image of retrograde aortic flow, and aortic tortuosity (small arrow) and kinking (large arrow) at 13 weeks and 6 days of gestation. (A) Frontal view; (B) Panoramic view; (C) Posterior view; (D) Right lateral view (Ao, aorta; DA, ductus arteriosus; DAo, descending aorta; HV, hepatic vein; IVC, inferior vena cava; PA, pulmonary artery; RA, right atrium; RV, right ventricle)

Figs 3A and B: Mitral atresia (arrow) and wide oval thoracic aorta (small arrows) with pericardial effusion (PeE) and pleural effusion (PIE) using conventional fetal echocardiography at 20 weeks of gestation. (A) Two-dimensional sonography; (B) Radiant flow (DAo, descending aorta; LA, left atrium; LV, left ventricle)

Figs 4A to D: HDlive Flow image of aortic tortuosity (small arrows) and kinking (large arrow) with ductus arteriosus aneurysm (DAA) at 20 weeks of gestation. (A) Spatial three-vessel view; (B) Panoramic view; (C) Posterior view; (D) Right lateral view (Ao, aorta; AoA, aortic arch; DAo, descending aorta; PA, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava)

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