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Prenatal Diagnosis of Interrupted Aortic Arch with Intact Ventricular Septum and Persistent Left Superior Vena Cava at 20 Weeks: A Case Report.

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ABSTRACT: Interrupted aortic arch (IAA) is a rare, duct-dependent congenital heart defect that usually occurs with a ventricular septal defect (VSD) and is strongly associated with 22q11.2 microdeletion. Prenatal detection is difficult because the four-chamber view may appear nearly normal. We report a 33-year-old woman referred for fetal echocardiography at 20 weeks 4 days of gestation. Abdominal and cardiac situs were normal, with levocardia, a normal cardiac axis, and a regular rhythm (fetal heart rate 145 bpm) with 1:1 atrioventricular conduction. There was marked ventricular disproportion: the left ventricle was small (diastolic diameter 3.90 mm, Z-score -3.68) compared with the right ventricle (9.14 mm, Z-score +1.69), and the left atrium was small. Flow across the foramen ovale was reversed (left-to-right). The interventricular septum was intact. The aorta arose from the left ventricle and was markedly hypoplastic. The aortic isthmus measured 1.04 mm (Z-score -3.85) on the transverse view of the upper mediastinum, and on sagittal greyscale and colour Doppler imaging the aortic arch could not be traced in continuity with the descending aorta. The pulmonary arteries were confluent and the ductus arteriosus was normal. A persistent left superior vena cava draining into the coronary sinus was also seen. The findings were consistent with IAA. The parents were counselled about the guarded prognosis, and amniocentesis was offered to rule out aneuploidy and 22q11.2 deletion. This case shows that ventricular disproportion, reversed foramen ovale flow, and a small ascending aorta should prompt careful evaluation of the aortic arch, and that early prenatal diagnosis supports genetic testing, informed counselling, and planned delivery at a centre with paediatric cardiac surgery.

INTRODUCTION

Interrupted aortic arch (IAA) is a complete loss of luminal continuity between the ascending and the descending aorta. It is rare and accounts for about 1% of critical congenital heart disease [1,2]. Celoria and Patton divided it into three types according to the site of interruption: type A, distal to the left subclavian artery; type B, between the left common carotid and left subclavian arteries; and type C, between the innominate and left common carotid arteries [1]. Blood flow to the lower body depends entirely on the ductus arteriosus. When the duct closes after birth, the newborn develops shock and severe acidosis, and the condition is fatal without treatment [2].

IAA almost always occurs with a ventricular septal defect (VSD), and type B in particular is strongly associated with 22q11.2 microdeletion (DiGeorge syndrome) [3,4]. IAA with an intact ventricular septum is an uncommon variant. Prenatal detection of IAA is difficult. The four-chamber view may appear nearly normal, and the diagnosis depends on careful assessment of the outflow tracts, the three-vessel and three-vessel-trachea views, and the sagittal view of the arch [5,6]. We report a fetus diagnosed at 20 weeks of gestation with IAA, an intact ventricular septum, and a persistent left superior vena cava (PLSVC), and discuss the echocardiographic features that led to the diagnosis and its implications for counselling and perinatal care.

Case Presentation

A 33-year-old woman was referred for fetal echocardiography at 20 weeks 4 days of gestation. Abdominal and cardiac situs were normal, with levocardia and a normal cardiac axis. The rhythm was regular, with a fetal heart rate of 145 bpm and 1:1 atrioventricular conduction on M-mode (Fig. 1). The four-chamber view showed marked ventricular disproportion. The left ventricle was small, with a diastolic diameter of 3.90 mm (Z-score -3.68), compared with 9.14 mm (Z-score $+1.69$) for the right ventricle [7] (Fig. 2). The left atrium was also small. Colour Doppler showed diastolic inflow predominantly into the right ventricle, with reduced filling of the left ventricle. Flow across the foramen ovale was reversed (left-to-right). The interventricular septum was intact.

The aorta arose from the left ventricle and was markedly hypoplastic. On the transverse view of the upper mediastinum, the aortic isthmus measured 1.04 mm (Z-score -3.85) (Fig. 3). On sagittal greyscale imaging, the aortic arch could not be traced in continuity with the descending aorta (Fig. 4). Colour Doppler with HDlive Flow rendering showed the ductal arch continuing into the descending aorta, and no flow was seen across a continuous aortic arch (Fig. 5, Fig. 6). The pulmonary arteries were confluent and the ductus arteriosus was normal. A persistent left superior vena cava draining into the coronary sinus was also seen.

The impression was interrupted aortic arch with an intact ventricular septum and a persistent left superior vena cava. The parents were counselled about the guarded prognosis, and amniocentesis was offered to rule out aneuploidy and 22q11.2 deletion.

Imaging Findings in Detail

Fetal echocardiography was performed with greyscale, M-mode, and colour Doppler imaging, including HDlive Flow rendering. Cardiac measurements were expressed as Z-scores for gestational age [7].

Fig. 1, an M-mode tracing through the fetal heart, shows a regular ventricular rhythm at 145 bpm with 1:1 atrioventricular conduction. Fig. 2, the four-chamber view, shows the marked ventricular disproportion, with a small left ventricle (3.90 mm, Z-score -3.68) and a dominant right ventricle (9.14 mm, Z-score $+1.69$). Fig. 3, a transverse view at the level of the upper mediastinum, shows the markedly hypoplastic aortic isthmus, measuring 1.04 mm (Z-score -3.85). Fig. 4, a sagittal greyscale view of the fetal thorax, shows that the aortic arch cannot be followed in continuity into the descending aorta. Fig. 5, a sagittal colour Doppler view, shows the great vessels and their relationship to the descending aorta, with no flow across a continuous aortic arch. Fig. 6, a sagittal view in greyscale and colour Doppler side by side, shows the ductal arch continuing into the descending aorta, again with no continuous aortic arch demonstrated. Fig. 7, a colour Doppler image of the heart in diastole, shows ventricular inflow directed predominantly into the dilated right ventricle, with reduced filling of the small left ventricle.

DISCUSSION

In this fetus, three findings on routine cardiac views pointed to an abnormality of the aortic arch: ventricular disproportion with a small left ventricle, a small ascending aorta, and reversed flow across the foramen ovale. None of these is specific, but together they indicate reduced flow through the left side of the heart and should lead to a targeted examination of the arch. In reported fetal series of IAA, a small ascending aorta relative to the pulmonary trunk and failure to demonstrate a continuous arch on the three-vessel-trachea and sagittal views were the main diagnostic features [4,5]. In our case, the sagittal greyscale and colour Doppler views showed only the ductal arch reaching the descending aorta.

The main differential diagnosis is severe coarctation of the aorta with tubular hypoplasia of the arch. In coarctation, the arch is narrow but continuous. In IAA, no connection can be shown between the ascending and the descending aorta. This distinction can be difficult in mid-gestation. In our fetus, a very small isthmus of 1.04 mm was measured on the transverse view, whereas

continuity could not be shown on sagittal imaging with greyscale or colour Doppler. A vessel of this size with very low flow is close to the limit of resolution at 20 weeks, so severe coarctation with extreme isthmal hypoplasia cannot be completely excluded on a single examination. Both lesions are duct-dependent after birth, and the perinatal plan is the same for both. Two features of this case are unusual. First, the ventricular septum was intact. Most fetuses with IAA have a VSD, which allows the left ventricle to fill and empty normally, so the four-chamber view is often nearly normal [5,6]. With an intact septum, the left ventricle has no alternative outlet, which may explain the marked ventricular disproportion and the reversed foramen ovale flow seen here. Second, the association between IAA and 22q11.2 deletion is strongest for type B interruption with a VSD [3,4]. The site of interruption relative to the head and neck vessels was not documented in this examination, so the Celoria and Patton type could not be assigned. Genetic testing remains indicated in every fetus with IAA, regardless of the type. A persistent left superior vena cava draining into the coronary sinus was also present. PLSVC is often an isolated finding with no clinical consequence, but when it is detected in fetal life it is frequently associated with other cardiac defects, and it should prompt a complete examination of the heart, including the aortic arch [8]. This report has limitations. The diagnosis rests on a single examination at 20 weeks 4 days. The type of interruption was not defined, and severe coarctation could not be fully excluded.

Management and Counselling

IAA is a duct-dependent lesion, and the main benefit of prenatal diagnosis is that the perinatal period can be planned. Delivery should take place at a centre with neonatal intensive care and paediatric cardiac surgery, so that a prostaglandin E1 infusion can be started promptly after birth to keep the ductus arteriosus open until surgical repair [2,9]. Serial fetal echocardiography is advisable to follow the growth of the left ventricle, the aortic valve, and the ascending aorta, because the size of the left heart structures influences the surgical approach and the prognosis. Genetic testing should be offered in every case. Amniocentesis with karyotype and testing for 22q11.2 deletion, by chromosomal microarray or fluorescence in situ hybridisation, is recommended because of the strong association with IAA [3,4]. A positive result has consequences beyond the heart, and it changes both the counselling and the care of the newborn [9]. Counselling should be multidisciplinary, should involve fetal medicine, paediatric cardiology, and genetics, and should cover the expected neonatal course, the need for surgery in the newborn period, and the options available to the parents.

In our patient, the parents were counselled about the guarded prognosis, and amniocentesis was offered to rule out aneuploidy and 22q11.2 deletion.

CONCLUSION

Interrupted aortic arch can be suspected at the mid-trimester scan. Ventricular disproportion with a small left ventricle, reversed foramen ovale flow, and a small ascending aorta should prompt careful evaluation of the aortic arch on the three-vessel-trachea and sagittal views with colour Doppler. IAA with an intact ventricular septum is an uncommon variant, and a persistent left superior vena cava is a further reason to examine the arch closely. Early prenatal diagnosis supports genetic testing, informed parental counselling, and delivery at a centre with paediatric cardiac surgery, where prostaglandin therapy can be started promptly.

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FIGURE LEGENDS

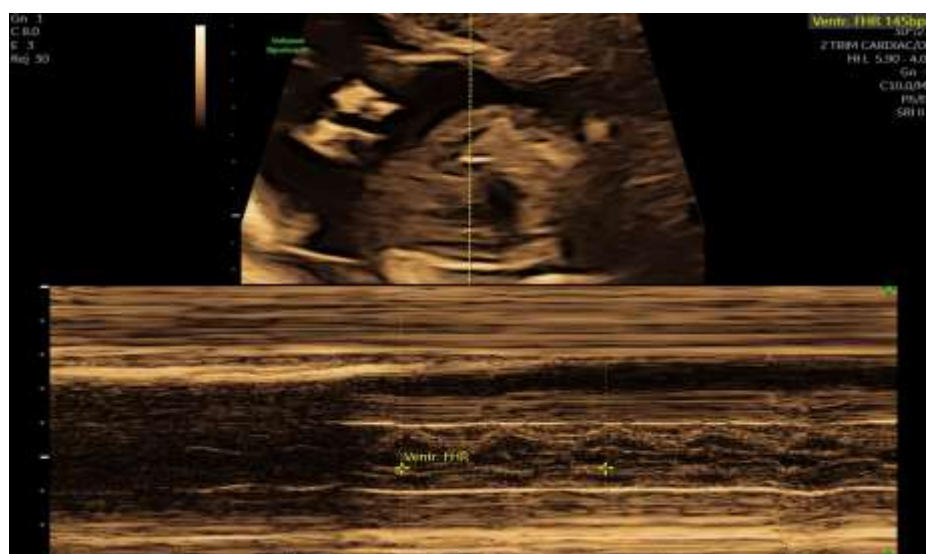


Fig. 1: M-mode echocardiogram through the fetal heart showing a regular ventricular rhythm with a fetal heart rate of 145 bpm and 1:1 atrioventricular conduction.



Fig. 2: Four-chamber view showing marked ventricular disproportion. The left ventricular diastolic diameter (LVD) is 3.90 mm (Z-score -3.68) and the right ventricular diastolic diameter (RVD) is 9.14 mm (Z-score +1.69).



Fig. 3: Transverse view at the level of the upper mediastinum showing a markedly hypoplastic aortic isthmus measuring 1.04 mm (Z-score -3.85).



Fig. 4: Sagittal greyscale view of the fetal thorax. The aortic arch cannot be traced in continuity with the descending aorta.



Fig. 5: Colour Doppler (HDlive Flow) sagittal view showing the great vessels and their relationship to the descending aorta; no flow is seen across a continuous aortic arch.

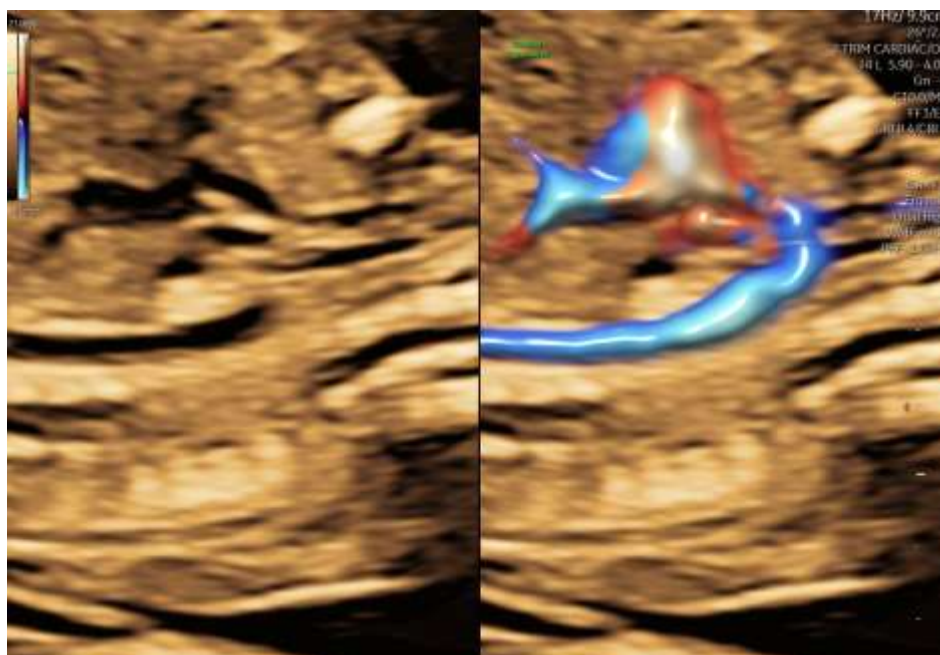


Fig. 6: Sagittal view in greyscale (left) and colour Doppler with HDlive Flow rendering (right) showing the ductal arch continuing into the descending aorta, with no continuous aortic arch demonstrated.

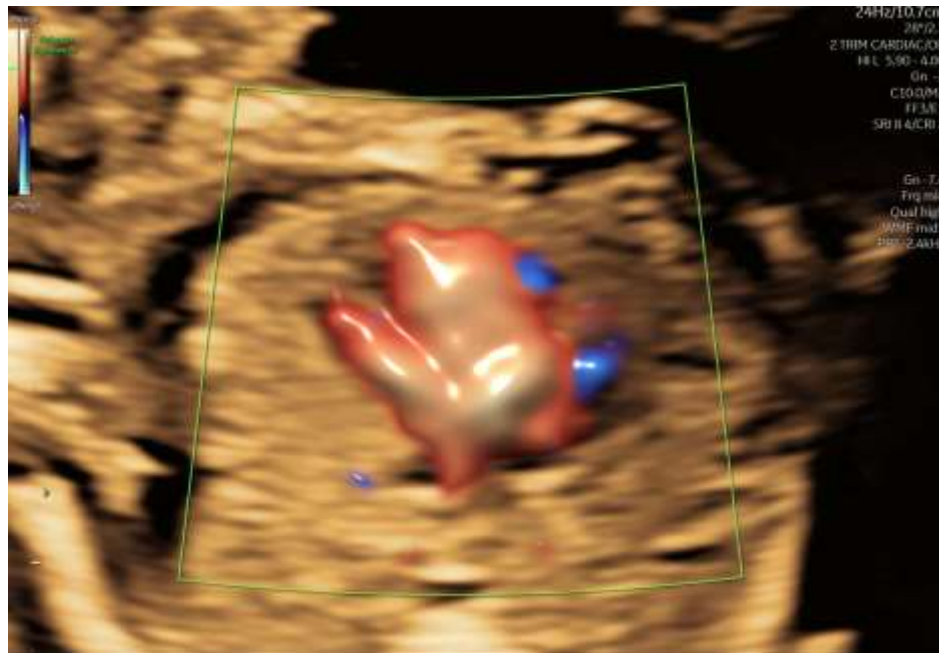


Fig. 7: Colour Doppler (HDlive Flow) of the fetal heart showing diastolic ventricular inflow, with flow predominantly into the dilated right ventricle and reduced filling of the small left ventricle.