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Case Report

Ultrasonographic approach to diagnosing coarctation of the aorta: case report

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ABSTRACT

Coarctation is the narrowing of the isthmus or aortic arch. It accounts for approximately 8% of all congenital heart defects; it is a cause of morbidity and mortality if not diagnosed early. Although it is a common congenital cardiac defect, it remains one of the most challenging cardiac defects to diagnose prenatally. Which is why the objective of this case report is to present a prenatal diagnosis of coarctation of the aorta and show how prenatal diagnosis with proper counseling can optimize management, and that learning about the ultrasonographic characteristics that raises suspicion of coarctation of the aorta and referring the patient to a maternal fetal medicine-specialist is a fundamental key to the diagnosis.

Keywords: Coarctation of the aorta, Prenatal diagnosis, Fetal echocardiography, Ultrasound

INTRODUCTION

Coarctation is the narrowing of the isthmus or aortic arch, typically located at the insertion of the ductus arteriosus or in the segment proximal to it.[1] It accounts for approximately 8% of all congenital heart defects; it is a cause of morbidity and mortality if not diagnosed early. After birth, the ductus arteriosus closes and may cause severe cardiac failure and poor perfusion of the lower body.

Although it is a common congenital cardiac defect, it remains one of the most challenging cardiac defects to diagnose prenatally. It is known that perinatal diagnosis improves survival of the neonates by allowing a planned delivery in a tertiary care hospital and early intervention with prostaglandin treatment to prevent closure of the ductus.

The suspicion of coarctation of the aorta is raised when the following ultrasonographic characteristics are visualized: ventricular disproportion, shelf or flow obstruction over the isthmus, discrepancy of the great vessels, or retrograde flow in the fetal aortic arch.

The objective of this case report is to present a prenatal diagnosis of coarctation of the aorta and show how prenatal diagnosis with proper counseling can optimize management.

CASE REPORT

A 30-year-old female with no past medical history, was referred for a fetal echocardiography examination at 24.4 weeks' gestational age due to a suspected coarctation of the aorta. During her second trimester ultrasound, we

noted right ventricular and atrial enlargement as well as great vessel disproportion.

The four chamber view of the fetal echocardiography revealed right ventricular and atrial enlargements. (Figure 1). In the aortic arch view, there was evidence of aortic stenosis in the proximal part of the descending aorta (Figure 2). On color Doppler imaging, the stenotic portion of the aorta was noted even more (Figure 3). According to findings, the prenatal diagnosis of coarctation of the aorta was suspected for the fetus. A non-invasive prenatal testing (NIPT) was requested to analyze small fragments of cell- free DNA circulating the pregnant woman's body to provide early detection of certain genetic abnormalities. The results were negative for trisomy 21, trisomy 18 and trisomy 13; no aneuploidy for sex chromosomes were found, and microdeletion was not detected.

The pregnancy was monitored by a multidisciplinary team including a psychologist, maternal fetal medicine specialist, pediatric cardiologist, neonatologist, and an interventional cardiologist until termination. At 38 weeks' gestational age, the mother had spontaneous rupture of membranes and a vaginal delivery of a male, weighing 3250 grams with Apgar scores of 8 and 9 at one and five minutes, respectively. Infusion with Prostaglandin E1 (PGE1) was started as soon as the newborn was born to keep the ductus arteriosus open.

After vaginal delivery, the pathological cardiovascular findings of coarctation of the aorta in the fetal echocardiography were confirmed with a computed tomography (CT) scan four hours after birth (Figure 4). The cardiac CT scan showed aortic arch hypoplasia.

Surgical relief of coarctation of the aorta was done on the day 7 of birth. No complications were seen during the operation and the newborn was stable after the surgical procedure. The patient is now in good condition and is being followed up with a pediatric cardiologist.

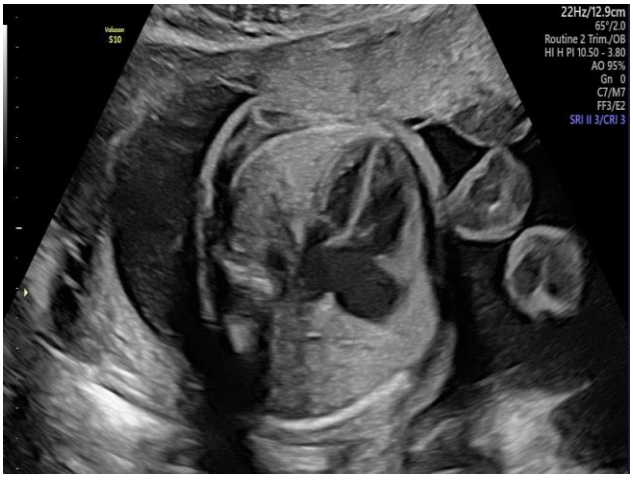

Figure 1: Four chamber view of the fetal echocardiography with right ventricular and atrial enlargements.

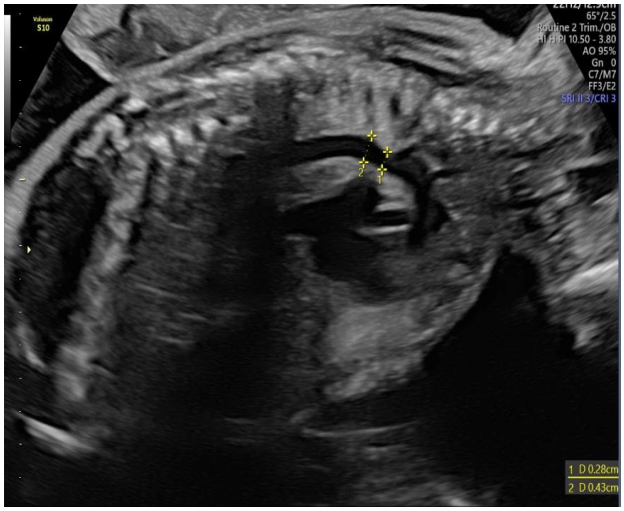

Figure 2: Aortic arch view with aortic stenosis in the proximal part of the descending aorta.

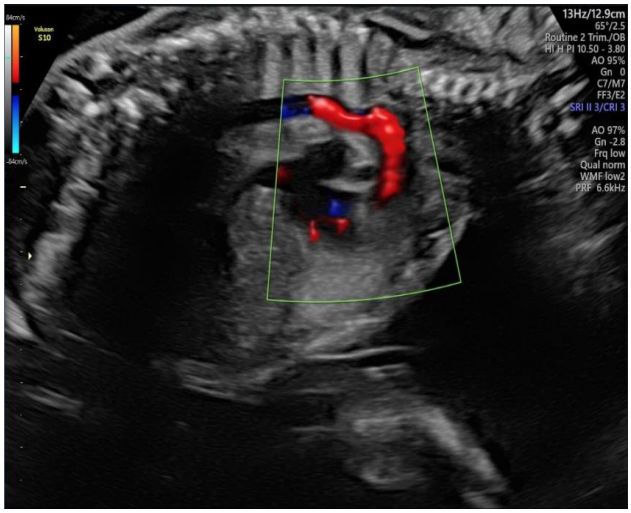

Figure 3: Color Doppler imaging with a stenotic portion of the aorta.

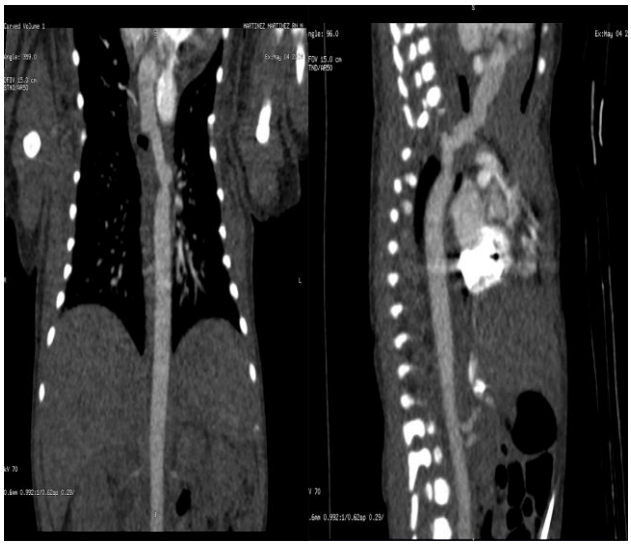

Figure 4: CT: coarctation of aorta.

DISCUSSION

Prenatal diagnosis of congenital heart defects has the potential of changing the epidemiological and public health impact in several ways, including the prevalence, mortality, and postnatal outcomes. Detection of congenital heart defects might be difficult because it depends on several factors, including the availability of adequate equipment, the sonographer skills, screening policies and access to prenatal services.[2]

The coarctation of the aorta represents the seventh most frequent congenital heart defect, with an incidence of 1 per 2,500 newborns. In Bakker´s cohort study, the prevalence reported in 12 different countries varied from 1.2 (in Slovak Republic) to 66.3 (in France) cases per 10,000 newborns, which means that coarctation of the aorta is one of the most poorly detected congenital heart defects.[2] This is why we are seeking to share the ultrasonographic approach to diagnosing coarctation of the aorta.

There are two theories that explain the pathophysiology of coarctation of the aorta: decreased ductal flow and ectopic ductal tissue. Due to reduced perfusion, there is decreased isthmic flow in the embryonic period; consequently, the isthmus region cannot develop properly and remains hypoplastic.[4] The ectopic ductal tissue theory is linked to the pathological invasion of the ductal tissue on the aortic wall covering its circumference. After birth, the ductal tissue contracts and closes the ductus, causing aortic narrowing.[5]

One of the ultrasonographic findings that raises suspicion for coarctation of the aorta is the ventricular disproportion, mainly a smaller left than right ventricle with a 70% sensitivity and 67% specificity and a PPV 73%.[6] This finding is more sensitive in the second trimester, before 25 weeks of gestational age and less in the third trimester, because there is a slight degree of physiological disproportion (normal: IV/RV<1.5).[6]

After the suspicion is raised, we should evaluate the aortic/ductal angle. Quarello et al defined a standard view in order to obtain and evaluate the aortic/ductal angle.[7] A sagittal view that includes the distal aortic and ductal arches and proximal descending aorta using power doppler 2D ultrasound. In normal fetuses the aortic/ductal angle ranges from 128.2° to 167° and in fetuses with coarctation of the aorta from 82.2° to 125°, being significantly smaller in the coarctation group.[7]

A coarctation of the aorta is almost always associated with a discrepancy of the great vessels where the diameter of the arteria pulmonalis is bigger than the diameter of the aorta during diastole. This is probably related to blood flow redistribution because of the increased resistance of the left ventricular out- flow tract.[6] Slodki et al found that the main PA: Ao diameter ratio can be a helpful tool for distinguishing true from false CoA. For a PA:Ao ratio of 1.60 or more, the sensitivity was 83%, specificity 85%, positive predictive value 62.5% and negative predictive value 94%.[8]

Using doppler flow, a retro- grade flow in the fetal aortic arch combined with a small left heart is suspicious for coarctation. B-flow imaging is potentially advantageous over color or power Doppler imaging when used in conjunction with stick for the evaluation of the fetal vasculature. A B-flow volume of the heart with off-line-mode tracing of the isthmic region (sampling at the level of the three-vessel view on the isthmus and ductal arch) can detect a diastolic run-off that is present in fetal aortic coarctation. In normal hearts the isthmus will show absence of diastolic-run off with a pulsatile flow pattern while in coarctation diastolic run-off with continuous forward flow throughout the cardiac cycle is present.[9]

Fetal coarctation can be associated with bicuspid aortic valve, aortic valve stenosis, and mitral stenosis.[10] Coarctation can also be associated with chromosomal abnormalities, especially when associated with other anomalies. In 14% of patients with Turner syndrome, a coarctation is present.[11]

In our case suspicion of CoA was made due to the dominance of the right articular and ventricular structures and great vessel disproportion, which led the physicians to order a fetal echocardiography examination in order to evaluate the fetal cardiovascular system status. Based on the American Society of Echocardiography, the absolute maternal indications include pregestational diabetes, anti-Ro antibodies, phenylketonuria, retinoid exposure, TORCH infections and fetal absolute indications include suspected structural cardiac anomaly, fetal extracardiac anomaly, hydrops fetalis, nuchal translucency >3.5 mm (11-14 SDG), genetic anomaly or monochorionic twin pregnancy. It also suggests considering performing an echocardiogram when the nuchal translucency is reported from 3.0 to 3.4 mm, exposure to teratogens (paroxetine, carbamazepine, lithium, sodium valproate), ACEs inhibitors or in vitro fertilization. The ideal gestational age for performing an echocardiogram is between 18 to 24 weeks and for follow-up, it is recommended to repeat this study every 2 to 8 weeks, specifically doing it at the 34-36 weeks for determining the severity.

CONCLUSION

Coarctation of the aorta is one of the most poorly detected congenital heart defects because it depends on several factors, including the availability of adequate equipment, the sonographer skills, screening policies and access to prenatal services. The increasing trends in prenatal diagnosis highlight the potential for significant changes in the clinical outcomes for these types of patients. This is why learning about the ultrasonographic characteristics that raises suspicion of coarctation of the aorta and referring the patient to a maternal fetal medicine-specialist is a fundamental key to the diagnosis.

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