Prenatal Diagnosis and Outcome of Right Aortic Arch without Significant Intracardiac Anomaly

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Background: Right aortic arch (RAA) is usually associated with the presence of a significant congenital heart disease, usually a conotruncal defect, which determines the postnatal outcome. In the absence of such cardiac defects, the significance of RAA has not been determined. The aims of this study were to evaluate the significance of recognizing RAA in fetuses with normal or near normal intracardiac anatomy and to determine which associations may be present.

Methods: A retrospective study was completed of all fetuses diagnosed with RAA with normal or near normal intracardiac anatomy between 1999 and 2011. The aim was to evaluate the presence of RAA with complete ultrasonic evaluation using two-dimensional imaging complemented by the Doppler color flow technique, paying particular attention to the three-vessel and tracheal view. We compared the prenatal findings with the postnatal outcomes and management of this cohort of fetuses.

Results: Among 16,450 fetal echocardiograms, 58 fetuses (0.35%) were diagnosed with RAA with normal or near normal intracardiac anatomy. Gestational age at diagnosis ranged from 19 to 34 weeks (mean, 23 weeks). Isolated RAAs were found in 50 fetuses, and double aortic arches (DAAs) were recognized in eight other cases. The postnatal cohort consisted of 44 newborns with RAAs and eight with DAAs (two were lost to follow-up, and four pregnancies were terminated). Postnatal echocardiography confirmed the prenatal diagnosis of RAA in 41 of 45 children, and four were found to have DAAs. Three of seven fetuses diagnosed prenatally as having DAAs were found to have only RAAs. Fourteen fetuses underwent karyotyping; two had 22q11 deletion and two had 47xxy. Eleven infants (21%) had respiratory symptoms, eight with DAAs, one with RAA, mirror-image head and neck vessels, and two with RAAs and aberrant left subclavian arteries. Surgery was indicated in all symptomatic patients except one, whose symptoms resolved. One asymptomatic patient underwent operation for significant compression of the trachea.

Conclusions: RAA on fetal ultrasonography may indicate vascular and chromosomal abnormalities that may complicate postnatal management. When RAA is identified, fetal karyotype analysis (including the integrity of chromosome 22) is warranted. RAA may herald an occult DAA and may be a clue to a tight vascular ring. Hence, it seems essential to conduct a careful postnatal evaluation of fetuses with RAAs on prenatal ultrasound. (J Am Soc Echocardiogr 2014;■:■-■.)

Keywords: Echocardiography, Vascular ring, Fetal anomalies

Right aortic arch (RAA) is a congenital vascular anomaly present in 0.086% to 0.1% of pregnancies.[1,2] It usually accompanies other congenital heart defects, most often tetralogy of Fallot (with or without pulmonary atresia).[3] Nevertheless, in 25% of cases, it is present in association with normal or near normal intracardiac anatomy.[3,4] A vascular ring can most commonly be formed by a double aortic arch (DAA) or by RAA with a left-sided ductus connected to a diverticulum of Kommerell, giving rise to an aberrant left subclavian artery. Such vascular rings may lead to respiratory distress or dysphagia during infancy or later in life.[5-8]

With the development of high-resolution ultrasound, the aortic arch course can now be identified prenatally. After birth, the prognosis of patients with RAAs with congenital heart defects is usually determined by the severity of the cardiac defects.[3,4] The prognosis of patients with RAAs with normal cardiac anatomy, however, is determined by vascular compression of the trachea or esophagus, giving rise to respiratory symptoms or dysphagia.[5] The purpose of the present study was to determine how the prenatal diagnosis of RAA with normal or near normal intracardiac anatomy might focus attention on appropriate postnatal management and treatment, thus improving outcomes.

METHODS

We performed a retrospective review of all fetal ultrasound studies at our institution between 1999 and 2011 and found 16,450 patients who had undergone fetal echocardiography.

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Abbreviations
DAA = Double aortic arch
RAA = Right aortic arch
VSD = Ventricular septal defect

Fetuses with normal or near normal intracardiac anatomy, diagnosed as having RAA alone or as part of a DAA were enrolled in the study. We did include fetuses and newborns with mild intracardiac lesions that do not have hemodynamically significant consequences, such as small ventricular septal defects (VSDs), bicuspid aortic valve, and left superior vena cava draining to the coronary sinus.

All patients underwent complete transabdominal fetal ultrasound studies using an Acuson Sequoia 512 ultrasound system (Siemens Medical Solutions USA, Inc, Mountain View, CA). The aortic arch and trachea were imaged in a transverse view of the fetal upper thorax (the three-vessel and tracheal view). The images were complemented with Doppler color flow imaging to enhance the morphologic information and to assess flow direction. Special attention was paid to the Nyquist scale, lowering it to levels appropriate to detect fetal blood flow. At this tomographic plane, the transverse aorta, pulmonary artery, superior vena cava, and trachea are demonstrated. In the normal heart, the pulmonary trunk is the largest and most anterior and leftward vessel, and the superior vena cava is the smallest most posterior and rightward vessel. The transverse aorta lies between these vessels to the left of the trachea, recognized as a circular structure with an echogenic wall and an echo-free lumen anterior to the spine (Figure 1). When RAA is present, the transverse aorta lies to the right of the trachea (Figure 2).[1,9-12] When an anomalous left subclavian artery is noted, its origin is noted to arise from the descending aorta, and it runs behind the trachea and to the left of the region where the left ductus and aorta join (Figure 3). In the case of DAA, it was possible to recognize the components of the arch surrounding the trachea (Figure 4).

We noted gestational age at the time of the fetal ultrasound study. We offered chromosomal analysis to all women who were examined to evaluate chromosomal anomalies. We evaluated the postnatal records, including surgical reports, of this cohort, noting the clinical findings, presence or absence of respiratory symptoms, and echocardiographic findings. We also assessed the magnetic resonance or computed tomographic x-ray reports and bronchoscopy reports, if the pulmonologist decided that they were appropriate. We also contacted all nonsurgical patients by phone to assess their well-being.

Statistical analysis was performed by means of two-by-two tables to assess sensitivity, specificity, and positive and negative predictive values. All pertinent patient data were recorded on a spreadsheet and made anonymous. Our institutional ethics committee approved the study.

RESULTS

We performed 16,450 complete fetal cardiac ultrasonic examinations between 1999 and 2011. RAAs with normal or near normal intracardiac anatomy were noted in 58 fetuses (0.35%). The mean gestational age was 22.7 weeks (range, 19–34 weeks; median, 22 weeks). Mean follow-up time after birth was 49 months (range, 17–112 months; median, 40 months).

There were 58 fetuses prenatally suspected to have RAAs, eight of whom were suspected to have DAAs (Figure 5; Table 1).

Among the group of 50 fetuses with RAAs, 37 had been referred with that diagnosis. Three of the 50 were referred for DAAs, and 10

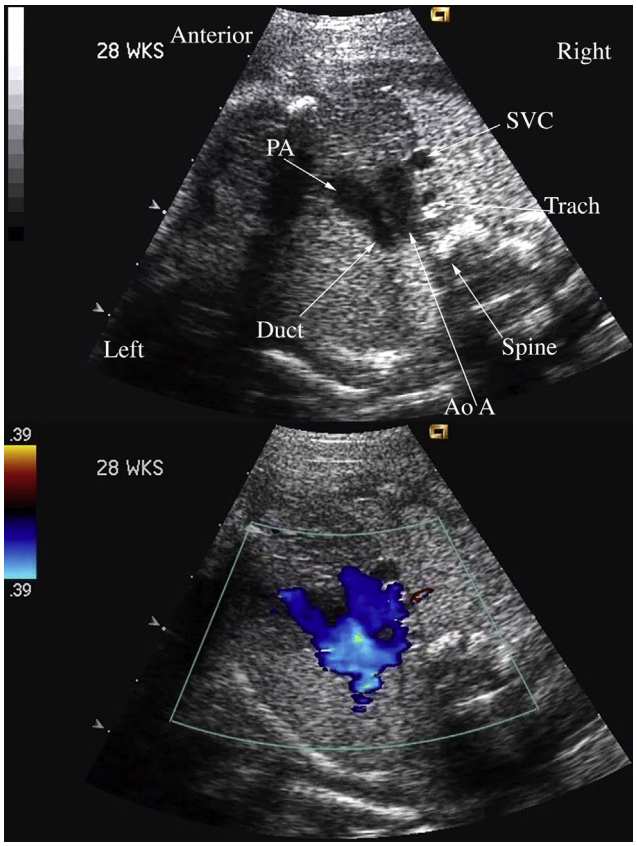

Figure 1 (*Top*) This figure shows the normal position of the aortic arch in the three-vessel view in a fetus at 28 weeks' gestation. The superior vena cava (SVC) is noted on the right. The aortic arch (Ao A), the pulmonary artery (PA), and the ductus arteriosus (*Duct*) are identified sequentially from right to left, as indicated on the figure. The aortic arch runs to the left of the trachea (*Trach*). The vertebral spine is also indicated, as are the right and left laterality markers. (*Bottom*) Doppler color flow image from the same fetus showing the directionality of flow in both the aortic and ductal and confirming the vascular structures to the left of the trachea.

were referred for other reasons, such as family history or nonconotruncal-associated heart defects. In this group, four pregnancies were terminated because of chromosomal anomalies, and one patient was lost to follow-up, leaving 45 postnatal records available for evaluation after birth. Postnatal echocardiography confirmed the prenatal diagnosis of RAA in 41 of 45 children, and four were found to have DAAs.

From the group of eight fetuses with DAAs, five were referred for diagnosis of RAA, and three were referred for other reasons. One patient was lost to follow-up, leaving seven postnatal records available for evaluation after birth. Three of the fetuses diagnosed prenatally as having DAAs were found to have only RAAs (Figure 6).

For 56 fetuses, information about the sidedness of the ductus arteriosus was available; only four fetuses (7%) had right ductus arteriosus (Table 1). A right ductus arteriosus does not complete a vascular ring, and all four fetuses were eventually asymptomatic after birth.

Only 14 fetuses underwent karyotype analysis, including fluorescence in situ hybridization analysis for 22q11 deletion, because of parental religious and cultural preferences. Two had chromosome