

# Perinatal outcomes and anomalies associated with fetal right aortic arch

## *Fetal sağ aortik ark: perinatal sonuçları ve ilgili anomaliler*

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### Abstract

**Objective:** To evaluate the prenatal findings, associated anomalies, and prognosis of right aortic arch (RAA) anomalies.

**Material and Methods:** All cases referred for detailed ultrasonography and fetal echocardiography between October 2006 and July 2009 were systematically examined for aortic arch anomalies and associated cardiac and extracardiac anomalies. Prenatal findings of all cases with aortic arch anomalies and intracardiac and extracardiac findings were prospectively registered in an electronic database that included fetal echocardiography. Outcomes of the cases were collected postnatally from the patients' obstetricians, neonatal unit archives, and pediatric cardiologists.

**Results:** We detected 12 cases of RAA (0.37%; n=12/3200). Mean gestational age at diagnosis was 24 weeks (range, 21–33 weeks). Of the 12 cases of RAA, five (41.7%) had a major cardiac defect, including tetralogy of Fallot (n= 3), atrioventricular septal defect (n=1), and ventricular septal defect (n=1). An extracardiac anomaly was observed in three cases (25%). The fetal karyotype was trisomy 21 in one case with increased nuchal translucency (6.6 mm). Microdeletion 22q11 analyses performed in three cases were normal. The postnatal courses of the cases with isolated RAA were uneventful. Two cases associated with major cardiac and extracardiac anomalies were lost in the early neonatal period. The case of trisomy 21 was terminated. The other four cases of RAA with an associated cardiac anomaly are currently in follow up.

**Conclusion:** Aortic arch anomalies, particularly RAA, can be diagnosed by fetal echocardiography. The prognosis for isolated RAA is relatively good compared with that for RAA with associated anomalies. (J Turkish-German Gynecol Assoc 2012; 13: 184-6)

**Key words:** Right aortic arch, aortic arch anomaly, fetal echocardiography, fetus, prenatal diagnosis

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### Özet

**Amaç:** Prenatal dönemde, sağ aortik ark tanısı alan hastaların prognozu ve diğer anomalilerle birlikteliğini değerlendirmek.

**Gereç ve Yöntemler:** Kasım 2006 ile Temmuz 2009 yılları arasında ünitemize obstetrik ultrasonografi ve fetal ekokardiyografi yapılması için refere edilen vakalar sağ aortik ark ve ilgili diğer kalp ve kalp dışı anomalilerin varlığı için sistematik olarak muayene edildi. Prenatal dönemde, sağ aortik ark tanısı ve diğer kalp ve/veya kalp dışı anomali tanısı alan hastalar prospektif olarak elektronik ortama kaydedildi. Postnatal dönemdeki sonuçlar hastanın kadın doğum hekiminden, neonatal ünite dosyalarından ve pediatrik kardiyoloji hekiminden alındı.

**Bulgular:** Çalışma süresi boyunca on iki hastaya sağ aortik ark tanısı konuldu (%0.37; n:12/3200). Tanı ortalama olarak 24'üncü gebelik haftasında kondu (ortalama: 21-33 hafta). Aortik ark anomali tanısı konan 12 hastanın beş (%41.7) tanesinde major kardiyak anomali saptandı. Bu anomaliler; fallot tetralojisi (TOF, n:3), atriyoventriküler septal defekt (AVSD, n:1) ve ventriküler septal defekt (VSD, n:1) idi. Çalışma grubumuzdaki üç hastada (%25) kalp dışı anomali varlığı tespit edildi. Ense saydamlığı (NT: 6.6 mm) artmış olan bir hastanın karyotip analizinde Trizomi 21 tespit edildi. Microdelesyon 22q11 analizi yapılan üç hastanın sonuçları normal olarak bulundu. Major kardiyak anomali ve kalp dışı anomali olan iki hasta erken neonatal dönemde ex oldu. Trizomi 21 tanısı konulan hastanın gebeliği sonlandırıldı. Diğer sağ aortik ark ve ilgili kalp anomali tanısı alan dört hastanın takipleri halen sürmektedir.

**Sonuç:** Aortik ark anomalilerinin, özellikle sağ aortik ark anomalinin tanısı fetal ekokardiyografi ile konulabilir. İzole sağ aortik ark anomalilerin prognozu, sağ aortik ark ve eşlik eden diğer anomaliler varlığı ile karşılaştırıldığında nispeten daha iyidir.

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**Anahtar kelimeler:** Sağ aortik ark, aortik ark anomali, fetal ekokardiyografi, fetus, prenatal tanı

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## Introduction

Right aortic arch (RAA) is a relatively rare anomaly. The true incidence is unknown, but it is estimated to be 1 in 1000 in low-risk populations (1). RAA may occur in isolation or in association with cardiac and extracardiac anomalies. Prenatal diagnosis of RAA anomalies has not been a focus of attention until recently. So far, a few case series and some case reports have been published, but information regarding outcomes and associated anomalies is still limited. In this study, we report the outcomes and associated anomalies in cases of RAA detected in the fetus (2-8).

## Material and Methods

All cases referred for detailed ultrasonography (US) and fetal echocardiography between October 2006 and July 2009 were systematically examined with a US 4-8 MHz curved array probe (GE Voluson 730, Vienna, Austria), and all cardiac and extracardiac anomalies were noted prospectively. Fetal echocardiography was carried out using the segmental approach with standardised anatomical planes and the three-vessels-trachea view as an integral part of all examinations. RAA was diagnosed when an aortic arch was present on the right side of the trachea or on the right side with a U-shaped confluence (Figure 1, 2). Other aortic arch anomalies associated with cardiac and extracardiac anomalies were also identified. Fetal karyotype and 22q11 microdeletion analyses were performed in three cases. Prenatal findings of all cases were registered in an electronic prenatal database that included fetal echocardiography. Outcomes of cases were collected postnatally from the patients' obstetricians, neonatal units, and pediatric cardiologists. Prenatal diagnoses of RAA and associated anomalies were confirmed by pathological examination or postnatal echocardiography.

## Results

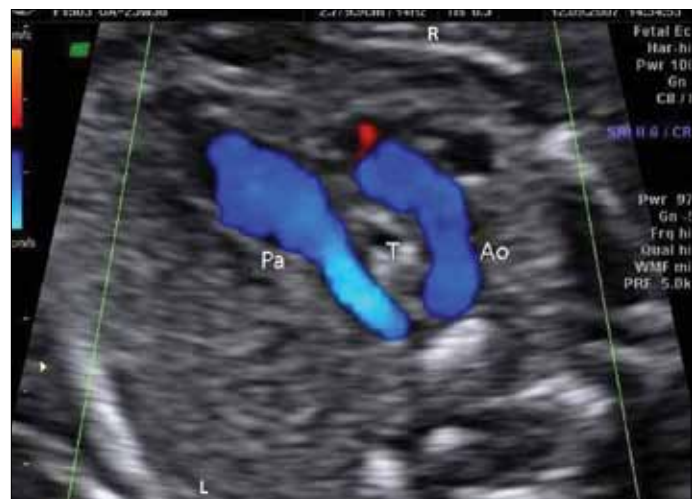
We detected 12 cases with RAA (0.37%; n=12/3200) (Table 1). Mean gestational age at diagnosis was 24 weeks (range, 21-33 weeks). Of the 12 cases of RAA, five (41.7%) were complicated with other major cardiac defects, including tetralogy of Fallot (TOF, n=3), atrioventricular septal defect (AVSD, n=1), and ventricular septal defect (VSD, n=1). Extracardiac findings were observed in three cases (25%). The fetal karyotype was trisomy 21 in one case with increased nuchal translucency (6.6 mm). Microdeletion 22q11 analyses were performed in three cases, and results were normal. The postnatal courses of the isolated RAA cases were uneventful. Two cases associated with major cardiac and extracardiac anomalies were lost during the early neonatal period. The case of trisomy 21 was terminated. The other four cases of RAA with associated cardiac anomalies are currently in follow-up.

## Discussion

Among the various anomalies of the aortic arch involving the vessel course and/or its branching pattern, those most

commonly seen include RAA with aberrant left subclavian or innominate arteries, RAA with mirror-image branching, double aortic arch, circumflex retroesophageal aortic arch, or left aortic arch with an aberrant right subclavian artery (1). The frequency of RAA among adults is approximately 0.1%, and a similar incidence was reported in a prenatal low-risk cohort (2, 3). Many undiagnosed cases are detected when RAA is investigated retrospectively.

RAA detected in the fetus is frequently associated with other cardiac and extracardiac malformations. The risk of concomitant congenital heart disease is >90% with the mirror-image branching type of RAA and only 10% with RAA and an aberrant left subclavian artery (3). Berg et al. (3) reported 71 fetuses with RAA, and additional cardiac abnormalities were noted in 22 (30%) of these cases. The most common cardiac lesions in that study were TOF, pulmonary atresia with VSD, and a common arterial trunk. Zidere et al. (4) reported 50 (66%) fetuses with other cardiac anomalies, the most common being TOF with pulmonary atresia. In our series, RAA was found in association



**Figure 1. Fetal right aortic arch**

Ao: aorta, Pa: pulmonary artery, T: trachea



**Figure 2. Fetal right aortic arch with tetralogy of Fallot**

Ao: aorta, Pa: pulmonary artery, T: trachea

**Table 1. Intracardiac, extracardiac anomalies and outcome in the cases of right aortic arch (n:12)**

No.	GA at diagnosis	Cardiac anomaly	Extracardiac anomaly	Outcome
1	23	-	-	Alive
2	22	-	-	Alive
3	21	-	-	Alive
4	24	-	-	Alive
5	23	-	-	Alive
6	33	-	Trachea and anal atresia, polyhydramnios	NND
7	24	VSD	Pelvic kidney	Alive
8	23	TOF	-	Alive
9	24	TOF	-	Alive
10	28	AVSD	-	Alive
11	30	TOF with absent pulmonary valve	-	NND
12	22	-	NT: 6.6 mm, Trisomy 21	Termination

GA: gestational age, week, RAA: right aortic arch, TOF: tetralogy of Fallot, VSD: ventricular septal defect, AVSD: atrioventricular septal defect, NT: nuchal translucency, NND: neonatal death

with additional intracardiac malformations in five (41.7%) cases (TOF, n=3; AVSD, n=1; VSD, n=1). Furthermore, RAA was associated with an extracardiac anomaly in three fetuses (25%). In a study by Berg et al., (3) this finding was observed in 12% of the entire series. These findings suggest that a meticulous inspection of fetal cardiac and extracardiac anatomy, including the brachiocephalic branching pattern, should be performed in prenatally detected cases and that cytogenetic testing for 22q11 deletions should be considered carefully.

A right-sided aortic arch may be asymptomatic. Sometimes, affected infants and children can present at any time with symptoms of airway obstruction, usually stridor. Symptoms in infancy are related to congenital heart anomalies or to compression of mediastinal structures such as the trachea or the esophagus.

### Conclusion

RAA can be prenatally diagnosed by fetal echocardiography and may be associated with cardiac anomalies. Once RAA is diagnosed, cardiac and extracardiac anomalies, fetal karyotype, and 22q11 deletion should be suspected. The prognosis depends on whether RAA is associated with cardiac and extracardiac anomalies. The prognosis is relatively good for isolated RAA.

### Conflict of interest

No conflict of interest was declared by the authors.

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