DOI: 10.37990/medr.1641836 Med Records 2025;7(2):412-8

MEDICAL RECORDS-International Medical Journal

Research Article

Aberrant Right Subclavian Artery in Prenatal Diagnosis: A Retrospective Study

Sadullah Ozkan[1], Alperen Aksan[2]

[1]Sivas Numune Hospital, Department of Perinatology, Sivas, Türkiye
[2]Şar Hospital, Department of Obstetrics and Gynecology, Rize, Türkiye

Abstract

Aim: Aberrant right subclavian artery (ARSA) is the most common anomaly of the aortic arch and occurs in 1-2% of the population. Although it is usually asymptomatic, its prenatal detection has gained importance due to associations with chromosomal abnormalities, including trisomy 21 and 22q11.2 microdeletion. This study examines isolated (iARSA) and non-isolated ARSA (niARSA), focusing on diagnostic approaches and neonatal outcomes.

Material and Method: In this retrospective study, 29 pregnancies diagnosed with ARSA between October 2022 and January 2024 were analyzed. Fetuses were classified as iARSA or niARSA based on additional structural or chromosomal findings. Data were collected from high-resolution ultrasound examinations and medical records, and statistical comparisons were performed using SPSS v25.0.

Results: There were a total of 29 cases of ARSA, of which 16 were iARSA (55.2%) and 13 were niARSA (44.8%). Non-invasive prenatal testing was performed in 68.7% of iARSA cases, all of which had normal results. In contrast, invasive testing was performed in 38.5% of niARSA cases, with chromosomal abnormalities detected in two cases (trisomy 21). Neonatal outcomes were favorable in iARSA, with 15 cases discharged without complications. NiARSA cases had higher morbidity, including NICU admissions (46%) and congenital heart defects, which in some cases required surgical intervention.

Conclusion: ARSA is an important marker in prenatal diagnosis. While iARSA generally indicates favorable outcomes, niARSA correlates strongly with unfavorable neonatal outcomes and chromosomal abnormalities. The distinction between iARSA and niARSA is crucial for tailored prenatal management and optimization of neonatal care strategies.

Keywords: Aberrant right subclavian artery, chromosomal abnormalities, congenital heart defects, neonatal outcomes, prenatal diagnosis

INTRODUCTION

Aberrant right subclavian artery (ARSA), the most common anomaly of the branching pattern of the aortic arch, occurs in about 1-2% of the general population (1,2). ARSA results from the failure of normal regression in embryonic development, causing the artery to originate distal to the aortic arch and cross behind the trachea and esophagus toward the right upper limb (2). Although ARSA is usually asymptomatic and considered benign in the general population, it has gained clinical importance as a marker in prenatal diagnosis, especially for chromosomal abnormalities such as trisomy 21 and 22q11.2 microdeletion syndrome (2,3).

The detection of ARSA has been linked to advances in prenatal imaging, particularly the use of color Doppler ultrasonography in the second trimester. Studies report that ARSA can be identified in 82-95% of cases using the three-vessel and tracheal view, allowing accurate visualization of its progression (1,4). However, ARSA is not only a marker for chromosomal abnormalities, but is also associated with other structural abnormalities that primarily affect the cardiovascular system, such as conotruncal defects (2,4).

Isolated ARSA (iARSA) carries a much lower risk of chromosomal abnormalities than non-isolated ARSA (niARSA), where additional structural or sonographic markers are present. This distinction serves as a basis for clinical decision making, with invasive testing recommended primarily in niARSA cases (2,3). In addition, cell-free DNA testing has been shown to be a non-invasive alternative for chromosomal risk assessment in fetuses with iARSA, demonstrating high sensitivity and specificity for conditions such as trisomy 21 (4,5).

CITATION
Ozkan S, Aksan A. Aberrant Right Subclavian Artery in Prenatal Diagnosis: A Retrospective Study. Med Records. 2025;7(2):412-8. DOI:1037990/medr.1641836

Received: 18.02.2025 **Accepted:** 05.03.2025 **Published:** 08.05.2025
Corresponding Author: Alperen Aksan, Şar Hospital, Department of Obstetrics and Gynecology, Rize, Türkiye
E-mail: alprnaksn@gmail.com

Despite these advances, there is still a need to clarify the implications of ARSA in different clinical contexts, particularly its association with rare genetic syndromes and long-term neonatal outcomes. This study aims to contribute to the growing body of knowledge by analyzing the co-occurrence of major cardiac and chromosomal abnormalities in fetuses diagnosed with ARSA.

MATERIAL AND METHOD

This retrospective cohort study was conducted in a tertiary perinatology center evaluating pregnancies diagnosed with ARSA by second trimester ultrasonography. Ethical approval was obtained from the local ethics committee before the study began.

The study included pregnant women aged 18–45 years who underwent detailed fetal anomaly screening between October 2022 and January 2024. Cases with ARSA identified on ultrasound were classified as either iARSA or niARSA based on the presence of additional structural abnormalities or sonographic markers, including increased nuchal fold thickness, nasal bone hypoplasia, echogenic bowel, intracardiac echogenic focus, and choroid plexus cysts. Exclusion criteria included pregnancies without ARSA findings on ultrasound or incomplete medical records.

The ultrasound examinations were performed with high-resolution transabdominal ultrasound systems which are Voluson E8 GE ultrasound machine (GE Medical Systems, Milwaukee, WI, USA) equipped with a convex 4–8 MHz transabdominal transducer, by experienced sonographers. The diagnosis of ARSA was confirmed in the three-vessel and tracheal view using color Doppler imaging to identify the typical course behind the trachea (Figure 1).

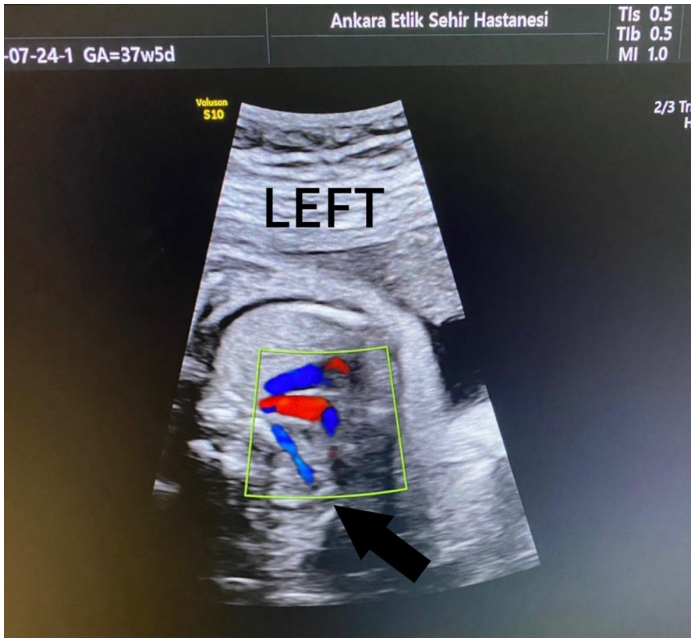

Figure 1. Ultrasound image of the three-vessel trachea view in a 37 week fetus; An ARSA (arrow) can be visualised arising from the distal aortic arch, coursing towards the right arm; Note the colour scale is set to approximately 20cm/s to enable visualisation of flow within the subclavian artery

Medical records were reviewed to collect data on maternal demographics, prenatal screening results, fetal structural findings, and chromosomal test outcomes. In cases where chromosomal analysis was performed, results were obtained through karyotyping or chromosomal microarray analysis.

The primary outcome was the prevalence of chromosomal abnormalities (e.g., trisomy 21, 22q11.2 microdeletion) and major congenital anomalies in fetuses with ARSA. Secondary outcomes included pregnancy and neonatal outcomes, such as delivery mode, gestational age at delivery, and postnatal complications associated with ARSA.

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional research committee at which the studies were conducted (Clinical Research Ethics Committee of Ankara Etlik City Hospital No. 1 (Decision No.: AEŞH-EK-2024-001, date: 31/01/2024) and with the 2013 Helsinki declaration and its later amendments or comparable ethical standards.

The statistical analyzes of this study were carried out using SPSS v25.0 software. Descriptive statistics were presented as median (interquartile range, IQR) for non-normally distributed variables and as percentages for categorical variables. For group comparisons, the Mann-Whitney U test was used for non-normally distributed continuous variables and Pearson Chi-Square was used for categorical variables. A p-value <0.05 was considered statistically significant.

RESULTS

A total of 29 pregnancies with diagnosed ARSA were included in the analysis, of which 16 (55.2%) were classified as iARSA and 13 (44.8%) as niARSA. Table 1 provides an overview of the demographic and clinical characteristics of the groups. The mean maternal age and BMI were similar in the groups (p=0.975 and p=0.638, respectively). The mean gestational age at diagnosis was comparable at 23.06±2.61 weeks for iARSA and 23.30±4.83 weeks for niARSA (p=0.690) (Table 1).

Non-invasive prenatal testing (NIPT) was performed in 68.7% of iARSA cases (11/16), with all results normal. In contrast, no NIPT was performed in any of the niARSA cases. Invasive prenatal testing was performed in 38.5% of niARSA cases (5/13), with chromosomal abnormalities detected in two cases (both trisomy 21). No invasive tests were performed in the iARSA group (Table 2, Table 3).

Neonatal outcomes were significantly different between the groups. In the iARSA group, 15 out of 16 neonates (93.8%) were discharged without complications, while one neonate had to stay in the neonatal intensive care unit (NICU) for 11 days due to transient neonatal tachypnea, congenital pneumonia and other complications, all of which resolved with treatment (Table 4).

Table 1. Demographic and clinical characteristics of pregnant women diagnosed with isolated ARSA and non-isolated ARSA

	Groups		p-value
	iARSA (n=16)	niARSA (n=13)	
Age(year) (median) (IQR)	30.5 (11.5)	30.0 (10.5)	0.982
Body mass index (kg/m²) (median) (IQR)	27.00 (7.75)	30.00 (5.50)	0.643
Gravida (median) (IQR)	2 (2)	2 (2)	0.424
Parity (median) (IQR)	0 (1)	1 (1)	0.777
Gestational week at the time of diagnosis (week) (median) (IQR)	22.00 (3.50)	23.00 (5.50)	0.690
Gestational week at birth (week) (median) (IQR)	38.00 (1.75)	37.00 (3.0)	0.173
Birth weight (gr) (median) (IQR)	3055.00 (272.00)	3305.00 (910.00)	0.430
Type of delivery (n)			
Spontaneous	5	4	
Cesarean section	11	8	0.525
Termination	0	1	

Values are presented as median (interquartile range, IQR) for non-normally distributed variables; P-values are calculated using the Mann-Whitney U test for continuous variables and Pearson Chi-Square test for categorical variables; A p-value <0.05 was considered statistically significant; ARSA: aberrant right subclavian artery, iARSA: isolated aberrant right subclavian artery, niARSA: non-isolated aberrant right subclavian artery

Table 2. Maternal and neonatal characteristics for isolated ARSA cases

	Age (years)	BMI (kg/m²)	Gestational age at the time of diagnosis ARSA	Additional ultrasonographic findings	Additional CVS anomalies	PIT	Non-invasive test	Gestational age at delivery	Birth weight	Type of delivery	Gender of the newborn	Results of the prenatal tests
1	20	24	26	None	None	-	-	40	3100	CS	Female	None
2	30	31	23	None	None	-	-	37	3500	CS	Female	None
3	33	33	26.3	None	None	-	-	36	3310	Spontaneous	Male	None
4	22	26	23	None	None	-	-	36	3070	CS	Male	None
5	30	31	20	None	None	-	-	39	3500	CS	Male	None
6	30	25	23	None	None	-	+	39	3600	CS	Female	Normal
7	32	27	22	None	None	-	+	39	3200	CS	Male	Normal
8	27	24	21	None	None	-	+	38	3050	CS	Male	Normal
9	35	32	21	None	None	-	+	37	2790	CS	Female	Normal
10	38	33	22	None	None	-	+	39	3270	CS	Female	Normal
11	29	34	20	None	None	-	+	40	3680	Spontaneous	Female	Normal
12	32	24	23	None	None	-	+	40	3460	Spontaneous	Male	Normal
13	26	24	22	None	None	-	+	39	3060	CS	Male	Normal
14	24	23	20	None	None	-	+	38	2970	Spontaneous	Female	Normal
15	40	31	24	None	None	-	+	38	3010	CS	Female	Normal
16	36	27	22	None	None	-	+	36	2650	CS	Female	Normal

Birth weight is measured in grams (g); Gestational age is presented in completed weeks unless otherwise specified; Results of prenatal tests denote findings of chromosomal or genetic testing when applicable; Neonatal outcomes include complications such as respiratory or cardiac anomalies identified postnatally; ARSA: aberrant right subclavian artery, CS: cesarean section, CVS: cardiovascular system

Table 3. Maternal and neonatal characteristics for non-isolated ARSA cases

	Age (years)	BMI (kg/m²)	Gestational age at the time of diagnosis ARSA	Additional ultrasonographic findings	Additiona CVS anomalies	PIT	Non-invasive test	Gestational age at delivery	Birth weight	Type of delivery	Gender of the newborn	Results of the prenatal tests
17	22	26	25	Echogenic cardiac	None	-	-	38	2750	Spontaneous	Female	-
18	21	24	30	Polyhydramnios	Type 1 TA, dilated trunkal artery, multiple valve dysplasia	-	-	40	3130	Spontaneous	Male	-
19	34	27	23	Pes equinovarus, scoliosis, renal fusion, single umbilical artery	Agenesis of ductus venosus	-	-	38	3460	CS	Male	-
20	30	26	13	Hydrops fetalis, cystic hygroma, hypoplastic nasal bone	Tricuspid regurgitation	-	-	14	-	Termination		-
21	24	24	20	Echogenic cardiac	None	yes	-	37	2640	CS	Female	Normal
22	25	31	26	PUV	None	yes	-	37	3050	CS	Male	Normal
23	25	24	22	Sandal gap	None	Yes	-	39	3200	CS	Male	Normal
24	27	29	21	Fetal renal pelviectasis	None	Yes	-	40	3300	CS	Male	Normal
25	22	30	25	Single umbilical artery	None	-	-	37	2740	Spontaneous	Male	-
26	44	37	22	Choroid plexus cyst	Mitral valve regurgitation	-	-	36	3020	CS	Female	-
27	42	23	26	Renal pelviectasis	Inlet VSD	yes	-	35	1500	CS	Female	Normal
28	41	30	34	Polyhydramnios, hypoplastic nasal bone	None	-	-	39	3880	Spontaneous	Female	-
29	37	32	27	Renal pelviectasis	DV shunt, inlet VSD	-	-	40	4200	CS	Male	-

Birth weight is measured in grams (g); Gestational age is presented in completed weeks unless otherwise specified (e.g., termination at 14 weeks); Results of prenatal tests denote findings of chromosomal or genetic testing when applicable; Neonatal outcomes include complications such as respiratory or cardiac anomalies identified postnatally; ARSA: aberrant right subclavian artery, VSD: ventricular septal defect, CS: cesarean section, PUV: posterior urethral valve, CVS: cardiovascular system, DV: ductus venosus, TA: truncus arteriosus

Table 4. Summary of neonatal outcomes based on ARSA subtypes

	Type of ARSA	Additional ultrasonographic findings	Neonatal outcomes
1	iARSA	None	Normal outcome
2	iARSA	None	Normal outcome
3	iARSA	None	11 days in NICU; TTN, congenital pneumonia, bilateral PTX, PDA, small secundum ASD
4	iARSA	None	Normal outcome
5	iARSA	None	Normal outcome
6	iARSA	None	Normal outcome
7	iARSA	None	Normal outcome
8	iARSA	None	Normal outcome
9	iARSA	None	Normal outcome
10	iARSA	None	Normal outcome
11	iARSA	None	Normal outcome
12	iARSA	None	Normal outcome
13	iARSA	None	Normal outcome
14	iARSA	None	Normal outcome
15	iARSA	None	Normal outcome
16	iARSA	None	Normal outcome
17	niARSA	Echogenic cardiac finding	Normal outcome
18	niARSA	Polyhydramnios, Type 1 truncus arteriosus, VSD	Operated on the 7th postnatal day, deceased on the 13th postnatal day
19	niARSA	Pes equinovarus, scoliosis, renal fusion, SUA	Normal outcome
20	niARSA	Hydrops fetalis, cystic hygroma, hypoplastic nasal bone	Pregnancy terminated
21	niARSA	Echogenic cardiac finding	Normal outcome
22	niARSA	PUV	Normal outcome
23	niARSA	Sandal gap	Normal outcome
24	niARSA	Fetal renal pelviectasi	Normal outcome
25	niARSA	Single umbilical artery, RDS, IUGR, polyhydramnios, VSD, ASD	118 days in NICU; postoperative complications: pneumothorax, pneumopericardium, pneumonia, deceased after surgery
26	niARSA	Choroid plexus cyst	5 days in NICU; indirect hyperbilirubinemia, maternal pregnancy-induced hypertension
27	niARSA	Renal pyelectasis, IUGR, ASD, PFO	Down syndrome (prenatal diagnosis), 18 days in NICU
28	niARSA	Polyhydramnios, hypoplastic nasal bone	Postnatal diagnosis of Down syndrome, ASD, minimal pericardial effusion
29	niARSA	Renal pyelectasis, AVSD	Postnatal diagnosis of Down syndrome, successfully operated

Birth weight is measured in grams (g); Gestational age is presented in completed weeks unless otherwise specified (e.g., termination at 14 weeks); Results of prenatal tests denote findings of chromosomal or genetic testing when applicable; Neonatal outcomes include complications such as respiratory or cardiac anomalies identified postnatally; ARSA: aberrant right subclavian artery, iARSA: isolated aberrant right subclavian artery, niARSA: non-isolated aberrant right subclavian artery, NICU: neonatal intensive care unit , TTN: transient tachypnea of the newborn, PTX: pneumothorax, PDA: patent ductus arteriosus, ASD: atrial septal defect, VSD: ventricular septal defect, CS: cesarean section, PUV: posterior urethral valve, IUGR: intrauterine growth restriction, RDS: respiratory distress syndrome, BPD: bronchopulmonary dysplasia, CVS: cardiovascular system, DV: ductus venosus, TY: tricuspid valve regurgitation

In contrast, the niARSA group exhibited a higher rate of adverse neonatal outcomes, with NICU admission required in 46.1% of cases (6/13). Major complications included respiratory distress syndrome, severe congenital heart defects and chromosomal abnormalities, such as Down syndrome (identified in two cases). Two neonates in this group died due to complications following surgery for congenital anomalies (Table 4). A comparison between niARSA cases with normal chromosomal results and iARSA cases revealed significant differences in neonatal outcomes. Among the eight niARSA cases with normal chromosomal results, three (37.5%) required NICU admission due to complications such as respiratory distress syndrome, IUGR, and congenital anomalies requiring postnatal intervention. In contrast, only one iARSA case (6.3%) required NICU admission, with transient tachypnea of the newborn and congenital pneumonia that resolved with treatment.

The mean birth weight was 3.103±270 g in the iARSA group and 2.910±420 g in the niARSA group (p=0.668) (Table 1). The gestational age at birth was slightly lower in the niARSA group (36.9±2.3 weeks) than in the iARSA group (37.8±1.9 weeks), but the difference was not statistically significant (p=0.173) (Table 1).

Cesarean section was the predominant mode of delivery in both groups and was performed in 68.7% of all cases. Spontaneous vaginal delivery was performed in 31.3% of cases, with no significant differences between the groups (p=0.525) (Table 1).

DISCUSSION

ARSA is a common vascular anomaly with significant impact on prenatal diagnosis and neonatal outcomes. This study evaluates the role of ARSA as a marker of chromosomal and structural abnormalities and compares iiARSA and niARSA cases. The results are consistent with the existing literature and provide insights into clinical practice and prenatal management strategies.

The prevalence of ARSA ranges between 0.5% and 2% in the general population and increases significantly in trisomy 21 cases, with studies reporting rates of 23–30% (2,3). Recent meta-analyzes show that isolated ARSA has no significant association with chromosomal abnormalities, with a likelihood ratio (LR+) of almost zero. However, in niARSA the LR+ increases dramatically to 199 when additional markers are present, underlining the diagnostic relevance (2,3).

In this study, niARSA cases had a higher prevalence of chromosomal abnormalities, particularly trisomy 21 and 22q11.2 deletion syndromes, which is consistent with other reports. Previous studies have shown that 22q11.2 deletion syndrome, which is associated with congenital heart defects such as truncus arteriosus, is significantly correlated with ARSA (1,2). In addition, ventricular septal defects and other cardiac malformations are the most common structural anomalies in niARSA cases (6).

Neonatal outcomes in iARSA cases were predominantly favorable, which is consistent with previous literature stating that isolated ARSA rarely leads to significant complications (7,8). In our study, 94% of iARSA neonates were discharged without requiring a prolonged stay in the NICU, emphasizing the limited clinical impact of this condition in the absence of additional abnormalities. However, the niARSA cases had a markedly different profile. Adverse neonatal outcomes such as RDS, low birth weight and prolonged ICU stays were significantly more common in this group. Structural anomalies, including ventricular septal defects (VSD) and other congenital heart defects, contributed to the increased morbidity and in some cases required early surgical intervention (1,6,9). This discrepancy underscores the importance of distinguishing between iARSA and niARSA prenatally to effectively tailor postnatal care strategies.

The use of NIPT in pregnancies with ARSA is a critical factor. In iARSA cases, NIPT has been shown to be a highly reliable tool for ruling out chromosomal abnormalities, obviating the need for invasive procedures such as amniocentesis or chorionic villus sampling (7-9). This is consistent with the findings that isolated ARSA has a minimal association with chromosomal abnormalities, making NIPT an efficient diagnostic option of first choice. In contrast, invasive testing remains essential for niARSA, especially when additional sonographic markers or structural abnormalities are present (1,2,8). Studies suggest that invasive procedures provide important insights into genetic and structural risk factors that support prenatal counseling and delivery planning (3,8).

The results confirm that ARSA, especially niARSA, should be considered as a marker of high clinical relevance in prenatal medicine. The strong correlation between niARSA and chromosomal abnormalities requires a detailed evaluation of coexisting sonographic markers and structural defects as well as other anomalies (3,8,10).

Although this study provides a robust analysis of ARSA, its retrospective design and single-center nature may limit its generalizability. Future multicenter, prospective studies are needed to validate these results and examine the long-term neonatal outcomes associated with ARSA (2,11).

CONCLUSION

ARSA is a valuable prenatal marker, with isolated cases generally associated with favorable outcomes and non-isolated cases associated with significant chromosomal and structural abnormalities. The distinction between iARSA and niARSA is crucial for appropriate prenatal screening and neonatal care. While non-invasive prenatal testing effectively rules out chromosomal abnormalities in iARSA, invasive methods remain essential in niARSA to ensure a comprehensive evaluation. The role of ARSA in prenatal diagnosis underscores its importance for personalized and multidisciplinary fetal care.

Financial disclosures: *The authors declared that this study has received no financial support.*

Conflict of interest: *The authors have no conflicts of interest to declare.*

Ethical approval: *All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional research committee at which the studies were conducted (Clinical Research Ethics Committee of Ankara Etlik City Hospital No. 1 (Decision No.: AEŞH-EK-2024-001, date: 31/01/2024) and with the 2013 Helsinki declaration and its later amendments or comparable ethical standards.*

REFERENCES

1. Annetta R, Nisbet D, O'Mahony E, Palma-Dias R. Aberrant right subclavian artery: embryology, prenatal diagnosis and clinical significance. Ultrasound. 2022;30:284-91.
2. Luo T, Liu S, Ran S, et al. Associated congenital anomalies and genetic anomalies in fetuses with isolated and non-isolated aberrant right subclavian artery. J Matern Fetal Neonatal Med. 2023;36:2211705.
3. Saraç T, Erzincan SG, Uygur L, et al. Isolated aberrant right subclavian artery: should invasive intervention be recommended in the era of noninvasive prenatal tests?. J Ist Faculty Med. 2023;86:37-43.
4. Morlando M, Morelli C, Del Gaizo F, et al. Aberrant right subclavian artery: the association with chromosomal defects and the related post-natal outcomes in a third level referral centre. J Obstet Gynaecol. 2022;42:239-43.
5. Aygün EG, Sarı U, Pata Ö, Dilek TUK. Isolated aberran subclavian artery diagnosed in the second trimester examination: how should we approach? Dicle Med J. 2022;49:119-24.
6. Scala C, Leone Roberti Maggiore U, Candiani M, et al. Aberrant right subclavian artery in fetuses with Down syndrome: a systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2015;46:266-76.
7. Borenstein M, Minekawa R, Zidere V, et al. Aberrant right subclavian artery at 16 to 23 + 6 weeks of gestation: a marker for chromosomal abnormality. Ultrasound Obstet Gynecol. 2010;36:548-52.
8. De León-Luis J, Gámez F, Bravo C, et al. Second-trimester fetal aberrant right subclavian artery: original study, systematic review and meta-analysis of performance in detection of Down syndrome. Ultrasound Obstet Gynecol. 2014;44:147-53.
9. Nedelcu AH, Lupu A, Moraru MC, et al. Morphological aspects of the aberrant right subclavian artery-a systematic review of the literature. J Pers Med. 2024;14:335.
10. Özkan S, Aksan A, Fıratlıgil FB, et al. Persistent right umbilical vein: clinical outcomes and prognostic factors in prenatal diagnosis. South Clin Istanb Eurasia. 2024;35:359-63.
11. Ranzini AC, Hyman F, Jamaer E, van Mieghem T. Aberrant right subclavian artery: correlation between fetal and neonatal abnormalities and abnormal genetic screening or testing. J Ultrasound Med. 2017;36:785-90.