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# Isolated Aberrant Right Subclavian Artery: Does It Increase the Risk of Aneuploidy?

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## Objective

Aortic arch anomalies constitute approximately 1% to 3% of congenital heart diseases, aberrant right subclavian artery (ARSA) being the most common. The prevalence of ARSA is reported as 23.6% in Down syndrome, and 1.02% in euploid fetuses. We presented the prevalence of ARSA, its relationship with cardiac and extracardiac anomalies, karyotype analysis results and pregnancy results in our center during the 2-year period.

## Methods

We retrospectively investigated women who had ARSA, in a population of women which were applied to our center due to fetal cardiac anomaly or with a suspected fetal anomaly. The study involved the period between January 2020 and January 2022. Fetal cardiac examination was performed during the 11-14$^{th}$ weeks and 18-23$^{th}$ weeks scan, at the axial plane investigating principally the three vessels and trachea view, while obtaining also the longitudinal and coronal planes by using color Doppler. For the detection of ARSA, color Doppler speed settings were set to 15-30 cm/sec. In Figure 1, there are ultrasound images of a case with ARSA at the 21$^{st}$ gestational week.

## Results

During the study period, a total of 4720 pregnant women were applied to our unit for the first and second trimester detailed fetal ultrasound examination. During the study period, Down syndrome was diagnosed in 21, and ARSA was detected in 34 women (0.72%). The mean±SD respective maternal age and gestational age of women with ARSA were 30.2±5.7 years and 20.4±3.3 weeks. ARSA was an isolated finding in 17 cases (50%). Extracardiac malformations were observed in 17 cases (47%) (three <15 weeks). Chorionic villus biopsy was performed in one and amniocentesis was performed in ten patients. Amniocentesis was performed in 3 of 17 patients with isolated ARSA and the karyotype was found to be normal. Trisomy 21 was found in 3 of 34 ARSA cases (8.8%). In cases with Trisomy 21, ARSA was accompanied by hypoplastic nasal bone, hyperechogenic cardiac focus plus hyperechoic bowel, and short femur. Since ARSA can be associated with Down syndrome or other congenital anomalies, prenatal diagnosis is important. ARSA is observed in approximately 3% of people with a congenital heart anomaly and in 0.1% of the general population without a heart defect. In our cohort, we did not detect ARSA accompanying any major cardiac anomaly, but we found intracardiac echogenic focus in 5 cases, ventricular septal defect in one, and cardiac arrhythmia in one patient. Previous studies have analyzed the frequency of fetal ARSA in chromosomally normal fetuses and fetuses with Down syndrome and identified its benefit as an ultrasound marker for Down syndrome in the second trimester. In fetuses with Down syndrome, the prevalence of ARSA was reported between 8% and 37%. We observed ARSA in 3 out of 21 fetuses with Down syndrome (14.2%). However, Down syndrome was not detected in cases with isolated ARSA. Although there is insufficient evidence for prenatal invasive diagnostic testing in cases with ARSA and low risk for trisomy in screening tests, detailed anatomical and cardiac screening should be performed to investigate additional markers for trisomy 21 and other possible anomalies such as 22q11 microdeletions.

## Conclusion

When ARSA is detected, fetal echocardiography and a detailed ultrasound scan for markers of trisomy 21 and other aneuploidies should be performed. In the presence of isolated ARSA, the probability of aneuploidy is low, but diagnostic testing for chromosomal anomalies should be recommended if accompanying anomalies existed.