Aberrant right subclavian artery: Correlation with neonatal and chromosomal abnormalities

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Objective
Variations in the anatomical course of the right subclavian artery are found in 1-2% of healthy individuals postnatally. In general, an aberrant right subclavian artery (ARSA) is a benign finding. However, ARSA is considered a very strong marker for trisomy 21 and may be associated with deletions in the DiGeorge spectrum. We reviewed neonatal outcomes of fetuses diagnosed with ARSA.

Methods
A retrospective chart review of all fetuses with antenatally diagnosed ARSA was performed. We collected data from fetal anatomy ultrasound scans, fetal echocardiograms, genetic studies (karyotype, arrays or cell free fetal DNA cfDNA screens, neonatal records and pediatric cardiac records. Fetuses were excluded if they delivered outside our hospitals without karyotype or cfDNA screening.

Results
Seventy-nine fetuses with ARSA were identified at an average gestational age of 20.1 weeks. Three were excluded. Thirty-eight fetuses underwent cfDNA or amniocentesis for chromosomal evaluation. Of those, 7 had trisomy 21. An additional fetus likely had trisomy 21 (age 42, absent nasal bone, thick nuchal fold, ARSA and VSD) but the mother declined invasive testing. Three additional fetuses had karyotype abnormalities: 8p23.1 (IUGR, small aortic valve, hypoplastic aortic arch), 46XX, 4p- (IUGR, VSD, persistent LSVC, small thymus), and 5p15.2p15.1(14, 444, 119-19, 375, 547)x1 deNovo (echogenic intracardiac focus). There were 3 spontaneous fetal deaths (2 with trisomy 21 and 1 with Wolf-Hirshhorn syndrome). Nine pregnancies were terminated (trisomy 21-5, deNovo deletion-1, retinoic acid embryopathy-1, VATER-1, arachnoid cyst-1). One neonate died due to hypoplastic left heart syndrome. Of fetuses diagnosed with trisomy 21, all had additional major abnormalities. No neonate was found or suspected to have DiGeorge spectrum by FISH, SNP array or neonatal examination. The ARSA was isolated in 50 fetuses. All of these fetuses had unremarkable neonatal outcomes, and none was readmitted in the six months after discharge.

Conclusion
As an isolated finding, ARSA is benign. All fetuses with trisomy 21 had additional findings on ultrasound consistent with this diagnosis, and all fetuses with less common karyotype anomalies had additional ultrasound findings.