Relevance of detection of isolated aberrant right subclavian artery (i-ARSA) in a low risk screened population.

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Objective:
In fetuses diagnosed with i-ARSA and no extra-cardiac anomalies, we aimed to establish the frequency of chromosomal anomalies diagnosed with single nucleotide polymorphism microarray analysis, particularly focusing on microduplications or micro deletions which would have gone undetected by conventional karyotyping or cfDNA screening.

Methods:
Retrospective study of fetal ultrasounds with ARSA as an isolated finding between 2012 and 2017 in an Australian tertiary referral centre. Outcomes of interest were survival and frequency of chromosomal anomalies.

Results
- 63 cases of ARSA, 45 low risk cases i-ARSA
- For the 45 i-ARSA
  - 38 i-ARSA without complications
  - 2 i-ARSA with symptomatic oesophageal compression
  - 1 i-ARSA postnatally diagnosed ASD
  - 4 i-ARSA with abnormal genetic results *

*includes IUFD at 38weeks, diagnosed post-mortem with Trisomy 21, in woman 24yrs of age who declined amniocentesis

Conclusion
In a population that has had low risk first trimester screening, the subsequent finding of ARSA should still trigger the following steps:
1. Detailed review of fetal morphology including fetal echocardiography.
2. Invasive testing for chromosomal microarray analysis should be offered.
3. Postnatal follow-up with should be organised.

Despite the relatively low risk of aneuploidies expected in this setting, our data shows a positive association between ARSA and underlying genetic pathology and postnatal complications.