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Aberrant right subclavian artery: chromosomal aberrations and the cell-free DNA

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Objective

To evaluate the outcome of fetuses with isolated Aberrant Right Subclavian Artery (ARSA) prenatally detected and the rate of association with chromosomal aberrations and to investigate the role of the cell-free DNA in the prenatal management.

Methods

This was a retrospective study including all pregnancies with prenatal ultrasound detection of ARSA at first and mid-trimester scans. In our institution, following the diagnosis of ARSA, all pregnancies underwent a detailed ultrasound assessment, including a dedicated cardiac examination within 16 weeks, at 20-21 and 30-34 weeks of gestation. In all cases fetal invasive testing for karyotype and chromosomal microarray was discussed and offered regardless of the combined risk for aneuploidies. The option of cell -free DNA testing for trisomy 21, 18 and 13 was also mentioned and discussed. Information on maternal characteristics, antenatal course and perinatal outcome were collected by reviewing our database, medical records and neonatal charts. We excluded from the study all cases when ARSA was found in association with other structural anomalies. Cases lost at follow-up or with unknown outcome were also excluded from the study.

Results

From 2015 to 2022, 340 cases of ARSA were detected within the second trimester of pregnancy. Ninety cases were excluded from the analysis: 88 cases in which ARSA was associated to other additional abnormalities and 2 cases because were lost to the follow-up. In 250 cases ARSA was an isolated finding throughout the pregnancy. During the follow up examinations, fetuses with isolated ARSA were found to have normal growth and normal gestational development. Parents declined further genetic investigations in 78 cases (31.2%). In 101 (40.4 %) cases an invasive test was performed: only in one fetus from this group a pathogenic copy number variation (CNV) was detected (0.99%). In 71 fetuses (28.4%) with isolated ARSA parents otherwise opted for cel-free DNA testing, which resulted in low risk for aneuploidies in 100% of cases. No significant difference was observed between the two groups regarding chromosomal aberrations ($p = 0.762$).

Conclusion

This study confirmed that fetuses with isolated ARSA have a low probability of being diagnosed with chromosomal aberrations and the overall associated outcome is good. Cell-free DNA testing seems to be a useful option that should be discussed with the parents for the improvement of the prenatal management.