Detection rate of trisomy 21 in fetuses with isolated aberrant right subclavian artery

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Objective

To evaluate the frequency of chromosomal abnormalities among the fetuses with isolated aberrant right subclavian artery (ARSA) diagnosed by prenatal ultrasonography.

Methods

Retrospective study conducted between January 2008 and December 2012 at three different maternal fetal medicine units: Istanbul University Faculty of Medicine, Istanbul Prenatal and Kanuni sultan Suleyman Research and Teaching Hospital. For each pregnancy with a diagnosis of ARSA a detailed ultrasonographic examination was performed in order to detect additional findings, and an invasive prenatal diagnostic test for karyotype analysis was proposed.

Results

Over the study period a total of 104 fetuses with isolated ARSA were examined. Six of them were excluded, since they were lost to follow-up and their karyotypes were unknown. Among the 98 cases, median gestational age at diagnosis was 21 (16-30) weeks' and the median maternal age was 32 (22-50) years. In five pregnancies karyotype analysis revealed trisomy 21, and in one pregnancy in which parents did not accept the invasive prenatal diagnosis, trisomy 21 was diagnosed postnatally. The frequency of trisomy 21 was found to be 6. 1%.

Conclusion

ARSA is an important sonographic marker for trisomy 21 and karyotype analysis might be considered even in the absence of associated findings.