Chromosomal microarray analysis in foetuses with aberrant right subclavian artery
Idit Maya, MD,1,*, Sarit Kahana, PhD,1 Josepha Yeshaya, PhD,1 Tamar Tenne, PhD,2 Shiri Yacobson, MS,1 Iltaf Agmon-Fishman, MSc,1 Lital Cohen-Vig, MD,3 Alex Levi, MD,4 Eyal Reinstein, MD, PhD2,8 Lina Basel-Vanagaite, MD, PhD1,5,6,8 Reuven Sharony, MD2,7,8
1The Raphael Recanati Genetics Institute, Rabin Medical Center, Beilinson, Campus, Petah Tikva, Israel, 2The Genetics Institute, Meir Medical Center, Kfar Saba, Israel, 3Schneider Children’s Medical Center of Israel, Petah Tikva, Israel, 4Department of Cardiology, Meir Medical Center, Kfar Saba, Israel, 5Pediatric Genetics Unit, Schneider Children’s Medical Center of Israel, Petah Tikva, Israel, 6Felsenstein Medical Research Center, Rabin Medical Center, Petah Tikva, Israel, 7Department of Obstetrics and Gynecology, Meir Medical Center, Kfar Saba, Israel, 8Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv, Israel

Objectives: To evaluate the correlation between aberrant right subclavian artery (ARSA), with or without other ultrasound abnormalities, and risk factors for aneuploidies and chromosomal microarray analysis (CMA) results. Methods and materials: This was a multicentre study. Genetic analyses of foetuses diagnosed with ARSA were evaluated by CMA in the same laboratory. The clinical investigation included nuchal translucency, first and second trimester maternal serum screening, early and late second trimester foetal anatomic scans and foetal echocardiogram. Comparative Genomic Hybridization (CGH) Microarray analysis or Single Nucleotide Polymorphism (SNP) Array technology was used for CMA. Results: CMA results were available for 63 foetuses diagnosed with ARSA. No pathogenic variants were found among 36 foetuses with ARSA as an isolated finding. Additional ultrasound findings and/or risk factors for aneuploidies were present in 27 foetuses, of which 5 had pathogenic CMA results. Trisomy 21 (T21) was detected in a foetus with eugenic intracardiac focus (EIF; 22q11 deletion) in which targeted copy number analysis (CNA) showed no pathogenic findings. Additional single major anomalies were found in 19 foetuses, with one patient having a 22q11 deletion and two patients with trisomy 18. Conclusion: CMA was efficient in detecting additional aneuoploidies. Our study demonstrated that additional ultrasound abnormalities or risk factors for aneuploidies should be considered when ARSA is diagnosed.