Combined use of cytogenetic and SNP array in prenatal diagnostics of chromosomal abnormalities

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Objective
To explore the genetic etiology of fetal abnormalities, detected by prenatal ultrasound, through single nucleotide polymorphism (SNP array) analysis.

Methods
979 fetuses were tested with SNP array and conventional karyotyping. Complex copy number variations (CNVs) were verified with fluorescence in situ hybridization (FISH), multiplex ligation - dependent probe amplification (MLPA) and quantitative fluorescence polymerase chain reaction (QF - PCR).

Results
For the 979 cases, the diagnostic yields of conventional karyotyping and SNP assay were 7.3% (72/979) and 9.3% (91/979), respectively. Most common observed pathogenic CNVs were founded in fetuses with central nervous system (11.6%, 5/43), cardiovascular system (10.8%, 23/212) and multiple systems defects (10.8%, 15/139), with intrauterine growth retardation (8.8%, 6/68 each) and with facial dysmorphism (5.4%, 3/55). No pathogenic CNVs were detected among those with abnormalities of the amniotic fluid, skeletal system, respiratory system or prenatal serum screening.

Conclusion
Compared with conventional cytogenetic genomics, SNP array analysis provides significantly improved detection of submicroscopic genomic aberrations in pregnancies. Based on these results, we propose that genomic SNP array is an effective method, which could be used in the prenatal diagnostic testing, in order to assist genetic counselling in pregnancies.