Chromosomal microarray analysis on fetuses with ultrasound anomalies
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Objective
To investigate the correlation between the abnormal ultrasonic images and detection rates by chromosomal microarray analysis (CMA), which could give evidence for clinical application of CMA in prenatal diagnosis.

Methods
Retrospective analysis on results of CMA and conventional karyotyping performed on 1274 fetuses with abnormal ultrasonic imagings (including major structure and non-structure anomalies). And to analysis the relationship between various sonographic abnormalities and chromosomal anomalies.

Results
Chromosome abnormalities were observed in 176/1274 cases (13.8%), and 63 (4.95%) of which identified clinically significant copy number variations, 25 (1.96%) were identified unclear clinical significance. 14/38 (36.84%) and (17/48) 35.42% were observed chromosome abnormalities in multiple ultrasound structural anomalies coupled with non-ultrasound structural anomalies and multiple ultrasound structural anomalies respectively, including 2.63% and 8.33% clinically significant copy number variations. 0~15% were identified chromosome abnormalities in single ultrasound structural anomaly in different anatomy system, (0~3.51% were clinically significant copy number variation) and 6.9~13% were identified chromosome abnormalities in single ultrasound non-structural anomaly in different anatomy system, (0.21~3.45% were clinically significant copy number variation).

Conclusion
This study in single center indicates that 13.8% of fetuses with ultrasound anomaly were observed chromosome abnormalities, 4.95% restricted to clinically significant copy number variations, even up to 35.42% were identified chromosome abnormalities in multiples, 8.33% restricted to clinically significant copy number variations, which could provides information for fetal prognosis. Therefore, we conclude that microarray has considerable diagnostic and prognostic value in fetuses with ultrasound anomalies.