Objective
To study the value of using chromosomal microarray analysis (CMA) in prenatal diagnosis to detect chromosomal abnormalities in high risk of the first trimester combined screening and ultrasonography anomalies.

Methods
From January 2013 to December 2015, we included 107 chorionic villus sampling sample from consecutive ongoing gestation, with combine test risk >1/270, fetal NT>3.5mm, NB(-), DV(+), TR(+) and structure abnormalities at 11-13 weeks' gestation, from a regional hospital. Subsequently, 93 amniocentesis sampling was performed when the structure abnormalities finding by ultrasonography scans at 18-22 weeks. Both of G-banding karyotype and CMA are parallel performed. DNA was extracted directed from the samples and examination with 80kb oligonucleotide array-based comparative genomic hybridization (n=200).

Results
In 200 cases, 12 cases of aneuploidies for 21, 18, 13, X or triploidy detected both of traditional G-banding karyotype test and CMA. Among 188 cases with normal karyotype, we detect pathologic copy number variants (CNVs) by CMA in 23(11.5%) fetuses including 13(6.5%) de novo and 7(3.5%) heritages. Variant of unknown significance (VOUS) were seen in 3 cases.

Conclusion
CMA is more an effective first tier than G-banding karyotype test in prenatal regime for the ultrasonography positively finding of fetal structural abnormalities and high risk of combined test in first trimester screening. The CMA is effect detection for the prenatal genomic microdeletion and microduplication.