Fetal karyotyping and CMA indicated for late onset abnormal sonographic findings are they really necessary?
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Objective
Late onset abnormal sonographic findings incorporate a cluster of cases characterized by normal biochemical and sonographic screening at first and second trimester up to 23 gestational weeks and “developing” abnormal findings later on. We aimed to analyze genetic testing results, including karyotype and chromosomal microarray analysis (CMA) for this unique group of patients.

Methods
We conducted a retrospective cohort analysis of all women who underwent amniocentesis, indicated for late appearing abnormal sonographic findings, from January 2012 to December 2015 at or beyond 24 weeks of gestation.

Results
Overall, 103 fetuses underwent amniocentesis, due to late onset genetic indications (Figure 1). The indications were: 1) Fetal abnormal sonographic findings: single finding, multiple findings and those accompanied by growth restriction or polyhydramnios; 2) Isolated FGR; 3) Isolated polyhydramnios. All 103 fetuses had normal karyotype. 95 women in the study population performed CMA, of whom 5 had clinical pathogenic result. The detection rate of abnormal CMA among our study population was 5.26%(5/95).

Conclusion
If amniocentesis is performed for late onset abnormal sonographic findings, CMA should be routinely applied as part of the genetic analysis: adds 5.26% to the Detection Rate-pathogenic CNV. Further, larger, studies are needed to elucidate the risk per specific findings.