Objective
To investigate the presence of copy number variation (CNV) using SNP array and determine which chromosomal region is changed in fetus with ventriculomegaly (VM).

Methods
Included on our study were 19 fetuses with VM diagnosed by prenatal ultrasound, normal karyotype studied by G band on fetal blood and negative PCR for congenital infections in the amniotic fluid. The DNA was extracted from fetal blood obtained by cordocentesis between 20 and 34 weeks and analyzed by SNP array. The CNVs found were compared to databases and literature and then classified in three groups: CNV pathogenic, uncertain clinical significance (VOUS) and benign.

Results
The mean atrial diameter at birth between the fetuses was 29.9mm (SD 15.1mm). Forty CNV were found and 15 loss of heterozygosis (LOH). VOUS was described for 16 CNV, and 23 CNV were benign. None of the CNV was pathogenic.

Conclusion
The present data suggest that SNP array analysis is a useful and relevant technique in prenatal diagnosis specially in ventriculomegaly, as we detected CNV in 94.7% of cases. Although, more studies are necessary to enrich the database and improve the prognosis of this complex disease.