Aim
To assess the clinical implication of chromosomal microarray analysis (CMA) and next-generation sequencing (NGS) in prenatal diagnosis of Congenital Anomalies of the Kidney and the Urinary Tract (CAKUT).

Methods
n=408 CAKUT
308 (75%) collecting system anomalies
43 (11%) renal dysplasias
57 (14%) number, fusion or location anomalies

164 (40%) invasive test (CVS/amnio) for CMA
(qGenomics(qChipCM,8x60K)

Results
17 genetic anomalies
4 VOUS

Collecting system 4 (24%)
Renal dysplasia 8 (47%)
Number, fusion or location 5 (29%)

- T21. Megacystis. TOP (2)
- 1.5Mb del 16q24.1q24.2.TOP (FOXF1)
- 800Kb del 22q11.21
- 1.2Mb del 17q12 (4)
- T13.TOP
- 47XYY (no array)
- Alagille syndrome (NOTHC2)
- Bardet-Biedl syndrome (TTC8)
- Steel sd. Horse-shoe kidney.TOP
- CHARGE.Horse-shoe kidney.TOP
- Del 4p16.3 crossed renal ectopia
- Del 3p26.2 unilateral renal agenesis
- Del 22q.11 unilateral renal agenesis+ARSA

12/17 chromosomal anomalies founded could not be diagnosed by conventional karyotype. CMA and NGS increased the detection rate of genetic disorders by 8% in our serie.

Conclusion
Chromosomal microarray analysis and next-generation sequencing are a valuable tool in the diagnosis and prenatal counseling of CAKUT, particularly in those cases that represent a diagnostic dilemma such as echogenic/dysplastic kidneys with normal amniotic fluid.