Copy number variations in multicystic dysplastic kidney: update for prenatal diagnosis and genetic counseling
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Objective
To assess the clinical implication of chromosomal microarray analysis (CMA) in prenatal diagnosis of MCDK.

Methods
Thirty-seven cases with MCDKs detected by prenatal ultrasound were enrolled in the study; 33 cases were isolated MCDKs and four cases were non-isolated MCDKs. CMA was performed on the Affymetrix CytoScan HD platform. The frequencies of the detected CNVs were compared with 461 cases underwent CMA for anomalies unrelated to congenital anomalies of kidney and urinary tract (CAKUT) or 124 healthy newborns as controls. All of the annotated CNVs were validated by MLPA or qPCR.

Results
Pathogenic CNVs were detected in 13.5% (5/37) of MCDKs. Two 17q12 deletions, one untypical 22q11.2 deletion, and one 22q11.2 duplication were detected in four isolated MCDK cases. Duplication of 1q31.3q44 was identified in a non-isolated MCDK case. Three of the five pathogenic CNVs were inherited. We also validated eight CNVs of uncertain significance only detected in MCDKs and five CNVs with higher frequency in MCDKs.

Conclusion
A substantial proportion of MCDKs were associated with pathogenic CNVs. Family members with the same CNV were asymptomatic or of different kind of renal malformations. It may be reasonable to perform CMA when MCDKs are identified prenatally.