Chromosomal disorders in fetal isolated echogenic kidneys

V. Borobio, M. Larroya, M. Perez-Cruz, E. Marimón, J. Fuenzalida, L. Rodríguez-Revenga, JM. Martínez, E. Gratacós
Department of Maternal-Fetal Medicine. BCNatal. Hospital Clinic-Sant Joan de Déu. University of Barcelona, Spain.

Aim
Fetal isolated echogenic kidneys with normal volume of amniotic fluid represents a dilemma in prenatal diagnosis.
The aim of this study is to assess the clinical implication of chromosomal microarrays analysis (CMA) in diagnosis of chromosomal disorders in fetal isolated echogenic kidneys.

Methods
We review 10 cases of isolated echogenic kidneys with normal volume of amniotic fluid (2015-2018).

Results
- 3 cases with normal array
- 1 case 47,XYY (no array)
- 4 cases (50%) 17q12 microdeletion (HNF1B)

*Three cases with abnormal appearance of kidneys in ultrasound. Normal renal function. Follow-up in Neuropediatrics and Nephro-Pediatrics Units.

Conclusions
A high proportion (63%) chromosomal anomalies are present in fetuses with isolated echogenic kidneys. Chromosomal microarray analysis strongly improves genetic disorders detection and should be offered in order to rule out 17q12 microdeletion syndrome, which has a high correlation with echogenic kidneys during fetal life. This syndrome is a cause of renal abnormalities with uncertain further renal function, maturity-onset diabetes of the young (MODY-5) and a wide spectrum of neurodevelopmental disorders. Early diagnosis of 17q12 microdeletion allows a closer follow-up of these children from birth.