

EP05.71. Juvenile Microcystic Kidney Disease in a Fetus with an Unclear Etiology: A Case report

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Introduction

Juvenile microcystic kidney disease is a rare congenital disorder characterized by abnormal renal development, leading to enlarged, hyperechoic kidneys with impaired function.

Case report

- A 27-week pregnant patient underwent an ultrasound examination, which revealed bilaterally enlarged, hyperechoic, horseshoe-shaped kidneys.
- Non-invasive prenatal testing indicated a low risk for the most common aneuploidies. Chromosomal microarray analysis revealed a normal female pattern, while whole-exome sequencing of both parents and the fetus failed to detect any pathogenic genetic variants.
- A follow-up ultrasound examination at 31 weeks of gestation, revealed additional findings:
 - Oligohydramnios
 - Cerebellar hypoplasia
 - Cavum septi pellucidi cyst
 - Intrauterine growth restriction
- Following approval of the Ethics Comittee feticide was performed and labor was induced.

Conclusion

Given the complexity of the observed fetal anomalies, whole-genome sequencing should be considered as an adjunctive diagnostic tool to enhance the precision of etiological assessment and improve the accuracy of prenatal counseling and management.

References:

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