IN UTERO DIAGNOSIS OF AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE

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Introduction

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common inherited renal disease. Diagnosis, which used to be made during adulthood, has now shifted towards earlier ages due to the development and widespread use of ultrasonography. Very-early onset (VEO) cases, diagnosed either in utero or within the first 18 months of life, are associated with adverse clinical outcomes, representing a particularly high-risk group of patients.

Clinical Case

We describe a case of prenatal diagnosis in a 35-year-old pregnant woman with ADPKD (mutation in PKD1) and a positive family history, diagnosed at the 21-week morphologic ultrasound. Diagnosis was based on the detection of bilaterally enlarged and hyperechogenic fetal kidneys with multiple cysts (figures 1 and 2). All other morphologic features were normal, including the renal corticomedullary

The couple was offered genetic counselling and opted for continuing pregnancy. Labour was induced at 37 weeks due to fetal growth restriction; vacuum-assisted vaginal delivery was performed due to labour dystocia and the baby was born with an Apgar score of 9/10.

Conclusion

ADPKD is a significant entity in childhood, with a wide clinical presentation spectrum. It may be indistinguishable from Autosomal Recessive PKD, and histological or genetic analysis may be necessary to differentiate them. This case shows how diagnosis of VEO-ADPKD through ultrasound may signal high-risk patients with poor prognosis that may benefit from early and timely treatment.