Ultrasound examination in week 32 showed anhydramnios and bilateral enlarged kidneys with hyperechoic pyramids, mimicking hypercalcinosis. The finding of small cystic lesions in the liver guided us to the presumptive diagnosis of ARPKD with bad prognosis. The primigravida had a spontaneous vaginal delivery of a liveborn boy at term who died 5 hours old due to hypoplastic lungs. Sequencing of the PKHD1 gene showed compound heterozygosity for a pathogenic mutation inherited from the mother and three VUS in cis inherited from the father, most likely explaining the patient’s phenotype. Autopsy showed changes pathognomonic for ARPKD.