# #1967 - Genetics of Autosomal Recessive Polycystic Kidney Disease: A case report with a novel mutation in the PKHD1 gene and a review of the literature


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**Introduction:** The majority of individuals with autosomal recessive polycystic kidney disease (ARPKD) present in the neonatal period with enlarged echogenic kidneys. Approximately 30% of affected infants die in the neonatal period or within the first year of life primarily of respiratory insufficiency or superimposed pulmonary infections. Fifteen-year survival is estimated to be 67%-79%, and may be improving. ARPKD occurs in about 1 in 20,000 live births among Caucasians. This corresponds to a carrier frequency of approximately 1:70 in non-isolated populations. The exact incidence is unknown since published studies vary in the cohorts of patients examined and some severely affected babies may die perinatally without a definitive diagnosis. Mutations in the PKHD1 gene are the primary cause of ARPKD; however, the disease is genetically not as homogeneous as long thought and mutations in several other genes can phenocopy ARPKD. Recently, mutations in DZIP1L have been described in patients with a moderate clinical course of ARPKD. Up to now, about 300 pathogenic mutations in PKHD1 gene and 20 mutations in DZIP1L gene have been reported. In this study, we review the available literature on ARPKD and present a family with an expired child affected by ARPKD and a novel mutation in PKHD1 gene.

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**Materials and Methods:** PubMed search performed using the terms "ARPKD" and "gene" in August 2018. For the asymptomatic father of expired affected child, Next Generation Sequencing was performed using Nimblegen chip capturing the genes of interest. Then direct sequencing of the region where the father had mutation in the PKHD1 gene was performed for child's mother.

**Result:** A possible pathogenic mutation (c.6469C>T, p.Gln2157Ter) on *PKHD1* (NM_138694) gene was detected in a heterozygous states in parents.

**Discussion and Conclusion:** Although there is no report that this mutation is ever identified in ARPKD patients, the early termination of amino acid production is expected to affect the protein's function as a possible pathogenic mutation.

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