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RECEIVED 31 July 2025

ACCEPTED 13 October 2025

PUBLISHED 03 November 2025

CITATION

Marquez J, Hawkins LM, Beck AE, Dipple KM,
Glass IA, Keefe AC and Nguyen ED (2025)
Case Report: An atypical case of ARPKD
highlights the utility and challenges of
implementing genetic testing in cystic kidney
disease.

Front. Pediatr. 13:1677417.

doi: 10.3389/fped.2025.1677417

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Case Report: An atypical case of ARPKD highlights the utility and challenges of implementing genetic testing in cystic kidney disease

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Background: Biallelic pathogenic variants in *PKHD1* cause a highly heterogeneous disease, predominantly involving the kidneys and liver. Although the correlation between genotype and phenotype remains unclear, many variants in this gene have been described.

Case: In this study, we describe a case of suspected autosomal recessive polycystic kidney disease (ARPKD) due to a novel variant in *PKHD1*. The patient in this instance presented with a novel *PKHD1* variant (c.2713C>A; p.Gln905Lys) in *trans* with a previously described pathogenic variant (c.7994T>C; p.Leu2665Pro).

Conclusions: The *PKHD1* variant c.2713C>A; p.Gln905Lys may contribute to an ARPKD phenotype with a delayed juvenile onset.

KEYWORDS

autosomal recessive polycystic kidney disease, exome sequencing, genetics, inherited kidney disease, cystic liver disease

Introduction

Autosomal recessive polycystic kidney disease (ARPKD) is a rare condition that arises due to biallelic pathogenic variants in a small subset of genes, the most common of which is *PKHD1*. *PKHD1* encodes the fibrocystin protein that participates in the development of kidney and liver tubular structures. Fibrocystin is understood to function through a variety of proposed roles that include acting as a transmembrane ciliary protein and signaling the release of C-terminal fragments (1–3). While the genetic etiology of ARPKD has long been appreciated, reports of this condition have predominantly focused on severe presentations during the perinatal period (4–6). As sequencing-based genetic testing has become more prevalent in the evaluation of individuals with kidney disease, a broader phenotypic spectrum of ARPKD, which has a later onset, has become apparent (7, 8). Indeed, juvenile- and adult-onset ARPKD has been reported and appears to follow a distinct clinical course with less severe kidney disease and

often a predominance of liver-related complications (9, 10). Yet, it remains unclear how different genetic variants may contribute to varying ARPKD phenotypes. In this study, we present the case of an individual with a clinical diagnosis of ARPKD, which is thought to have arisen due to compound heterozygous variants in *PKHD1* and includes a novel variant in this gene detected through exome sequencing (ES).

Case report

A 14-year-old XY boy initially presented to the nephrology clinic following an abdominal ultrasound that found multiple cortical kidney cysts and multiple hepatic cysts (Figures 1A,B). On initial laboratory evaluation, he was found to have abnormalities

consistent with chronic kidney disease (CKD). He had a serum creatinine level of 1.15 mg/dL, albumin of 4.7 g/dL, blood urea nitrogen of 26 mg/dL, potassium of 4.3 mmol/L, bicarbonate of 21 mmol/L, calcium of 9.9 mmol/L, and 1+ protein in his urine. He underwent magnetic resonance imaging (MRI) of the abdomen to further delineate the cystic structures (Figures 1C,D). His liver and kidney cysts had a maximal volume of 14,137 and 179 mm³, respectively. His history was significant with a previously identified small unspecified number of hepatic cysts found incidentally during ultrasound evaluation in the setting of a prior gastrointestinal illness at 8 months of age. His kidneys were not cystic during the prior imaging, and he had no dietary history that would contribute to decreased kidney function. Similarly, he had no history of severe or frequent infections that would increase his risk for decreased kidney function. He was not taking any

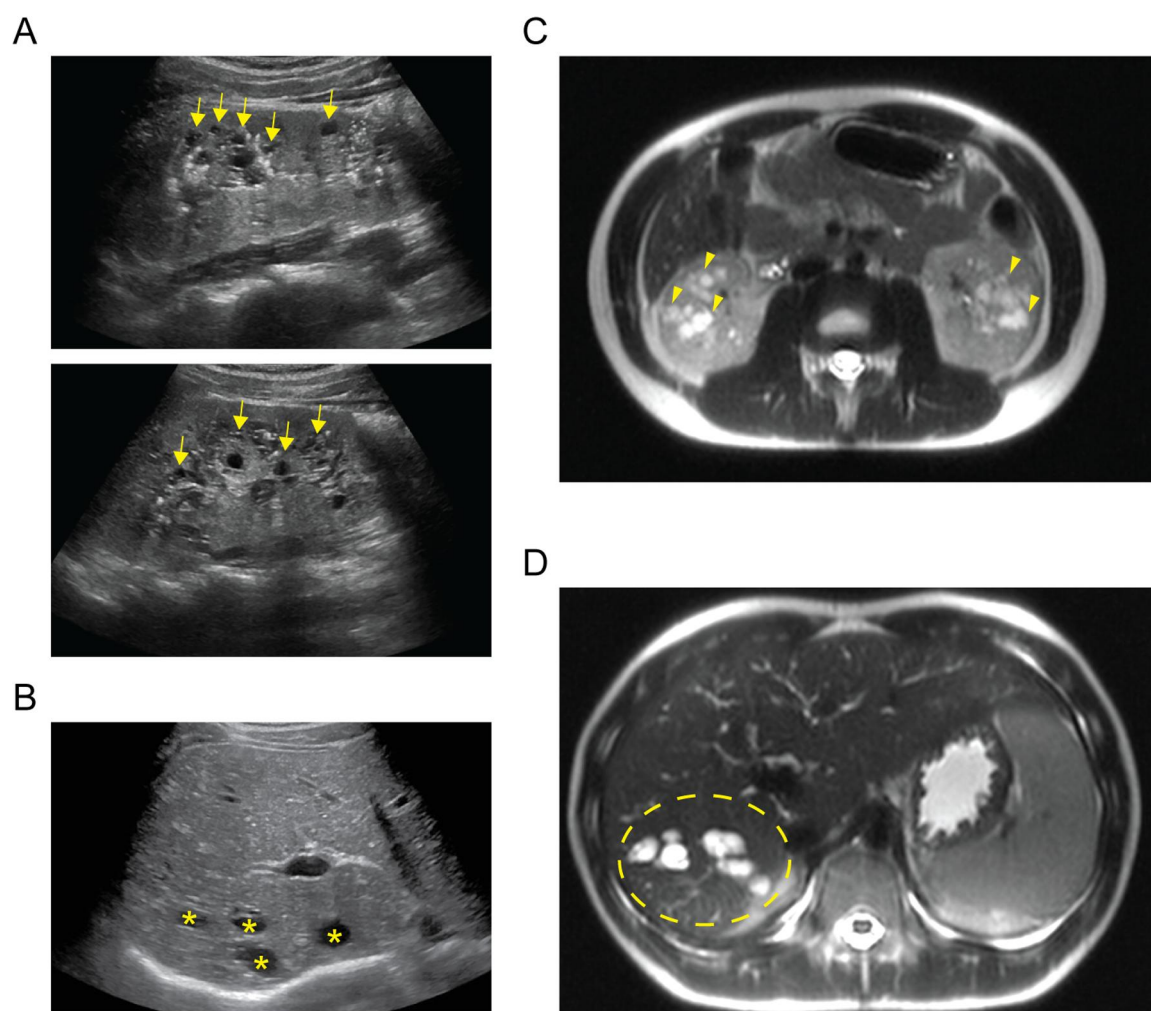


FIGURE 1

Imaging of cystic organ changes. (A) Sagittal ultrasound images of right (upper) and left (lower) kidneys demonstrating diffuse cystic changes predominantly affecting the medullary parenchyma (examples indicated with yellow arrows). (B) Transverse ultrasound image of the liver demonstrating cystic spaces of varying size affecting the hepatic parenchyma (examples indicated with yellow asterisks). (C) Axial T2-weighted MRI cross section demonstrating extensive kidney cystic changes bilaterally (examples indicated with yellow arrowheads). (D) Axial T2-weighted MRI cross section demonstrating extensive cystic changes within the right hepatic lobe (affected area within area demarcated by yellow dashed ellipse).

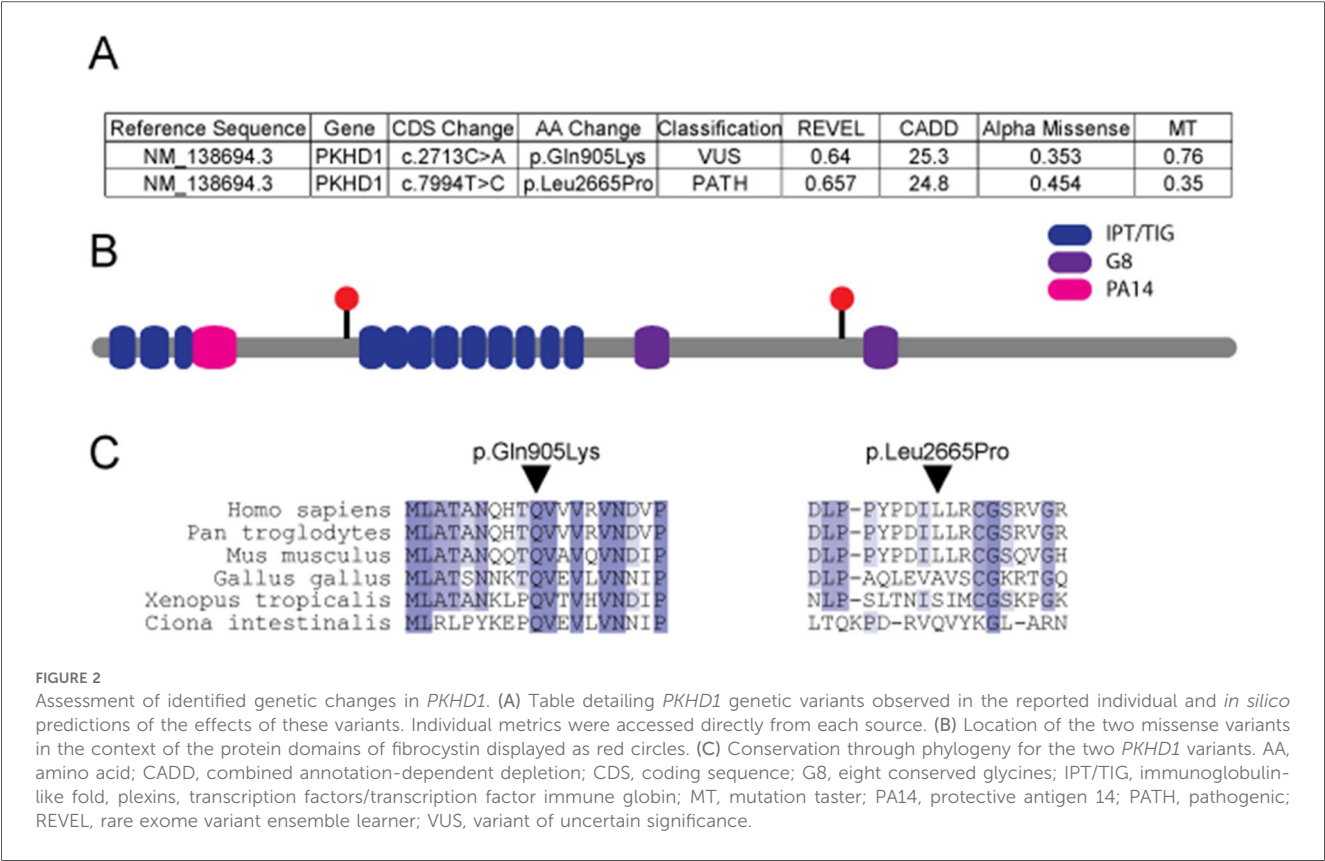
routine medication and had no history of exposure to known nephrotoxic agents.

Due to the presence of multi-organ cysts along with evidence of proteinuria, he was referred to the medical genetics department for further evaluation. His results did not show evidence of dysmorphic craniofacial features or of a well-described syndrome. His development had progressed typically following an uncomplicated pregnancy. His family history included notable episodes of hepatic cysts and hepatic hemangiomas in his father, which had been attributed to a history of hepatitis B infection. As a genetic cause of cystic hepatorenal disease was suspected, diagnostic testing in the form of clinical ES was performed, with concurrent copy number variant analysis. This test was carried out by a clinical laboratory improvement amendments (CLIA)-certified laboratory and included exon capture and sequencing of coding exons plus ±10 bp of flanking non-coding DNA for each exon. Exome sequencing when carried out in this manner, with analysis that considers the phenotype, can detect pathogenic variants in many of the potential causes of cystic kidney and/or cystic liver disease.

Analysis of sequencing results was completed with particular emphasis on those genes known to be associated with cystic kidney and/or liver disease. This testing identified compound heterozygous variants in *PKHD1* (Figure 2A). One of these was maternally inherited (NM_138694.3 *PKHD1* c.7994T>C; p.Leu2665Pro) and classified as pathogenic. The other variant was paternally inherited (NM_138694.3 *PKHD1* c.2713C>A; p.Gln905Lys) and classified as a variant of uncertain significance (VUS). The classifications were

completed by the clinical laboratory in accordance with American College of Medical Genetics (ACMG) guidelines (11). These variants were deposited in the ClinVar database under accession numbers SCV005339882.1 and SCV005363611.1. While the pathogenicity of the detected VUS remains unclear, given the patient’s clinical course and imaging findings, *PKHD1*-related autosomal recessive polycystic kidney disease (*PKHD1*-ARPKD) was deemed a likely diagnosis. Indeed, another variant at fibrocystin amino acid 905 has been classified as pathogenic. However, this is a truncating variant (Gln905Ter) previously identified in individuals with severe neonatal ARPKD (12, 13). The p.Gln905Lys missense variant is extremely rare in the population, and multiple *in silico* prediction tools further support the deleterious nature of the missense change for this VUS (Figure 2A). While some variants implicated in ARPKD, such as splice site variants (14), are amenable to testing to determine possible pathogenicity, missense variants such as the one we describe are not easily tested for effect on function. The amino acid position lies near the immunoglobulin-like fold, plexins, transcription factors/transcription factor immune globin (IPT/TIG) domains of fibrocystin that have an immunoglobulin-like fold where numerous missense variants putatively implicated in ARPKD phenotypes are found (Figure 2B). In addition, this amino acid position is evolutionarily conserved to the level of invertebrates (Figure 2C).

The patient’s treatment proceeded in a manner consistent with the surveillance and interventions recommended for ARPKD. His blood pressure was closely monitored and was predominantly within reference ranges for age, and his 24-h ambulatory blood



pressure monitoring was also within normal limits. His treatment included evaluation by hepatologists, along with ongoing care in both nephrology and genetics clinics. His kidney function remained relatively stable with modest improvement in proteinuria after initiation of an angiotensin-converting enzyme inhibitor, which he tolerated without a significant rise in creatinine. His hepatic function was thus far within reference laboratory ranges. However, this represented a limited window for clinical observation, and the long-term kidney and hepatic disease course for this patient remains uncertain.

Discussion

We identified biallelic *PKHD1* variants in an individual presenting with kidney dysfunction and cystic kidney and liver morphology in adolescence. This included one VUS in *trans* with a pathogenic variant. This individual's family history was suggestive of an autosomal dominant disorder based on his father's cystic liver finding, which nevertheless rendered this less straightforward due to the father's hepatitis history. Indeed, though autosomal dominant polycystic kidney disease (ADPKD) is much more common (15, 16), it follows a different disease course compared to ARPKD (17). This individual's imaging findings of medullary predominance of small kidney cysts and biliary cystic changes are less consistent with ADPKD presenting in childhood and more consistent with what has been described in ARPKD with later childhood and adult onset (18). While we considered whether his findings could be caused by monoallelic *PKHD1* variants, which have been previously described (19–21), this apparently distinct disorder typically includes increased medullary echogenicity of the kidney and multiple small liver cysts, which are inconsistent with the patient's radiological findings. We also considered whether another genetic cause could have remained undetected by the clinical exome sequencing approach used. Our knowledge of all potential genes in which variants might lead to cystic kidney disease is likely incomplete, and this may serve as a limitation to clinically identifying relevant genetic variants. However, many variants that give rise to cystic kidney and liver disease often also occur alongside other clinical manifestations. For instance, variants in genes such as *ALG8* or *ALG9* often result in a complex neurodevelopmental disorder of glycosylation (22, 23). As additional manifestations were not observed in this case, many other genetic causes of cystic tissue morphology were deemed less likely.

An individualized approach to treatment is beneficial in children and adolescents with ARPKD, and interventions should be tailored to the evidence of CKD progression, persistent hypertension, and/or complications from hepatic involvement, such as portal hypertension or cholangitis (24). Disease surveillance to detect these indications for further intervention should be multidisciplinary and tailored to disease severity. Appropriate measures typically include regular blood pressure monitoring, laboratory evaluations of kidney function and electrolytes, hepatic surveillance through both laboratory evaluations and imaging of the liver, and monitoring for psychosocial complications of disease

(25). Particular emphasis and increased frequency of evaluation may also be useful during puberty, as CKD has been described to accelerate during this physiological life stage, which could also be the case in the context of CKD due to ARPKD (26).

Even among large cohorts of children and adolescents diagnosed with ADPKD from high- and middle-income countries, only approximately half individuals or fewer have undergone genetic testing (27). The genetic etiology of cystogenesis in individuals with a clinical diagnosis of ADPKD may well range beyond variants in *PKD1* and *PKD2*. ARPKD is more frequently considered in the evaluation of perinatal hepatic and kidney disease, given the many reports of early-presenting ARPKD in the literature (28, 29). Indeed, the patient has a personal and family history that could have led to the clinical diagnosis of ADPKD, albeit with an earlier presentation than typical. However, genetic testing, despite a VUS in *trans* with a pathogenic variant in *PKHD1*, facilitated a more tailored approach to his treatment and likely a more accurate prognosis. Recent efforts have attempted to delineate the clinical course of ARPKD in young adults (30). Based on these data, it appears that individuals with ARPKD have a greater likelihood of stable native kidney function into adulthood despite varying levels of CKD compared to other diagnosed causes of cystic kidney disease, such as ADPKD. In addition, such individuals frequently develop fibrotic liver disease and resultant portal hypertension, necessitating multi-drug blood pressure management later in life. Despite these insights, even in larger cohorts of individuals with ARPKD diagnoses, fewer than half of the reported individuals have undergone genetic testing (30). This limits our ability to identify genotype–phenotype correlations that could further improve counseling related to ARPKD diagnoses.

Therefore, while genetic testing in the setting of cystic kidney disease certainly necessitates comprehensive genetic counseling tailored to each individual, as has been suggested by others (31), we recommend that the benefits of testing to achieve tailored care are considerable and should be more widely available. Nevertheless, we also acknowledge the challenges of uncertain results, such as seen in this case. While VUSs are often a component of genetic testing results, it seems likely that these may be an even more prevalent component of *PKHD1*-ARPKD-related testing for older children and adults, since missense variants do seem to yield a milder and later-onset phenotype that has not been as well captured in the literature thus far (28). Therefore, interpretation of testing may well rely on careful consideration of predicted molecular effects of a variant, taken along with the clinical presentation in the case of uncertain results, as illustrated here.

Conclusion

This case demonstrates a rare phenotype of suspected *PKHD1*-ARPKD manifesting in adolescence. We describe a novel variant in *PKHD1* that likely contributes to a later-onset liver-predominant phenotype with cystic kidney changes. Genetic testing is an important consideration for individuals with cystic kidney disease, as a suggestive or conclusive molecular diagnosis can greatly influence management.

Data availability statement

The original contributions presented in the study are included in the article/Supplementary Material, further inquiries can be directed to the corresponding authors.

Ethics statement

Written informed consent was obtained from the minor(s)' legal guardian/next of kin for the publication of any potentially identifiable images or data included in this article.

Author contributions

JM: Conceptualization, Data curation, Formal analysis, Investigation, Methodology, Writing – original draft, Writing – review & editing. LH: Formal analysis, Investigation, Writing – original draft, Writing – review & editing, Conceptualization. AB: Investigation, Writing – review & editing. KD: Investigation, Writing – review & editing. IG: Investigation, Writing – review & editing. AK: Investigation, Supervision, Writing – original draft, Writing – review & editing. EN: Investigation, Supervision, Writing – original draft, Writing – review & editing.

Funding

The author(s) declare that no financial support was received for the research and/or publication of this article.

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Acknowledgments

The authors thank the patients and families who are the inspiration for this report.

Conflict of interest

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