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A case report of atypical autosomal dominant polycystic kidney disease presenting as glomerulocystic kidney superimposed with thin basement membrane nephropathy

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Abstract

Background Autosomal dominant polycystic kidney disease (ADPKD) is the most frequent monogenic polycystic kidney disorder, but ADPKD presented as Glomerulocystic kidney (GCK) in adults is uncommon. Thin basement membrane nephropathy (TBMN) seems to account for major causes of familial hematuria and can coexist with other glomerular diseases. Here, we report a case of atypical manifestation of ADPKD presenting as GCK superimposed with TBMN in an adult man.

Case presentation A 40-year-old male presented with moderate proteinuria, microhematuria and renal insufficiency. He has no family history of kidney disease. Ultrasound revealed slightly enlarged kidneys with echogenic cortex. 2 to 3 visible small cortical cysts on both kidneys, and no anatomical abnormalities were detected by CT scan. Renal biopsy demonstrated that 33.3% (9/27) of the glomeruli had marked dilatation of Bowman's space. The glomerular cysts were lined by a simple layer of cuboidal epithelium, which was stained positive for Claudin-1 (parietal epithelial cell marker), but negative for LTA and DBA (tubular epithelial cell markers). There were foci of mild chronic interstitial fibrosis with few inflammatory infiltrates. Immunofluorescence stains were negative. Transmission Electron microscopy (TEM) revealed extensive glomerular basement membrane (GBM) thinning, without splitting or lamellation. The average thickness of GBM was 221 ± 25 nm. No electron dense deposits were identified by TEM. The next-generation sequencing indicated pathogenic heterozygous deletion of *PKD1* exon 3, and the mutation was determined to be a de novo mutation by familial variant analysis. No pathogenic mutations of *COL4A3*, *COL4A4*, *COL4A5*, *UMOD*, *TCF2* and *HNF1β* were identified.

Conclusion We report a rare case of atypical ADPKD presenting as GCK superimposed with TBMN. GCK is a rare disease and often overlooked. It is important for practicing nephrologists to have a clear understanding of GCK. GCK involves in various conditions, thus genetic analysis should be considered.

Keywords Autosomal dominant polycystic kidney disease, Glomerulocystic kidney, Thin basement membrane nephropathy, Next-generation sequencing analysis, Case report

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Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is the most common monogenic kidney disease, affecting 1/1000 individuals worldwide, and frequently progresses to end-stage renal disease (ESRD) by the sixth decade in life [1, 2]. ADPKD results from mutations in *PKD1*(in 85% of the cases) or *PKD2*, encoding polycystin-1 and polycystin-2 respectively [3]. However, the genetic test is not commonly used due to expensive costs and complexity. In addition, ADPKD presenting with atypical clinical symptoms and negative family history is still a diagnostic challenge to clinicians [4].

Glomerular cyst is defined as Bowman capsule dilation greater than 2 to 3 times normal glomeruli [5, 6]. While, glomerulocystic kidney (GCK) refers to a kidney with more than 5% glomeruli with cysts [6]. GCK can be classified further based on clinical presentation and associated multisystem disorders [7]. GCK often occurs in early childhood but rarely in adults, which makes it difficult to diagnose in adults.

Thin basement membrane nephropathy (TBMN) is characterized by extensive (>50% of the total glomerular surface area) thinning of the glomerular basement membrane (GBM) [8, 9]. TBMN can also coexist with other glomerular diseases in the diagnosis of renal biopsy [10]. Here, we report a case of atypical manifestation of ADPKD presenting as GCK superimposed with TBMN in an adult man.

Case presentation

A 40-year-old male was admitted to our division for foamy urine. He had neither a past medical history nor a family history of kidney diseases by laboratory findings and imaging. Physical examinations were as follows: body mass index (BMI) 20.4 kg/m^2, blood pressure 132/73 mmHg, normal respiratory sound, and abdominal tenderness. No hearing loss, skin rash, edema or joint pain was detected. Urinalysis showed proteinuria and microhematuria, and total protein excretion was 0.87 g/24hr. Serum creatinine was 103μmol/L, and eGFR was 81 ml/min/1.73m^2. Hematocrit was 37.7%, serum albumin was 44 g/L, fasting blood glucose and HbA1c were normal, and serology was negative. Laboratory findings were showed in Table 1.

Ultrasound revealed slightly enlarged kidneys (right, 125 × 51 mm, left, 120 × 60 mm) with echogenic cortex. CT scan showed 2 to 3 visible small cortical cysts on both kidneys. The biggest cyst was 28 × 29 mm (Fig. 1A). The number of cysts did not meet Ravine criteria [11]. Whereas, hydronephrosis or anatomical abnormalities were not detected and there were no cysts on the liver or pancreas. In addition, numerous small renal cysts were identified on magnetic resonance imaging after genetic diagnosis (Fig. 1B).

Renal biopsy

Percutaneous needle renal biopsy was performed. Immunofluorescence was negative for immunoglobulins or complements. 27 glomeruli were examined by light microscopy, of which 4 glomeruli were obsolescent global sclerosed, and 33.3% (9/27) of the glomeruli had marked dilatation of Bowman's space (Fig. 2A, and 2 C). The diameter of glomerular cysts varied from 608.7 μm to 961.0 μm (the average diameter, 784.9 μm). The glomerular cysts were lined by a simple cuboidal epithelium with periglomerular fibrosis (Fig. 2D). The glomerular tuft appeared normal. In addition, there was no significant cystic dilatation of tubules. There were foci of mild chronic interstitial fibrosis (Fig. 2B) with few inflammatory infiltrates (Fig. 2B). Arteries and arterioles were unremarkable. Transmission electron microscopy (TEM) revealed extensive glomerular basement membrane thinning, without splitting or lamellation (Fig. 2E). The average thickness of GBM was 221 ± 25 nm. Neither electron dense deposits nor extensive podocyte foot process effacement were identified by TEM. The structure and thickness of Bowman's capsule and parietal epithelial cells (PECs) appeared normal (Fig. 2F).

Genetic analysis

The differential diagnosis of TBMN and GCK in this patient included Alport syndrome, PKD, GCKD and syndromic GCKD. To examine whether there were any genetic mutations, the next-generation sequencing (NGS) of whole exome was performed by BGI (Shenzhen, China). NGS indicated pathogenic heterozygous deletion of *PKD1* exon 3 and heterozygous missense mutation of *NPHP1* (Table 2). No pathogenic mutations of *COL4A3*, *COL4A4*, *COL4A5*, *UMOD*, *TCF2* and *HNF1β* were identified. In addition, we performed familial segregation analysis. Both of his parents had normal renal function, negative urine test, and normal ultrasound results without any renal cysts. Both of parents' Sanger sequencing of the pathogenic variant of *PKD1* was negative. The mutation of *PKD1* was confirmed to be a de novo mutation. The patient is the only child in his family, and his grandparents passed away years ago. Moreover, the Sanger sequencing of *NPHP1* variants revealed that both the patient's father and his son carried the heterozygous missense mutation of *NPHP1.* And the son of the patient did not carry the mutation of *PKD1*.

Immunohistochemical staining

Renal tissue sections were immunostained for Claudin-1 (a PEC marker) (51-9000, Invitrogen), *Lotus tetragonolobus* agglutinin (LTA, a proximal tubular marker) (L32480, Invitrogen), and *Dolichos biflorus* agglutinin (DBA, a distal tubular marker) (L32474, Invitrogen). The immunostaining showed that the cells lining glomerular cysts were

Table 1 Laboratory findings

Laboratory Test	Admission Result	Reference values
White blood cell count	6.02×10^9/L	3.9-9.5 ×10^9/L
Hemoglobin	129	130–175 g/L
Hematocrit	37.7	40–50%
Platelets	237	125–350 ×10^9/L
Serum creatinine	103	57–97 μmol/L
eGFR	81	ml/min/1.73m^2
Iohexol GFR	83	ml/min
Uric acid	381	208–428 μmol/L
Asparate aminotransferase	27	9–50 U/L
Alanine aminotransferase	39	15–40 U/L
Glucose	4.8	3.9–6.1 mmol/L
Hemoglobin A_{1c}	5.4	3.9–6.2%
Potassium	3.62	3.5–5.3 mmol/L
Sodium	143	137–147 mmol/L
Calcium	2.2	2.11–2.52 mmol/L
Phosphorus	1.17	0.85–1.51 mmol/L
Bicarbonate	26.3	21–31 mmol/L
Triglyceride	1.85	< 1.7 mmol/L
Cholesterol	5.2	3-5.7 mmol/L
Low-density lipoprotein cholesterol	3.92	1.5–3.88 mmol/L
Complement C3	1.4	0.9–1.8 g/L
Complement C4	0.27	0.1–0.4 g/L
Anti-Nuclear antibody	Nonreactive	Nonreactive
Anti-dsDNA antibody	Nonreactive	Nonreactive
PR3-Antineutrophil cytoplamic antibody	Nonreactive	Nonreactive
MPO-Antineutrophil cytoplamic antibody	Nonreactive	Nonreactive
Hepatitis B surface antigen	Nonreactive	Nonreactive
Hepatitis B core antibody	Nonreactive	Nonreactive
Hepatitis C virus antibody	Nonreactive	Nonreactive
HIV antibody	Nonreactive	Nonreactive
Urinalysis		
Dipstick protein	+	Negative
RBC	4–5/HP	0–3/HPF
WBC	0/HP	0–5/HPF
Albumin-creatinine ratio	1057.6	< 30μg/mg
Total urine protein excretion	0.87	0.02–0.14 g/24 h
Urinary culture	Negative	Negative

Abbreviations: ds-DNA, double-stranded DNA, eGFR, estimated glomerular filtration rate, MPO, myeloperoxidase, PR3, proteinase 3, RBC, red blood cell, WBC, white blood cell

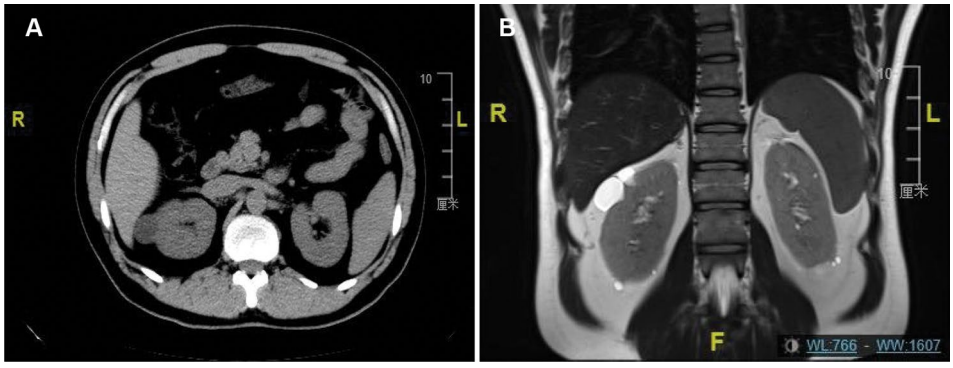

Fig. 1 Kidney imaging. (**A**) Computed tomography scan showed one cyst on right kidney. (**B**) Magnetic resonance imaging showed numerous small cysts on both kidneys besides the large cysts

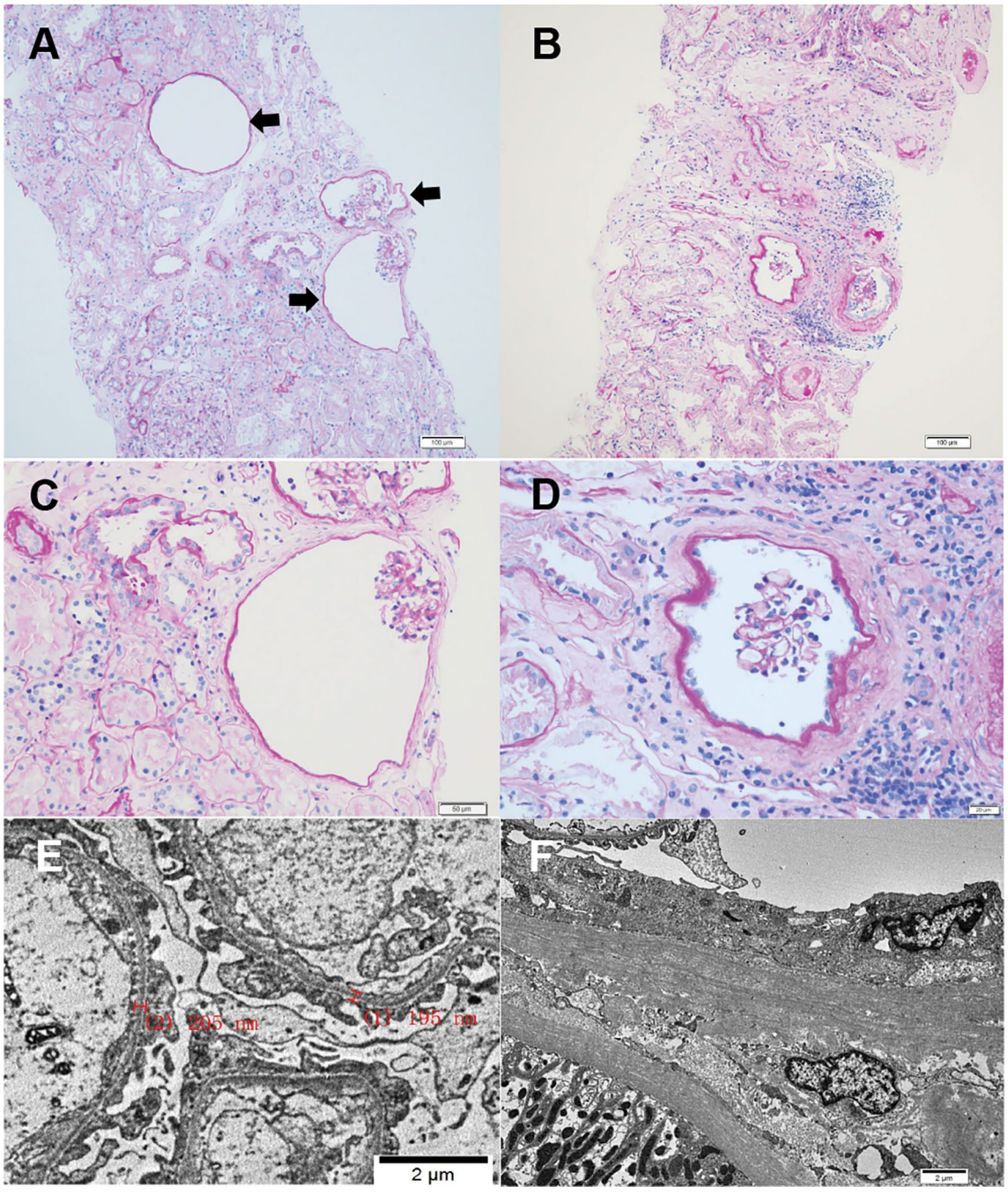

Fig. 2 Histological findings in renal biopsy. (**A**) Scatter dilatation of Bowman's capsule, the arrows indicate dilatation of Bowman's capsule. (PAS stain, Scale bar represents 100 μm) (**B**) Interstitial fibrosis. (PAS stain, scale bar represents 100 μm) (**C**) Dilatation of Bowman's capsule with normal glomerular tuft. (PAS stain, scale bar represents 50 μm). (**D**) Periglomerular fibrosis. (PAS stain, scale bar represents 20 μm). (**E**) Thinning of the glomerular basement membrane was showed by transmission electron microscopy, no electron dense deposits were found. (Scale bar represents 2 μm). E. The structure and thickness of Bowman's capsule and parietal epithelial cells appear normal by transmission electron microscopy. (Scale bar represents 2 μm)

Table 2 Summary of the pathogenic mutations

Gene	Genomic Location	Mutation Designation	Exon Region	Zygosity
PKD1	chr16:2169114–2,169,186	NM_001009944.2:EX3 Del	Exon 3	Heterozygote
NPHP1	chr2:110905572	NM_000272.3:c.1358G > T(p.Gly453Val)	EX13/CDS13	Heterozygote

Details of NGS sequencing: The NGS was performed on MGISEQ-2000 sequencing platform. ACMG criteria for the *PKD1* variant was PVS1_Moderate + PP4 as reported previously [12], ACMG criteria for *NPHP1* variant was PM2 + PP3 by SIFT, MutationTaster, Condel, and SpliceAI(1.3) softwares

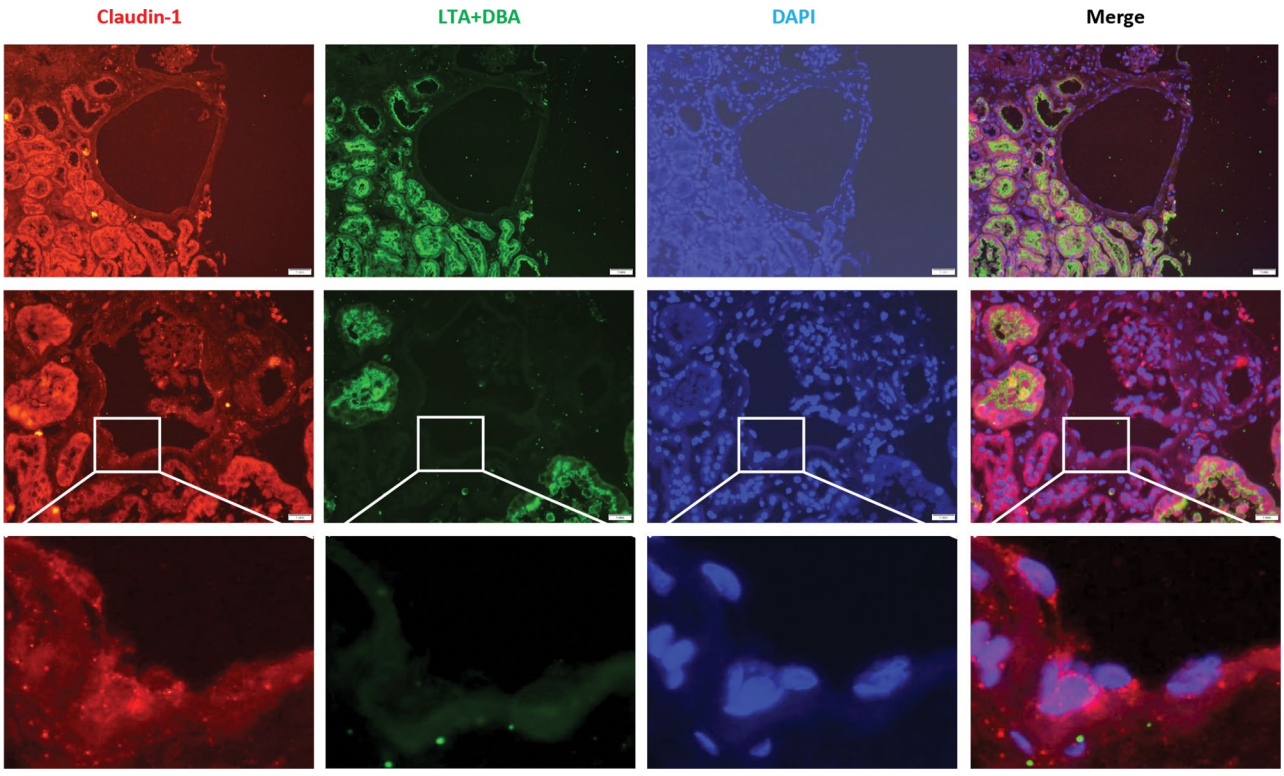

Fig. 3 Immunohistochemical staining showed glomerular origin of the cysts. Claudin-1(red), *Lotus tetragonolobus* agglutinin + *Dolichos biflorus* agglutinin (green), DAPI(blue). Original magnification ×20 in upper panel, ×40 in middle panel, and ×100 in bottom panel

positive with Claudin-1, and did not co-localized with LTA nor DBA (Fig. 3).

Discussion

The patient in this case report presented as TBMN superimposed with GCK. There are no standard diagnostic criteria for TBMN, because the reported mean thickness of GBM varies widely. Normal thickness is usually 373 ± 42 nm in adult males and 326 ± 45 nm in adult females [13, 14]. GBM thickness < 250 nm is the lower limit cutoff of diagnostic criteria in most of studies [8, 9, 15]. The average thickness of the GBM (221 nm) in this case met the diagnostic criteria of TBMN [8, 9]. TBMN is the most frequent cause of benign familial hematuria. About 40% of patients with TBMN carry mutations at type IV collagen α3 (*COL4A3*) or α4 (*COL4A4*) gene locus [16]. However, no pathogenic mutations of *COL4A3*, *COL4A4* and *COL4A5* were identified in our case. In addition, moderate proteinuria and mild renal insufficiency in this patient were atypical in TBMN. TBMN are also seen in early Alport syndrome (AS), the carrier state of X-linked Alport, and the carrier states of autosomal recessive Alport [8, 17, 18]. AS is usually manifested by increased proteinuria and progressive renal insufficiency. Hence, the diagnosis of AS was also considered in this case. But, the absence of GBM splitting and lamellation, anterior lenticonus, and hearing loss did not correspond with AS. Furthermore, gene analysis ruled out AS.

GCKD was brought to our attention because of the scatter ectasia of Bowman's space. GCK is not one disease, but an entity encompassing many conditions. Lennerz and colleagues divided GCK into five categories: type I, PKD presenting as a GCK variant of ADPKD/ARPKD; type II, hereditary GCK synonymous with GCKD; type III, syndromic GCK (GCK associated with heritable malformation syndromes); type IV, obstructive GCK; and type V, sporadic GCK (with 2 subcategories: ischemic GCK and drug-induced GCK) [7]. We did not find any indication for syndromic GCKD associated malformation indicative of tuberous sclerosis, Down syndrome or Zellweger syndrome. And obstructive GCKD was ruled out in this case due to the absence of radiology evidence of urinary tract obstruction. In addition, there was no past history of primary diseases which could cause sporadic GCKD including ischemic damage, such as systemic sclerosis, hemolytic uremic syndrome, or exposure to drugs, such as lithium. Furthermore, NGS analysis did not find mutations of *UMOD*, *TCF2*, *HNF1β* or other previously reported genes associated with hereditary GCKD.

Gene analysis confirmed *PKD1* mutation, although the patient had no positive family history of PKD. About 10–25% patients with ADPKD have no positive family

history [19]. *PKD1* is highly polymorphic and the type of the *PKD1* mutation is associated with the age of ESRD onset and disease severity, suggesting an allelic influence on ADPKD phenotype. Truncating mutations of *PKD1* and in-frame insertions/deletions are often associated with early onset and rapidly progression, while missense mutations generally with milder disease [20, 21]. Deletion of exons, which belongs to large re-arrangement, is not very common in *PKD1* mutations. About 4% of Consortium for Radiologic Imaging Study of PKD (CRISP) population was reported to have large re-arrangement [22, 23]. In addition, deletion of Exon 3 had been previously demonstrated in a Japanese ADPKD cohort [12].

Because TBMN occurs up to 5% in population [24], it is not uncommon that TBMN is recognized superimposed with other kidney diseases, such as IgA nephropathy, focal segmental glomerulosclerosis (FSGS), pauci-immune crescentic glomerulonephritis, mesangioproliferative glomerulonephritis, acute interstitial nephritis, lupus nephritis, and acute endocapillary glomerulonephritis [10]. But there was only one case reported that TBMN was superimposed with GCK in the literature. Recently, Hashimoto and his colleagues reported a case of TBMN accompanied by sporadic GCKD without mutation of *COL4A3*, *COL4A4*, *UMOD*, *MUC1*, and *SEC61A1*. However, in the case described by Hashimoto et al., the authors did not analyze whether the patient had pathological mutations in *PKD1*, *PKD2* or *PKHD1* [25].

Although renal cysts were easily identified on imaging in our case, the number of renal cysts was under the age-related ultrasound Ravine criteria of ADPKD [11]: the presence of four (two or more cysts in each kidney) for individuals aged 40–59 years with positive family history for cystic kidney diseases. There are no established imaging-based criteria for diagnosis of ADPKD in patients without a positive family history. A study showed that individuals who had 10 or more cysts in each kidney could be diagnosed ADPKD by imaging studies [26]. Ultrasound can detect the cysts with a diameter of 10 mm or greater, while MRI can detect cysts of 2–3 mm. After genetic test, we confirmed more cysts on both kidneys by MRI. Thus, MRI is more useful in diagnosis of polycystic kidney disease [27, 28].

ADPKD is a systemic disease, usually has extrarenal cysts in the liver or pancreas. And other extrarenal manifestations of ADPKD include hypertension, abdominal pain, cardiac valvular disease, cerebral aneurysms, and intestinal diverticulosis [29]. In this patient, we did not find any extrarenal cysts or manifestations. Cases of polycystic kidney diseases resembling GCK in neonates and children have been reported. In those cases, cysts of liver or other organs were not identified either [30, 31]. GCK in PKD usually occurs from early childhood, and we seldom encounter adult GCK in PKD, making it difficult to diagnose in the present case. Therefore, genetic tests were necessary in suspicious case of GCK.

Cysts in PKD originate in renal tubules or glomeruli, and are thought to occur due to enhanced proliferation of the epithelial cells lining the cysts [1]. In addition, the lack of tubular cysts in this case is not consistent with the typical histopathological features of ADPKD [7]. The immunostaining showed that dilatation of Bowman's capsule was not tubule-derived, and the ultrastructure of Bowman's capsule and PECs appeared normal by TEM in this case. But selective dilatation of the Bowman capsule still remains largely unexplained. Other cases reported that the stenosis at the glomerulotubular junction increased pressure of Bowman's space, and remodeling of the basement membrane of the Bowman's capsule might be the cause of glomerular cysts [32–34].

Heterozygous missense mutation of *NPHP1* was also detected in this case. But this heterozygous mutation is not pathogenic because homozygous or compound heterozygous defect of *NPHP1* can cause Nephronophthisis, which is transmitted through an autosomal recessive inheritance pattern [35].

In conclusion, we report a case of atypical ADPKD presenting as GCK accompanied by TBMN in an adult man. GCK is a rare disease and often overlooked. It is important for practicing nephrologists to have a clear understanding of GCK. Moreover, GCK involves in various conditions, and gene test should be considered.

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None.

Author contributions
W.W. and L.C. prepared clinical data and figures, F.D. and X.L. wrote the main manuscript text. All authors reviewed the manuscript.

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Data availability
The authors confirm that the data supporting the findings of this study are available with the article or from the corresponding author on reasonable request. The datasets generated during the current study are available in online repository, The names of the repository/repositories and accession number can be found below: https://www.ncbi.nlm.nih.gov/sra/PRJNA1251727.

Declarations

Ethics approval and consent to participate
This study was approved by the Ethics Committee of Shanghai Ninth People's Hospital Affiliated Shanghai Jiaotong University School of Medicine, the reference number is SH9H-2025-T129-1. The study adhered to the Declaration of Helsinki. Written informed consent were obtained from patient for ethics approval and consent to participate.

Consent for publication
Written informed consent were obtained from the patient and all family members for publication of this case report and any accompanying images.

Competing interests
The authors declare no competing interests.

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