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HNF1B mutation as a cause of polycystic kidney disease – a case report

Mutacja *HNF1B* jako przyczyna wielotorbielowatości nerek – opis przypadku

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Abstract

Mutations in the *HNF1B* gene represent an important cause of kidney disease and monogenic diabetes of the MODY type. These mutations are characterised by marked phenotypic variability – ranging from isolated cystic changes in the kidneys to complex syndromes involving metabolic disturbances, diabetes, and developmental anomalies of other organs. In this report, we describe the case of an 8.5-year-old boy in whom the first and only clinical manifestation consisted of renal cystic changes. Molecular testing revealed a pathogenic mutation in the *HNF1B* gene, confirming the genetic basis of the disease. This case exemplifies the diagnostic challenges posed by the broad phenotypic spectrum of *HNF1B*-related disorders and highlights the fact that renal abnormalities may precede the onset of other clinical features. The patient was placed under nephrological, diabetological, and genetic follow-up, with further management focused on monitoring renal function, electrolyte balance, and glycaemic control. This case underscores the necessity of including *HNF1B* mutations in the differential diagnosis of renal cystic disease, emphasises the pivotal role of genetic testing in identifying the underlying aetiology, and demonstrates the importance of early recognition for ensuring comprehensive and personalised care.

Keywords: monogenic diabetes, *HNF1B*, MODY, polycystic kidney disease

Streszczenie

Mutacje w genie *HNF1B* są ważną przyczyną chorób nerek i cukrzycy monogenowej typu MODY. Charakteryzują się dużą zmiennością fenotypową – od izolowanych zmian torbielowatych w nerkach po złożone zespoły obejmujące zaburzenia metaboliczne, cukrzycę i wady rozwojowe innych narządów. W pracy przedstawiono przypadek 8,5-letniego chłopca, u którego pierwszą i jedyną manifestacją kliniczną były zmiany torbielowate w nerkach. W badaniach molekularnych stwierdzono obecność patogennej mutacji w genie *HNF1B*, co potwierdziło genetyczne podłoże choroby. Opisany przypadek dobrze ilustruje trudności diagnostyczne wynikające z dużej zmienności fenotypowej oraz to, że zmiany nerkowe mogą wyprzedzać inne objawy. Pacjenta objęto opieką nefrologiczną, diabetologiczną i genetyczną, a dalsze postępowanie koncentrowało się na monitorowaniu czynności nerek, gospodarki elektrolitowej i glikemii. Opis przypadku podkreśla konieczność uwzględniania mutacji *HNF1B* w diagnostyce różnicowej oraz rolę zarówno badań genetycznych w diagnostyce przyczyn torbielowatości nerek, jak i wczesnego rozpoznania w zapewnieniu kompleksowej, spersonalizowanej opieki.

Słowa kluczowe: cukrzyca monogenowa, *HNF1B*, MODY, wielotorbielowatość nerek

INTRODUCTION

Renal cysts are fluid-filled cavities of varying density and composition, located within the renal cortex or medulla, arising as a consequence of dilatation of different segments of the renal tubules. Renal cysts are a common finding in imaging studies and may have diverse aetiologies[1].

Cystic kidney diseases resulting from mutations in genes regulating renal development and function are classified as ciliopathies. These include autosomal recessive polycystic kidney disease (ARPKD), autosomal dominant polycystic kidney disease (ADPKD), and nephronophthisis. A distinct form of cystic kidney disease is associated with mutations in the hepatocyte nuclear factor-1β (*HNF1B*) gene[1].

The product of this gene, HNF1β, is expressed in multiple organs, predominantly in the kidneys, liver, intestines, pancreas, and genitourinary tract. Consequently, carriers of *HNF1B* mutations may present with developmental abnormalities in any of these organs[2]. This mutation is also responsible for a distinct form of monogenic diabetes – *HNF1B*-MODY (maturity-onset diabetes of the young), formerly referred to as MODY 5 – which represents the fourth most frequent MODY subtype (after MODY 2, 1, and 3) and accounts for approximately 5–10% of all monogenic diabetes cases[2,3].

It is characterised by a multisystem phenotype that includes not only disturbances of carbohydrate metabolism but also other metabolic abnormalities such as hypomagnesaemia, hypermagnesuria, hyperuricaemia, and hypokalaemia, as well as structural and functional alterations of the kidneys, liver, pancreas, and genitourinary tract[4–7]. Importantly, in children, the clinical presentation most often begins with renal abnormalities, which typically precede the onset of diabetes[8].

In the majority of affected individuals, developmental renal anomalies are observed, including renal cysts and dysplasia, as well as structural malformations such as horseshoe kidney and renal hypoplasia[2,9–13].

Whereas metabolic and electrolyte disturbances usually manifest later in life, diabetes typically develops during the second to third decade[14–17]. In clinical practice, this implies that the diagnosis of *HNF1B*-MODY is often established during the differential work-up of kidney diseases, primarily renal cystic disease, rather than in the context of overt diabetic symptoms.

Genetic testing plays a pivotal role in establishing a definitive diagnosis, particularly when using next-generation sequencing (NGS) technologies, which encompass genes responsible for various forms of renal cystic disease, including *HNF1B*[2,18].

Furthermore, tools have been developed to support the qualification of patients with congenital anomalies of the kidney and urinary tract for molecular testing, such as the *HNF1B* score and the *HNF1B* Mutation Probability Calculator[19,20]. These instruments enable estimation of the likelihood of a mutation and thus facilitate decision-making regarding the implementation of genetic testing.

This paper describes the case of an 8.5-year-old boy in whom an *HNF1B* mutation was identified during genetic evaluation for renal cystic disease, prompted by a positive family history and the presence of cystic changes on imaging studies.

CASE REPORT

An 8.5-year-old boy was admitted to the clinic with a diagnosis of polycystic kidney disease. Postnatal abdominal ultrasonography revealed small renal cysts: two in the right kidney, measuring 5 × 3 mm and 5 × 5 mm, and one in the left kidney, measuring 3 mm. The family history was notable for diabetes mellitus in the patient's mother, maternal uncle, and maternal grandmother. In addition, the grandmother was diagnosed with gout, while both the grandmother and great-grandfather had renal cystic disease of unknown aetiology. Between 2017 and 2025, the patient was hospitalised multiple. Renal function indices remained within normal limits at each admission. Serum concentrations of thyroid-stimulating hormone, parathyroid hormone, fasting glucose, electrolytes (magnesium, sodium, and potassium), and uric acid were consistently within the reference range (Tab. 1). Serial 24-hour urine collections demonstrated neither albuminuria nor proteinuria. Assessment of crystalloid excretion in 24-hour urine was not performed.

Over the years, abdominal ultrasonography revealed bilaterally small kidneys with poor corticomedullary differentiation and multiple small cortical and medullary cysts measuring 2–5 mm (up to approximately 10 in each kidney). In the upper pole of the left kidney, cysts with distinct septations measuring 12 × 10 mm were noted. No interval progression of lesions was observed during subsequent

Year of hospitalisation	Creatinine [mg/dL]	Urea [mg/dL]	Cystatin C [mg/L]	Serum glucose [mg/dL]	Serum uric acid [mg/dL]	Serum magnesium [mg/dL]	Serum potassium [mmol/L]	Urine specific gravity [mg/dL]
2017	0.4	30	1.54	93	4.6	3.9	4.6	1.010
2018	0.5	46	1.53	87	4.9	2.2	4.2	1.008
2020	0.5	35	1.32	88	4.9	2.5	4.5	1.009
2023	0.6	32	1.26	94	5.2	2.0	4.2	1.005
2024	0.7	46	1.35	98	5.3	2.1	4.6	1.009
2025	0.7	35	1.25	87	5.6	1.7	4.2	1.008

Tab. 1. Blood serum test results over the years

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hospitalisations. As part of the diagnostic work-up, abdominal computed tomography was performed in 2022, which demonstrated a polycyclic, hypodense focal lesion measuring 11 × 10 mm with possible thin septations in the upper pole of the left kidney. In the upper pole of the right kidney, an oval hypodense lesion measuring approximately 6 × 3 mm was identified. In addition, single hypodense foci up to 3 mm in diameter, consistent with small cysts, were present in both kidneys.

In 2020, renal scintigraphy showed both kidneys with preserved secretory and excretory function. In 2017, genetic testing for selected renal cystic disease–associated genes revealed no pathogenic or likely pathogenic variants, but variants of uncertain significance were identified in the *RPGRIP1L* (*RPGRIP1*-like) gene. This test did not allow determination of whether the variants were located in *cis* or *trans* alleles of *RPGRIP1L*. Given that *RPGRIP1L* mutations may underlie Joubert syndrome or Meckel syndrome, brain magnetic resonance imaging was performed, revealing no abnormalities. The patient was evaluated twice at a Clinical Genetics Outpatient Clinic, where no genetic basis for polycystic kidney disease was established in the available results. However, due to concerning ultrasonographic findings (small kidneys with multiple cysts) and persistently low urine specific gravity (1.009 g/L, with normal range 1.016–1.035 g/L), repeat genetic testing was recommended in 2024. NGS analysis identified a pathogenic variant in the *HNF1B* gene, which is associated with the renal cysts and diabetes syndrome. The disease is inherited in an autosomal dominant manner; thus, a mutation in a single copy of the *HNF1B* gene is sufficient to cause clinical manifestations. Following the identification of the *HNF1B* mutation, the boy has remained under continuous genetic, diabetological, and nephrological follow-up.

DISCUSSION

The spectrum of clinical manifestations associated with *HNF1B* mutations is remarkably broad, ranging from isolated renal abnormalities, through the *HNF1B*-MODY phenotype (formerly referred to as MODY 5), to complex multisystem syndromes involving the pancreas, liver, and reproductive tract[10,19].

The presented case of an 8.5-year-old boy carrying a pathogenic *HNF1B* gene mutation illustrates the diagnostic challenges inherent in identifying *HNF1B*-MODY, particularly when the clinical presentation is limited to renal involvement. In this patient, the only clinical manifestation consisted of cystic kidney changes, without any accompanying metabolic or electrolyte disturbances, further highlighting the marked phenotypic heterogeneity of the disorder.

An additional diagnostic challenge in this case stemmed from the fact that the initial genetic testing did not reveal any pathogenic or likely pathogenic variants. This was most likely due to the use of an earlier gene panel that did not include large deletions or gene rearrangements. It was only upon re-evaluation using NGS that a definitive diagnosis was established. This finding underscores the growing importance of advanced molecular techniques in the diagnosis of monogenic disorders, enabling the identification of cases that would previously have remained undiagnosed[9,21].

Furthermore, the family history of the patient, which was positive for both diabetes mellitus and renal cystic disease, suggested a hereditary aetiology. However, it is important to note that approximately 50% of *HNF1B* mutations occur *de novo*[9].

Variable penetrance of *HNF1B* mutations has been observed, and the clinical phenotype may differ significantly even among individuals within the same family[19].

In the present case, the ability to definitively determine the familial nature of the mutation was limited by the lack of genetic testing in other family members.

It is worth emphasising that although *HNF1B* was originally identified as a gene responsible for MODY, in clinical practice, the most common initial manifestation of *HNF1B* mutations is renal involvement rather than diabetes mellitus. Therefore, genetic testing for *HNF1B* mutations should be considered in patients with unexplained renal cysts, particularly when accompanied by diabetes, early-onset gout, or congenital uterine anomalies[17].

In recent years, efforts have been made to develop tools to assist in the selection of patients for genetic testing. To this end, Faguer et al. developed the *HNF1B* score, a scale based on 17 criteria, including family history, as well as biochemical and imaging findings assessing the involvement of organs such as the kidneys, pancreas, liver, and reproductive tract (Tab. 2)[20].

A similar approach was undertaken by Kołbuc et al., who developed the *HNF1B* Mutation Probability Calculator, based on clinical and laboratory criteria such as the presence of renal abnormalities, hypomagnesaemia, and hypermagnesuria, among others. This tool allows for the estimation of the likelihood of a mutation, thereby facilitating decisions regarding referral for genetic testing[19]. Furthermore, in their study, they emphasised the risk of underestimation of hypomagnesaemia in the paediatric population when determining the probability of a mutation, due to inadequately low reference values for children[19]. In the described patient, the serum magnesium levels ranged from 3.9 mg/dL to 1.7 mg/dL (1.61–0.7 mmol/L). According to generally accepted reference ranges, hypomagnesaemia is defined as a magnesium concentration of <0.7 mmol/L. Based on the reference values established by Kołbuc et al. (for boys aged 0–9 years, a reference range of 0.82 mmol/L), the most recent test results for the patient indicated hypomagnesaemia. The aforementioned tools help enhance diagnostic efficiency and reduce the number of unnecessary tests.

It is important to note that *HNF1B* regulates the transcription of genes involved in polycystic kidney and hepatic disease 1 (PKHD1), polycystin 2 (PKD2), and uromodulin (UMOD). The homologues of these genes are mutated in ARPKD, ADPKD, and autosomal dominant tubulointerstitial kidney disease (ADTKD) associated with uromodulin,

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Characteristics	Item	Value
Family history		+2
Antenatal renal abnormalities	Uni-/bilateral abnormality by renal echography	+2
Kidney (points for each kidney separately)	Hyperechogenicity	+4
	Renal cysts	+4
	Hypoplasia	+2
	Multicystic and dysplastic kidney	+2
	Urinary tract malformation	+1
	Solitary kidney	+1
Electrolyte or uric acid disorders	Low serum Mg^{2+} (<0.7 mmol/L)	+2
	Low serum K^{+} (<3.5 mmol/L)	+1
	Early-onset gout (>30 years of age)	+1
Pathological findings	Oligomeganephronia or glomerular cysts	+4
Pancreas	MODY, hypoplasia of pancreatic tail and neck, or pancreatic exocrine insufficiency	+4
Genital tract	Genital tract abnormality	+4
Liver	Liver test abnormalities of unknown origin	+2
Negative predictive value: ≤8.		

Tab. 2. HNF1B *score. Author's own elaboration based on Faguer et al.*[20]

respectively. A specific form of ADTKD, known as ADTKD-*HNF1B*, is referred to as *HNF1B*-MODY.

In clinical practice, the differential diagnosis primarily includes ADPKD and ARPKD. These conditions may present with similar ultrasonographic findings; however, they differ in the age of onset, prognosis, and the frequency of associated metabolic disturbances[23].

On ultrasonographic examination, ARPKD typically presents as a generalised abnormal appearance of the renal parenchymal layer (characterised by a "salt-and-pepper" pattern and loss of corticomedullary differentiation), whereas *HNF1B*-MODY and ADPKD are characterised by the presence of cysts. However, the number of cysts alone is not fully discriminatory. In patients with *HNF1B* mutations, there are typically fewer than five cysts, whereas the diagnosis of ADPKD in the population aged between 15 and 39 years can be established based on the identification of three cysts in both kidneys on ultrasonography[23,24].

Hypertension in children with ARPKD is detected prenatally in 90% of cases, while it is observed in 35% of children with ADPKD, and only 22% of children with *HNF1B*-MODY present with elevated blood pressure[25,26].

An important differentiating marker in children is the presence of hypomagnesaemia and hypokalaemia (62% vs. 46%), which are frequently encountered in patients with *HNF1B* mutations, but less commonly observed in ADPKD and ARPKD[25]. A comparative overview of *HNF1B*-MODY, ADPKD, and ARPKD is presented in Tab. 3[23,27,28].

Although *HNF1B*-MODY most commonly presents with renal involvement and MODY-type diabetes, manifestations involving other organs have also been documented.

Liver dysfunction occurs in approximately 40% of patients with *HNF1B* mutations, with the prevalence rising to up to 85% among individuals with MODY5 diabetes. Additionally, cholestasis, elevated gamma-glutamyl transpeptidase (GGTP) levels, and liver fibrosis may occur; in some cases, these conditions can progress to the point where liver transplantation is required at a young age[29,30].

Moreover, pancreatic atrophy is observed in 30% of patients with *HNF1B* mutations and in approximately 50% of those with diabetes[31].

It is also worth noting that around 40% of female patients exhibit reproductive tract anomalies, most commonly severe anatomical defects, such as rudimentary uterus, bicornuate uterus, or vaginal aplasia[31]. In contrast, males with *HNF1B* mutations may present with reproductive system anomalies such as cryptorchidism, abnormal testicular descent, hypospadias, and prostate hypoplasia.

In the presented patient, no involvement of the reproductive system, pancreas, or liver was observed. This was confirmed by both clinical examination and numerous abdominal ultrasonographic studies performed over several years.

The diagnosis of an *HNF1B* mutation carries significant prognostic and therapeutic implications. *HNF1B* nephropathy is characterised by a slow progression, and chronic kidney disease may not manifest until adulthood, typically becoming apparent in the fifth decade of life[20,32].

Characteristics	*HNF1B*-MODY	ADPKD	ARPKD
Inheritance	AD/50% *de novo*	AD	AR
Arterial hypertension[1]	22%	35%	90%
MODY	+	–	–
Gout	+	–	–
Hypomagnesaemia	+	–	–
Renal cysts	<5	Numerous[2]	Microcystic in early childhood
Liver abnormalities	Elevated liver enzymes	"Liver cysts" common in adults but rare in children	From mild cholestasis to hepatic fibrosis, portal vein hypertension, oesophageal varices, cholangitis, and cirrhosis
Chronic kidney failure	Rare	Approximately 50% by age of 60 years	Approximately 50% by age of 20 years

[1] In children.
[2] In individuals aged 15–39 years, the presence of ≥3 cysts in both kidneys is sufficient for ultrasonographic diagnosis of ADPKD.

Tab. 3. Table comparing HNF1B-*MODY, ADPKD and ARPKD. Author's own elaboration*

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Mutations in the *HNF1B* gene are responsible for 5–10% of MODY cases, while diabetes is observed in approximately 25% of *HNF1B* mutation carriers[33].

In the presented case, the patient did not exhibit signs of diabetes. However, due to the predisposing mutation, the boy was enrolled in a diabetes clinic, which will enable proper disease diagnosis in the future and early initiation of appropriate treatment. For patients with *HNF1B* mutations, a comprehensive care approach is recommended, involving a nephrologist, diabetologist, and clinical geneticist. Regular assessment of renal function is necessary, along with ultrasonographic monitoring of the size and number of cysts. Adult patients with normal renal function should have their serum creatinine levels checked and glomerular filtration rate calculated annually. For children with normal renal function, the frequency of follow-up examinations should be guided by the clinical situation. Regular monitoring of electrolyte levels, especially potassium and magnesium, is also recommended, with appropriate supplementation if deficiencies are identified. Increased fluid intake is advised for nephroprotection. Additionally, regular monitoring of glucose levels in serum should be conducted to detect the onset of *HNF1B*-MODY diabetes at an early stage. Patients diagnosed with diabetes should have their haemoglobin A_{1c} (HbA_{1c}) levels measured regularly. Adults without a diagnosis of diabetes should have their HbA_{1c} tested annually. Children without a diagnosis of diabetes should undergo urine glucose screening once a year. Most cases of diabetes in patients with renal cysts and diabetes syndrome progress rapidly to insulin therapy, making early diagnosis crucial to prevent metabolic complications. The patient should be referred to a genetics clinic, which will provide comprehensive genetic counselling and support future family planning in an informed manner[34].

CONCLUSIONS

The presented case underscores that mutations in the *HNF1B* gene are characterised by significant phenotypic variability. In children, the first and sometimes the only manifestation of the disease may be cystic renal changes, preceding the onset of carbohydrate or electrolyte disturbances. In clinical practice, this fact should prompt consideration of *HNF1B* mutations in the differential diagnosis of unexplained renal cystic disease, particularly in patients with a positive family history of diabetes or renal disorders. Early diagnosis is crucial both prognostically and therapeutically, as it allows for comprehensive nephrological, diabetological, and genetic management, along with regular monitoring of renal function, electrolyte levels, and glycaemia. Such an approach facilitates the early detection of complications and the implementation of appropriate interventions. Given the lack of causal treatment, long-term monitoring, nephroprotective prophylaxis, and family education are of paramount importance. The family should be made aware of the hereditary nature of the disease and the potential risk of its occurrence in future generations.

Conflict of interests

The authors do not report any financial or personal connections with other persons or organisations which might negatively affect the content of this publication and/or claim authorship rights to this publication.

Author contribution

Original concept of study: NG, JM. Collection, recording and/or compilation of data: JM. Analysis and interpretation of data: KM, MS. Writing of manuscript: NG, KM, MS, JM, MP. Critical review of manuscript: MP, BK. Final approval of manuscript: BK.

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