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A Rare Case of MODY12 Complicated with Gitelman Syndrome: A Case Report and Literature Review

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Abstract

Background: Maturity-onset diabetes of the young type 12 (MODY12) and Gitelman syndrome are both classified as rare diseases. Gitelman syndrome increases the risk of type 2 diabetes by contributing to hypokalemia, hypomagnesemia and hyperaldosteronemia, all of which disrupt insulin secretion and lead to insulin resistance. Nevertheless, other types of diabetes are rarely associated with Gitelman syndrome. This report presents an uncommon case of MODY12 co-occurring with Gitelman syndrome, associated with a novel mutation in the ABCC8 gene.

Case Summary: A 21-year-old male patient was admitted to our hospital with the symptoms of polyuria and polydipsia for 3 months. Laboratory tests revealed a fasting plasma glucose (FPG) of 13.7 mmol/L, a 2-hour postprandial glucose level of 24.2 mmol/L, and a glycosylated hemoglobin (HbA1c) of 13.6%. The patient demonstrated impaired insulin secretion (fasting C peptide: 0.37 nmol/L; 2 hours oral glucose tolerance test (OGTT) C peptide: 0.85 nmol/L). Additionally, the patient presented with hypokalemia (2.62 mmol/L), hypomagnesia (0.35 mmol/L), metabolic alkalosis (pH 7.439, HCO3- 33.3 mmol/L) and hyperaldosteronism (renin 66.52 ng/mL/h; aldosterone 942.228 pg/mL), while blood pressure remained within normal limits (99/77 mmHg). Furthermore, genetic analysis through next-generation sequencing (NGS) identified a missense variant in the ABCC8 gene (NM_00352.4: c.892C>T; NM_005544.2: c.1435C>T) and the IRS1 gene (/NM_005544.2: c.1435C>T).

The patient received a diagnosis of MODY12 co-occurring with Gitelman syndrome. Genetic evaluation revealed a paternal heterozygous carrier status for the ABCC8 gene mutation. Sulfonylurea therapy resulted in better glycemic control. Potassium homeostasis was achieved through daily supplementation with 6.0 g of potassium chloride sustained-release tablets (262 mg/tablet).

Conclusion: This case report presents a rare co-occurrence of MODY12 and Gitelman syndrome with a novel stop-gain mutation in the ABCC8 gene. Although no correlation was found between the ABCC8 gene mutation and Gitelman syndrome in this case, the possibility of Gitelman syndrome should be evaluated in MODY12 patients exhibiting hypokalemia, hypomagnesemia, hypochloremia, metabolic alkalosis, elevated renin and aldosterone levels, and normotension.

Keywords: Diabetes, MODY12, Gitelman syndrome, ABCC8, IRS1

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Introduction

Maturity-onset diabetes of the young (MODY) is an infrequent form of monogenic diabetes. There are at least 14 pathogenic genes associated with MODY, accounting for about 1% to 2% of all diabetic cases [1,2]. MODY12, which accounts for roughly 1% of MODY cases, results from mutations in the ABCC8 gene. This gene encodes the sulfonylurea receptor of the KATP channel [3], and its mutation may disrupt the potential of β cell membrane which then impairs insulin secretion [4].

Gitelman syndrome is an autosomal recessive inherited disorder resulting from pathogenic variants in the *SLC12A3* gene. This gene encodes the Na-Cl co-transport protein (NCC). Mutations in the *SLC12A3* gene impair this sodium reabsorption process, leading to diuretic effects. The clinical presentation typically includes hypokalemia, hypocalcemia, hypomagnesemia and metabolic alkalosis [5]. Due to its rarity and the non-specific nature of its symptoms, Gitelman syndrome is

frequently subject to misdiagnosed [6].

Gitelman syndrome elevates the risk of developing type 2 diabetes through hypokalemia, hypomagnesia and hyperaldosteronemia, all of which contribute to impaired insulin secretion and cause insulin resistance. Nevertheless, the association between Gitelman syndrome and other forms of diabetes remains uncommon. This report details a rare case of MODY12 concomitant with Gitelman syndrome with a novel missense mutation in the ABCC8 gene.

Case Presentation

Chief complaints

A 21-year-old male patient was admitted to our hospital with a history of polydipsia, polyuria and emaciation for 3 months.

History of present illness

The patient reported the onset of excessive thirst and frequent urination 3 months prior to admission, with no identifiable precipitating factors. Over this period, he experienced a weight loss of approximately 10 kg. The patient was diagnosed with diabetes at a local hospital, where the fasting plasma glucose was found to be 13.19mmol/L, and urine testing revealed 3+ glucose and positive ketones.

Past medical and Family history

The patient reported no previous history of similar medical conditions and relevant family history.

Physical examination

On physical examination, the patient appeared emaciated, with a height of 180 cm and weight of 60 kg. Vital signs included a body temperature of 36.2°C, a respiratory rate of 18 breaths per minute, blood pressure of 99/77 mmHg, and a regular heart rate of 111 bpm. Thyroid palpation revealed no abnormalities. Pulmonary auscultation was clear, and cardiac examination showed normal heart sounds without murmurs or friction rubs. There was no edema in the lower extrimities and bilateral dorsal foot arterial pulse were palpable. Reflexes were intact, with no pathological reflexes observed.

Laboratory test

The patient's laboratory biochemical findings are summarized in Table 1. The blood routine was normal. Notably, the patient exhibited an elevated HbA1c level (13.1%), a decreased fasting C-peptide level (0.37 nmol/L) and negative results for diabetic antibodies. The serum potassium was low (2.62 mmol/L), although a relatively elevated 24 hours urinary potassium (41.4 mmolmmol/24h) was observed. No identifiable causes for the hypokalemia, such as diuretics usage, diarrhea or vomiting, were found. Additionally, the patient had reduced serum magnesium (0.35 mmol/L), low chloride (96.1 mmol/L), metabolic alkalosis (pH: 7.439, $HCO3^-$: 33.3 mmol/L). Liver function tests revealed elevated transaminases (ALT: 100 U/L, AST: 52 U/L). Hyperaldosteronism was confirmed (upright position renin: 66.52 ng/mL/h; aldosterone: 942.228 pg/mL). Additionally, 24-hour urinary protein excretion was increased (455 mg/24 h). Thyroid antibody (TPOAb and TgAb) levels were elevated, though thyroid functions remained normal. Immunological testing was negative for anti-keratin antibodies, anti-citrullinated peptide antibodies, nucleosome antibodies, anti-dsDNA, Smith antibodies, and so forth. Serum levels of immunoglobulin A (IgA), immunoglobulin M (IgM), complement C3 and C4, and total IgE were within normal ranges, while immunoglobulin G (IgG) was found to be low at 6.23 g/L (normal range: 7-16 g/L).

Table 1: Plasma and urine biochemical parameters of the patient on admission.

	Value	Reference range
Blood tests		
WBC, White blood cell (×10^9/L)	5.34	3.5-9.5
RBC, Red blood cell (×10^9/L)	5.62	4.3-5.8
PLT, Platelet count (×10^9/L)	277	125-350
FPG, Fasting plasma glucose (mmol/L)	13.7	3.9-6.1
HbA1c (%)	13.6	4-6.3
ICA	Negative	
IAA	Negative	
GADA	Negative	
Mg^+, magnesium, mmol/L	0.35	0.75-1.02
K^+, potassium, mmol/L	2.62	3.5-5.3
Na^+, sodium, mmol/L	139.7	137-147
Ca^+, calcium, mmol/L	2.18	2.11-2.52
phosphorus, mmol/L	1.39	0.85-1.51
chlorine, mmol/L	96.1	96-108
ALT, alanine aminotransferase,U/L	100	Sep-50
AST, aspartate transaminase,U/L	52	15-40
BUN, blood urea nitrogen, mmol/L	3.9	3.1-8.0
CR, Serum creatinine, mmol/L	51	57-97
TRAb, thyroid-stimulating hormone, IU/L	<0.800	0-1.75
TgAb, anti-thyroglobulin antibodies, U/mL	226.8	0-60
TpoAb, thyroid peroxidase antibody, U/mL	>1300	0-60
TSH, Thyrotropin, μIU/mL	2.948	0.38-4.34
FT3, Free triiodothyronine, pmol/L	5.64	2.77-6.31
FT4, Free thyroxine, pmol/L	20.49	10.45-24.38
Supine-Upright test		
Renin (ng/mL/h) (upright)	66.52	Apr-38
Aldosterone (ng/mL) (upright)	942.228	40-310
Arterial blood gas analysis		
pH	7.439	7.35-7.45
HCO3-, mmol/L	33.3	22-26
24-h Urine tests		
Potassium (mmol/24h)	41.4	25-125
Protein(mg/24h)	455	42-225

Based on the patient's clinical presentation and biochemical findings, a diagnosis of a specific form of diabetes in conjunction with Gitelman syndrome was considered. Following the acquisition of written informed consent from the patient and his family, peripheral blood samples were collected from the patient and both parents for genetic screening, including NGS and copy number variation analysis of whole exome sequencing. A missense variant in the ABCC8 gene (/ nm_00352.4: C.892c > t) and the IRS1 gene (/nm_005544.2: c.1435C>T) were detected (Table 2). The impact of ABCC8_ex6 c.892C>T(p.Arg298Cys) mutation on protein function was predicted using SIFT and Polyphen-2 software, both of which indicated a deleterious effect. In contrast, the mutation in IRSI_exl c.1435C>T(p. Pro479Ser) was predicted to have both neutral and harmful effects,

Table 2: Genetic testing results.

Gene	Reference sequence	cDNA level	Protein level	Status	Chromosomal location	Classification of variation
ABCC8	/NM_00352.4	c.892C>T	P.Arg298Cys	Heterozygosis	chr11:17482154	VUS
IRS1	/NM_005544.2	c.1435C>T	p.Pro479Ser	Heterozygosis	chr2:227662020	VUS

Table 3: The plasma and urine ion level of the patient.

Date	FPG	HbAlc (%)	K^+	Na+	Mg^+	Cl-	24h UK^+ (44-90)	24h UNa^+ (94-222)	24h UCl^+ (95-237)	ALT	AST
2021.04.28	7.1	13.6	2.6		0.4					100	52
2021.06.28		5.2								174	159
2021.07.12			3.7							38.3	43.7
2021.08.16	5.2									36.3	40.9
2021.09.13	5.9		3.4							73.2	118
2021.10.11			3.8							35.8	43.3
2021.11.15	5.2									32.4	62.8
2021.12.13	5.3		3.7							23.3	35.9
2022.01.13	4.8		3.6							20.4	38.4
2022.03.24	4.9	5.5	3.5							59.7	37.2
2022.04.21	5.4		3.6	142	0.5	101	42.73	162.5	185.25	30.7	37.1
2022.07.18		5.7	3.5	146	0.5	101	84.51	302.1	284.4	50.6	30.6
2023.01.16	4.9	5.3	3.3							30.3	25.4
2023.4.20	5.8	5.7	3.6	141	0.5	98.6	64.98	246.24	343.58	52.9	46.1

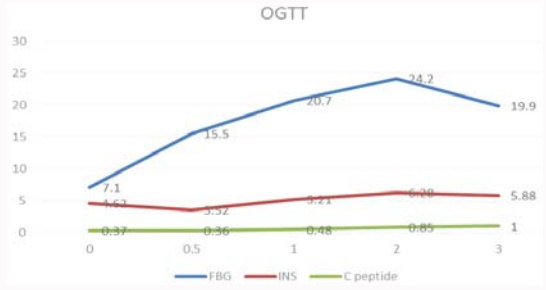

Figure 1: OGTT results of the patient on admission.

according to the respective software analyses.

Imaging and electrophysiological findings

Abdominal ultrasound revealed no significant abnormalities. Thyroid ultrasound indicated diffuse changes consistent with Hashimoto's thyroiditis. Renal ultrasound identified cysts in both kidneys, with the largest measuring 0.9 cm in diameter. Electromyography (EMG) finding included normal motor nerve conduction velocity (MCV) for the right median nerve, the the common peroneal nerve, and the tibial nerve, while the ulnar nerve MCV was slowed. Sensory conduction studies showed decreased amplitude in the right median nerve, with slowed conduction velocity and prolonged latency in the ulnar nerve, superficial peroneal nerve, and tibial nerve. F-wave latency was prolonged in right median nerve, ulnar nerve, and tibial nerve. Electrocardiography revealed sinus tachycardia with nonspecific T-wave abnormalities.

Final diagnosis

MODY12 with Gitelman syndrome.

Treatment, outcome and follow-up

Upon admission, the patient was initially managed with intensive insulin therapy due to significantly elevated blood glucose levels, with a daily insulin dose ranging from 12-15 units. In addition, he was prescribed with two tablets of potassium magnesium aspartate and 2g sustained-release potassium chloride three times daily. Based on the results of genetic testing, insulin therapy was gradually replaced with glimepiride 3mg twice daily. Over a one-year follow-up period, the patient achieved a fasting plasma glucose (FPG) range of 4.8-5.9 mmol/L, a glycosylated hemoglobin level of 5.2-5.7%, and maintained a serum potassium level above 3.5 mmol/L (Table 3). However, his serum magnesium remained low due to insufficient supplement. To support liver function, the patient was also treated by bicycloalcohol and silibinin.

Discussion

We present a case of a patient with MODY12 and Gitelman syndrome, who carries a missense variant (/NM_00352.4: c.892C>T) in the ABCC8 gene and a novel missense variant (/NM_005544.2: c.1435C>T) in the IRS1 gene. To date, more than 30 families of ABCC8-MODY characterized by heterozygous variants have been reported worldwide [7]. In the ClinVar database, ABCC8_ex6 c.892C>T(p.Arg298Cys) variant is classified as having unknown clinical significance. This mutation has been linked to several conditions, including permanent neonatal diabetes type 3, familial hyperinsulinemia type 1, leucine-induced hypoglycemia, and neonatal transient diabetes [8-10]. However, no cases of MODY12 have been reported associated with the specific ABCC8 missense variant (/NM_00352.4: c.892C>T) at this particular locus. ABCC8-MODY is characterized by the absence of ketosis, detectable C-peptide, negative pancreatic islet cell autoantibodies, and sensitivity to sulfonylureas. In contrast, this patient exhibited early-onset hyperglycemia (≤25 years old), an acceptable C-peptide level, and negative islet cell autoantibodies, distinguishing him from type 1 diabetes. Additionally, the patient was emaciated without acanthosis nigricans or insulin

resistance, which is also atypical for type 2 diabetes.

Genetic testing identified a heterozygous missense mutation in ABCC8 gene(c.3976G>A, p.Glu1326Lys), a novel causative mutation for MODY12. Futher genetic testing revealed that the patient's father was a heterozygous carrier of the mutation, though he had no history of diabetes, and there was no history of diabetes in the patient's mother or grandparents. Due to the patient's elevated blood glucose and relatively low body weight, insulin therapy was initially commenced. After 6 months of insulin treatment, and based on genetic test results, insulin therapy was switched to sulfonylureas, which resulted in improved glucose control.

The ABCC8 gene encodes sulfonylurea receptor 1 (SUR1) subunit of the ATP-sensitive potassium channel [7]. Sulfonylureas are recommended for the treatment of MODY12, as they specifically bind to SUR1 subunit, thereby closing the channel and facilitating insulin release in an ATP-independent manner [11]. In this case, glimepiride 4 mg/day was initially prescribed to minimize the risk of hypoglycemia based on previous published literature [12]. The dose was later increased to 6mg/day, resulting in satisfactory blood glucose control.

This case also presented with neuropathic changes. Several studies have suggested that sulfonylureas may ameliorate peripheral nerve damage by modifying ion channels [13]. Additionally, a novel missense variant in the IRS1 gene (/NM_005544.2: c.1435C>T) was identified. This gene having been associated with T2DM [14]. However, given the patient's low C-peptide level and insulin sensitivity, it is unlikely that the IRS1 mutation contributed to the development of diabetes in this case.

The patient also manifested by concurrent hypokalemia, hypomagnesemia, hypochloremia, hyperaldosteronism, and metabolic alkalosis, all of which are typical laboratory findings of Gitelman syndrome. Most cases of Gitelman syndrome are attributed to loss-of-function mutations in the *SLC12A3* gene, which encodes the thiazide-sensitive sodium-chloride co-transporter. Additionally, mutations in *CLCNKB* or *HNF1B* genes have been implicated in some cases [15]. However, genetic analysis in this patient did not reveal mutations in *SLC12A3*, *CLCNKB* or *HNF1B*.

It is important to note that Gitelman syndrome, particularly in adults, may result from factors beyond these known genetic mutations. A study by Mori et al., involving 70 adult patients with Gitelman syndrome, found that 27 cases (38.6%) had genetic confirmation, while 37 cases (52.8%) lacked any identified pathogenic variants linked to the condition [6]. Furthermore, Gitelman syndrome has been reported in association with Sjogren's syndrome and systemic sclerosis, potentially due to impaired NCC function resulting from autoantibody production [16]. Moreove, it has also been reported in patients with autoimmune thyroid diseases, including Grave's disease and Hashimoto's thyroiditis [17]. In this case, the patient exhibited elevated levels of TgAb and TPOAb. However, the potential association between Hashimoto's thyroiditis and Gitelman syndrome remains unclear.

To the best of our knowledge, this represents the first reported case of MODY12 coexisting with Gitelman syndrome. It is important to recognize that Gitelman syndrome may elevate the risk of type 2 diabetes through mechanisms such as hypokalemia, hypomagnesemia, and hyperaldosteronism, all of which contribute to disturbances in glucose metabolism. Hypokalemia can impair insulin secretion by hindering the closure of K-ATP and L-type Ca2+ channels on the β cell surface [18]. Magnesium, a crucial cofactor in various enzymatic reactions involved in energy metabolism plays a significant role in insulin secretion. Hypomagnesemia has been shown to inhibit insulin secretion and reduce the insulin activation of receptor tyrosine kinases. Moreover, magnesium supplementation in diabetic patients has been found to lower fasting plasma glucose (FPG) levels [19]. Hyperaldosteronism can elevate reactive oxygen species, and promote endothelial remodeling, thereby impairing insulin delivery and glucose metabolism [20-21].

A previous study found that a combination of oral potassium and magnesium supplements along with spironolactone treatment, leading to better control of glucose level in patients with Gitelman syndrome; eliminating the need for hypoglycemic drugs over a two-year follow-up period [22]. However, in the present case, the partient's diabetes aligns more closely with MODY12 rather than type 2 diabetes, based on the following reasons: (1) hyperglycemia persisted despite the correction of hypokalemia and hypomagnesemia; (2) sulfonylureas result in better glycemic control compared to insulin; (3) the patient was young, emaciated, exhibited low C-peptide levels, and demonstrated insulin sensitivity. Therefore, this patient was untimatey diagnosed with MODY12 complicated by Gitelman syndrome.

Currently, there is no literature that supports a direct connection between ABCC8 gene mutations and Gitelman syndrome. However, it has been reported patients with ABCC8-MODY12 may be at risk for early-onset and severe diabetic kidney disease. Some patients diagnosed with Gitelman syndrome have impaired tubular structure and function [23]. Impaired tubular function could be a significant factor contributing to the pathogenesis of Gitelman syndrome. Additionally, the ABCC8 gene encodes the sulfonylurea receptor of the KATP channel, which facilitates the inflow of K+ when KATP channels are blocked [24]. The ABCC8 gene mutations may lead to an increase in intracellular K+ while lowering serum K+, which could potentially be responsible in the development of Gitelman syndrome. This patient also carries a mutation in the IRS1 gene. However, no existing literature supports that there is a role for IRS1 mutations in the pathognesis of Gitelman syndrome. Therefore, the underlying cause of Gitelman syndrome in this patient remains unclear and warrants further investigation.

Conclusion

We presented a rare case of MODY12 complicated by Gitelman syndrome. Gitelman syndrome should be considered in patients with MODY12 who present with hypokalemia, hypomagnesemia, hypochloremia, metabolic alkalosis, elevated renin/aldosterone levels, and normal blood pressure. Sulfonylureas are effective in managing MODY12. Symptomatic treatments, including potassium and magnesium supplementation, can improve glycemic control, liver function and tubular function in such patients. However, the underlying cause of Gitelman syndrome in this patient remains uncertain and warrants further investigation.

Author Contribution Statement

Jinlan Xie analyzed the data and wrote the original draft. Yuan Li, Qingqing Ma and Feifei Zhong collected the data. Juhong Yang reviewed and revised the manuscript. All authors read and approved the final manuscript.

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Competing Interests

The authors have no competing interests in this article.

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