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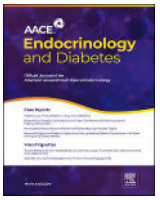

Case Report

When Weight Loss Surgery Meets Hidden Genetics: Lessons From a Case of Post–Bariatric Surgery Hypoglycemia With *ABCC8* and *GCK* Mutation

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ABSTRACT

Background/Objective: Postbariatric hypoglycemia is a recognized complication of Roux-en-Y gastric bypass. We report a case of refractory hypoglycemia following gastric bypass where genetic testing revealed variants that may have contributed to her presentation.
Case report: A 26-year-old woman with a body mass index of 48.4 kg/m^2 underwent Roux-en-Y gastric bypass and achieved ~63 kg of weight loss. Subsequently, she developed recurrent postprandial hypoglycemia with glucose values in the 2.2 to 2.8 mmol/L range, with loss of consciousness. Evaluation excluded insulinoma as a cause. Continuous glucose monitoring confirmed delayed postprandial hypoglycemia. Targeted genetic testing using a 120-gene panel identified a heterozygous pathogenic *ABCC8* variant c.1332G>T (p.Gln444His) and a *GCK* variant c.757G>A (p.Val253Ile) of uncertain significance. Sequential pharmacotherapy with acarbose +/− octreotide 50 μg sc 3 times daily + diazoxide 175 mg 3 times daily yielded only partial improvement (Fig. 1). The patient underwent an open Branco-Zorron switch procedure, after which hypoglycemic episodes decreased in frequency and severity on low-dose octreotide at 4 months of follow-up.
Discussion: The identification of a genetic susceptibility in this patient suggests that latent genetic susceptibility to hypoglycemia may predispose to severe hypoglycemia when unmasked by the metabolic changes following bariatric surgery. This is especially relevant given that hyperinsulinemia is associated with weight gain.
Conclusion: This case raises the possibility that preoperative genetic screening for hypoglycemia-associated variants may improve risk post-bariatric hypoglycemia stratification in bariatric surgery candidates.

Introduction

Recurrent postprandial hypoglycemia after bariatric surgery, termed postbariatric hypoglycemia (PBH), is an increasingly recognized and potentially serious complication that can range from mild autonomic symptoms to severe neuroglycopenic episodes including seizures, loss of consciousness, and hospitalization.[1] Depending on the diagnostic method, biochemical hypoglycemia (glucose <3.0 mmol/L) is found in roughly half of postbariatric patients on provocation tests or continuous glucose monitoring, often without accompanying symptoms.[2] In contrast, hypoglycemia with varying degrees of symptom severity is reported in about one-third of patients. The frequency of hypoglycemia with more severe symptoms is reported in 0.1% to 15% in different studies.[3,4] Despite this, hospitalization for hypoglycemia remains uncommon at the population level (well under 1% of Roux-en-Y gastric bypass [RYGB] patients), but these events carry

Abbreviations: CHI, congenital hyperinsulinism; PBH, post-bariatric hypoglycemia; RYGB, Roux-en-Y gastric bypass.

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substantial morbidity and highlight the need for early recognition and management.[1,4-6] It is likely that genetic predisposition contributes to this heterogenicity in outcome, though evidence to examine this is limited at the current time.

The pathophysiology of PBH is heterogeneous. The most common mechanism involves rapid nutrient transit provoking exaggerated glucagon-like peptide-1 secretion and inappropriately elevated postprandial insulin release.[3,7] Less frequently, insulinoma or islet cell hyperplasia may cause autonomous insulin hypersecretion.[8,9] Genetic variants in the ATP-sensitive potassium channel (KATP channel) genes *ABCC8* and *KCNJ11* and other regulators of insulin secretion (*GCK, GLUD1, HNF4A,* and *HNF1A*) are well-established causes of congenital hyperinsulinism, and their potential role as susceptibility factors for severe PBH has gained increasing recognition in recent years.[10,11]

We describe the case of a patient who developed medically refractory hypoglycemia following RYGB in whom targeted genetic testing identified a heterozygous pathogenic *ABCC8* variant (p.Gln444His) and a *GCK* variant of uncertain significance (p.Val253Ile). This case illustrates that latent genetic defects in β-cell function, clinically silent prior to surgery, may become manifest in the altered metabolic environment following RYGB, and supports the consideration of preoperative genetic screening in selected bariatric surgery candidates at risk for severe PBH.

Highlights

- Postbariatric hypoglycemia (PBH) is a recognized complication of bariatric surgery
- ABCC8 variant c.1332G>T; p.Gln444His; NM_000352.4 and other genetic variants associated with hyperinsulinemia may contribute to genetic susceptibility to PBH
- Genetic screening for hypoglycemia or hyperinsulinemia associated genetic variants prior to bariatric surgery may aid risk stratification for PBH

Clinical Relevance

This case highlights the importance of considering genetic screening for hyperinsulinemia or hypoglycemia-associated genetic variants in bariatric surgery candidates, as latent genetic predisposition may indicate an elevated risk for severe, refractory post-bariatric hypoglycemia.

Case Report

A 26-year-old woman with a high body mass index of 48.8 kg/m^2, and a height of 1.676 m, underwent RYGB surgery and subsequently achieved ~63 kg of weight loss in the following year (Table 1).

Eight months postoperatively, she began experiencing recurrent postprandial hypoglycemia characterized by dizziness, tremor, and lightheadedness ~60 minutes after meals. Several times, these episodes progressed to loss of consciousness requiring assistance. Dietary interventions emphasizing small, frequent meals with reduced simple carbohydrates produced only modest improvement. She denies having experienced hypoglycemic episodes—either by symptoms or on routine laboratory studies—prior to her bariatric surgery. She also denies prior episodes of loss of consciousness, seizures, or hospitalization due to symptoms that could conceivably be related to hypoglycemia. As a child, she had undergone surgical correction of cleft palate.

Over the subsequent years, she developed gastrojejunal stricture and marginal ulcers, a well-described complication following Roux en Y procedure.[12,13] Five years after her original surgery, she presented for the first time to our institution. At that, she was hospitalized for recurrent obstructive symptoms and underwent surgical revision including gastrojejunostomy repair, dilation, partial omentectomy, and adhesiolysis. During that hospitalization, she experienced recurrent hypoglycemias with glucose levels of 2.2 to 2.8 mmol/L requiring intravenous dextrose.

Despite surgical revision and optimized nutritional therapy, hypoglycemia persisted and the patient was referred for endocrine evaluation. Pharmacologic therapy including acarbose, diazoxide, and octreotide at varying doses yielded only partial improvement. Continuous glucose monitor (CGM) monitoring with verification by capillary glucose measurement confirmed delayed postprandial hypoglycemia with values as low as 2.8 mmol/L, typically following modest hyperglycemic excursions (Fig. 1 *A*). She also gained a significant amount of weight, likely in part due to diazoxide-related edema and frequent need for correction of hypoglycemia through intake of simple carbohydrates.

The patient denies any family history of hypoglycemic disorders. The patient has 3 daughters, 2 of whom are alive (7 and 10 years old). One of whom passed away at the age of 3. This daughter had a developmental disorder, a cleft palate, and seizures, and required a feeding tube early on in her life. She was found to have an *SCNA3* mutation. No hypoglycemic events were reported in this offspring. It could not be ascertained whether genotyping of *ABCC8* or *GCK* was performed. Her maternal grandmother was overweight and had type 2 diabetes, diagnosed at about age 30 years. She required management with insulin soon after diagnosis.

Routine laboratory evaluation revealed normal thyroid, hepatic, and renal function (Table 2). A1C was 4.7%, and adrenocorticotropic hormone (ACTH) stimulation testing using the ACTH analogue tetracosactide 250 mcg demonstrated an appropriate cortisol response. Abdominal computed tomography scan showed normal pancreatic morphology.

Continuous glucose monitoring showed recurrent delayed postprandial hypoglycemia with values 3.0 to 3.9 mmol/L, preceded by modest upward glycemic excursions as well as mild fasting hypoglycemia (Fig. 1 *A*). These values were confirmed by capillary glucose measurement on a regular basis.

A supervised fasting study demonstrated only mild hypoglycemia, with a glucose nadir of 3.8 mmol/L. Insulin (<2 μU/mL), C-peptide (0.4 ng/mL), and proinsulin (0.9 pmol/L) were appropriately suppressed (Table 3), and beta-hydroxybutyrate was detectable (0.30 mmol/L), effectively excluding insulinoma.

Given persistent postprandial hypoglycemia despite optimized nutritional and pharmacologic therapy, targeted genetic testing using a 120-gene panel was obtained. Testing identified a

Table 1
Weight Loss Trajectory Prior to and Following Roux-en-Y Gastric Bypass Surgery

Timeline	Weight (kg)	BMI (kg/m^2)
Prior to surgery	136	48.8 kg/m^2
1 mo after surgery	116.6	41.5
1 y after surgery	72.8	25.9
2 y after surgery	57.7	20.5
3 y after surgery	67.7	24.1
4 y after surgery	73.5	26.2
5 y after surgery	62.6	22.3
Year 6 after initial surgery prior to Branco-Zorron switch	84.4	30.0
4 mo following Branco-Zorron switch	57.8 kg	20.5

Abbreviation: BMI, body mass index.

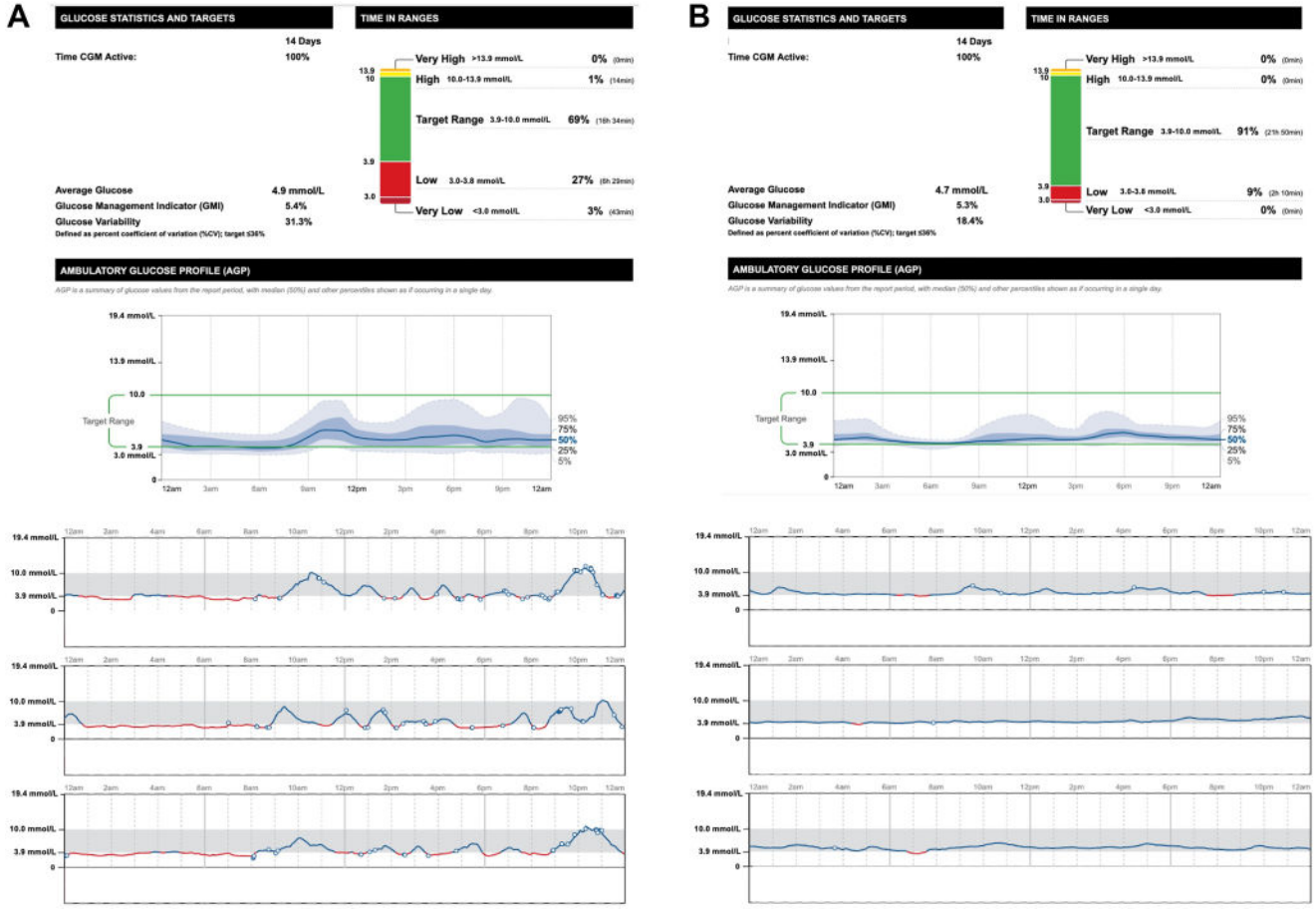

Fig. 1. CGM tracing prior to surgery, while being treated with octreotide 50 μg sc 3 times daily + diazoxide 175 mg 3 times daily (*A*) and 4 months following open Branco-Zorron switch procedure, while being treated with octreotide total daily dose of 200 μg daily (*B*). CGM, continuous glucose monitor.

Table 2
Laboratory Evaluation Results at Presentation to our Institution 5 Years After Bariatric Surgery

Variable	Value	Reference range
Thyroid stimulating hormone	1.50 mIU/mL	0.27-4.20 mIU/mL
Free thyroxine	0.94 ng/dL	0.93-1.70 ng/dL
A1C	4.7%	4.0%-6.0%
Alanine aminotransferase	7 U/L	<33 U/L
Aspartate aminotransferase	17 U/L	<32 U/L
Alkaline phosphatase	72 U/L	35-104 U/L
Creatinine	0.45 mg/dL	0.50-0.90 mg/dL
Glomerular filtration rate	>90 mL/min/1.73 m^2	>59 mL/min/1.73 m^2
Baseline cortisol	8.3 μg/dL	6.0-18.4 μg/dL
Cortisol, 60 min after ACTH stimulation	20.7 μg/dL	>17.9 μg/dL

Abbreviation: ACTH, adrenocorticotropic hormone.

Table 3
Results of Supervised Fasting Test

Variable	Value	Reference range
Serum glucose	3.8 mmol/L	3.9-7.8 mmol/L
Insulin	<2.0 μU/mL	2.6-24.9 μU/mL
Proinsulin	0.9 pmol/L	0.0-10.0 pmol/L
C-Peptide	0.4 ng/mL	0.8-5.2 ng/mL
Beta-hydroxybutyrate	0.30 mmol/L	<0.60 mmol/L
Oral hypoglycemic agents screen	Negative	Negative

heterozygous pathogenic *ABCC8* variant (c.1332G>T; p.Gln444His; NM_000352.4) and a heterozygous *GCK* variant of uncertain significance (c.757G>A; p.Val253Ile).[14]

After multidisciplinary case review, the patient underwent an open Branco–Zorron switch procedure, a surgical technique in which a jejunal segment is interposed between the gastric pouch and the alimentary limb antrum restoring a more controlled physiologic nutrient transit pathway and reducing the rapid carbohydrate delivery to the distal gut that drives exaggerated incretin-mediated insulin secretion.[15] Partial pancreatectomy was considered but deferred following shared decision making. Postoperatively, glycemic control improved moderately, though she continued to require octreotide to maintain symptomatic stability. At 4-month follow-up, hypoglycemic episodes had decreased in frequency and severity on an octreotide only regimen (total daily dose 200 μg). She also had lost a significant amount of weight, likely in part due to decreased diazoxide-associated edema and reduced need for carbohydrate intake to treat symptomatic hypoglycemia. However, complete discontinuation of pharmacotherapy was unsuccessful, with recurrence of more pronounced hypoglycemia upon weaning attempts. A timeline of the case report's most relevant events are summarized in Figure 2.

Discussion

Genetic variations in KATP channel genes are well-recognized causes for hyperinsulinemia and hypoglycemia of varying degrees.

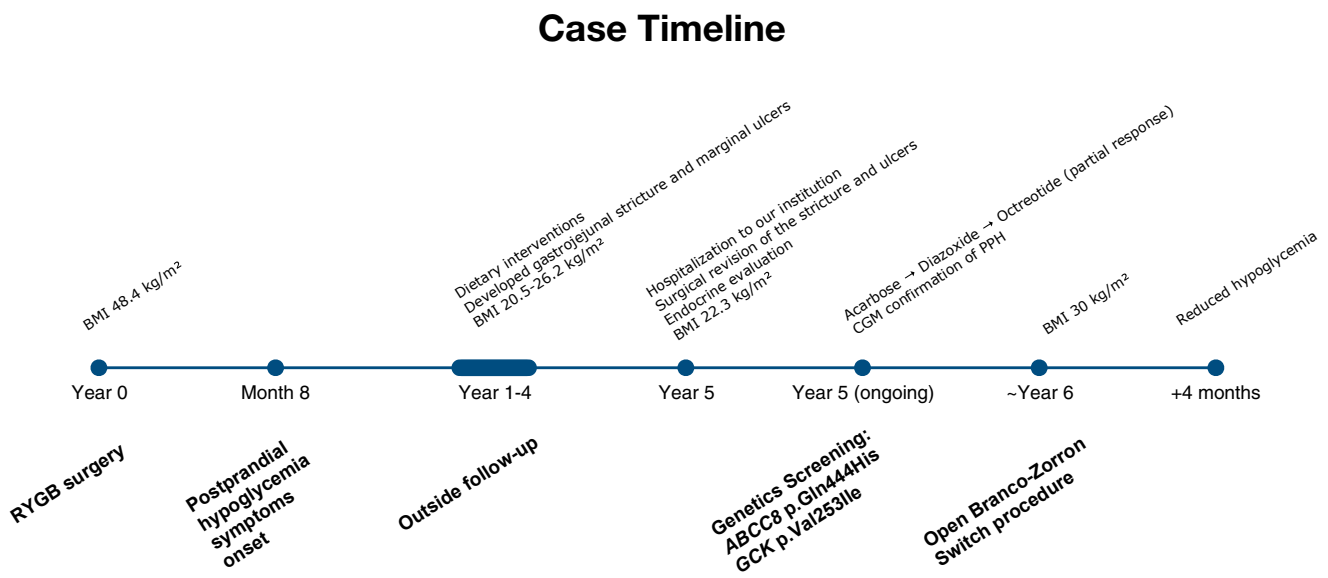

Fig. 2. The timeline summarizes the clinical course of the case reported herein. Primary clinical phases are displayed below the timeline, whereas associated diagnostic and therapeutic actions are shown above the timeline.

The KATP channel comprises Kir6.2 (*KCNJ11*) and SUR1 (*ABCC8*) subunits. Loss-of-function mutations impair channel opening, and cause persistent β-cell depolarization and constitutive insulin secretion, the hallmark of congenital hyperinsulinism (CHI).[10,16] *ABCC8* and *KCNJ11* variants account for 40% to 50% of genetically confirmed CHI.[17,18] Biallelic mutations cause severe, diazoxide-unresponsive CHI, whereas heterozygous variants produce milder, diazoxide-responsive disease with incomplete penetrance.[18-20] Our patient's heterozygous status and partial diazoxide response are consistent with this pattern—preserved function of the intact allele likely permits residual KATP channel activity and partial pharmacologic response.[21]

The p.Gln444His variant has been reported in multiple CHI cohorts in both focal and diffuse disease. Founder *ABCC8* variants are documented in European and Ashkenazi Jewish populations with carrier rates reaching 1 in 52.[22,23] Our patient's Northern European ancestry—with paternal lineage from Germany and maternal roots from Poland and Spain—is consistent with the geographic distribution of this variant and underscores the relevance of ancestry-informed variant interpretation.

The coidentified *GCK* p.Val253Ile variant (c.757G>A; rs748964205) is extremely rare, with an allele frequency of 4.3×10^{-6} in gnomAD v4.1 (7 alleles in 1,613,502 total; no homozygotes).[24,25] This variant has conflicting classifications of pathogenicity in ClinVar: one submitter classified it as likely pathogenic based on its location at a conserved position with nearby pathogenic residues, whereas another classified it as a variant of unknown significance based on biophysical modeling suggesting it is unlikely to disrupt protein function.[26] The only published report of this variant is in a 61-year-old individual with type 2 diabetes from a large-scale sequencing study of monogenic diabetes genes, where in silico predictions were predominantly benign (PolyPhen2: benign; SIFT: tolerated; CADD: 18.4) and the variant was classified as uncertain significance.[27] Its clinical contribution to our patient's presentation therefore remains uncertain and cannot be determined without functional characterization.

A key insight is that latent KATP channel defects may remain silent during chronic caloric excess but become unmasked following RYGB, where restricted intake, weight loss, rapid nutrient transit, and exaggerated incretin secretion create conditions for clinically significant hypoglycemia.[3] However, the absence of functional studies and the single-case nature of this observation preclude definitive conclusions about a causal relationship.

In this family, the exact mode of inheritance cannot be determined, as segregation analysis has not been performed. If the variant were to act in an autosomal dominant manner, each of the patient's surviving daughters would theoretically have a 50% chance of carrying the maternal ABCC8 variant. As of now, none of the children have undergone targeted genetic testing, and no hypoglycemic episodes have been reported. Given the well-described incomplete penetrance and variable clinical expression of heterozygous ABCC8 variants, the lack of symptoms does not rule out carrier status.[18]

After failure of medical therapy, our patient underwent an open Branco-Zorron switch procedure. Surgical reversal has been described as a salvage strategy for refractory PBH, with symptomatic improvement reported in the majority of cases, though complete resolution is not universal.[7,15] Our patient's partial response—reduced hypoglycemic frequency but continued need for low-dose octreotide—further supports a multifactorial etiology driven by both surgically altered anatomy and underlying genetic susceptibility.

Conclusion

This case raises the important question of whether preoperative genetic screening may have a role in the evaluation of selected bariatric surgery candidates. Although prospective data on the prevalence of CHI-associated genetic variants in PBH populations are currently lacking, the identification of pathogenic or likely pathogenic gene variants before surgery could, in principle, inform risk stratification and clinical decision making. We suggest that genetic panel testing for hyperinsulinism-associated genes may potentially be beneficial in patients with a personal or family history of unexplained hypoglycemia, atypical biochemical profiles, or features suggestive of a monogenic β-cell disorder. In such individuals, preoperative identification of a pathogenic variant could influence the choice of surgical procedure—potentially favoring those with lower PBH risk—and guide the intensity of

postoperative glucose monitoring and early intervention. This hypothesis-generating observation requires validation in larger, prospective cohorts to determine whether targeted genetic screening can meaningfully improve outcomes in bariatric surgery candidates.

Also for patients with postbariatric surgery hypoglycemia, even though routine genetic screening cannot currently be recommended it could be a new diagnostic option to obtain targeted genetic testing particularly in patients with severe, refractory, or biochemically unusual presentations, to better characterize potential genetic susceptibility and further define genotype-phenotype relationships in PBH clinically and academically.

Limitations

Several limitations should be acknowledged. This is a single case report, and causal inferences cannot be drawn regarding the role of the identified genetic variants in this patient's hypoglycemia. The relatively short follow-up after surgical reversal (4 months) limits conclusions about long-term outcomes. Finally, a comprehensive pre- and postoperative characterization of insulin secretory dynamics with serial provocative testing was not available, which would have provided additional insight into the temporal evolution of β-cell dysfunction.

Disclosure

The authors have no conflicts of interest to disclose.

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Consent

A written consent for this case report was obtained from the patient as directed by our IRB.

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