



## Case Report

# A Case of Isolated Renal Glycosuria from Nepal

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**Abstract:** Isolated renal glycosuria is a rare genetic disease caused by mutations in SLC5A2 gene. The mutation leads to a defect in glucose transporter, sodium-glucose co-transporter 2 (SGLT2), which is involved in the reabsorption of glucose from proximal tubules. Defect in this transporter leads to loss of glucose in urine. This rare disease has not been reported from Nepal previously, and here we report the first case. A 38-year-old female with complaint of tiredness and fatigue visited our medical centre. On clinical examinations, no other signs and symptoms were reported. The patient had no history of any other disease and the patient was not taking any supplements or medications. On repeated laboratory investigations, there were no signs of abnormal glucose metabolism and proximal tubular dysfunction. No evidence of hepatic, renal, and blood disorders, infection, haematuria, and proteinuria was present. Based on clinical and laboratory investigations, the patient was diagnosed as having isolated renal glycosuria. We report the first case of isolated renal glycosuria in Nepal. Because the disease is often asymptomatic, physicians and health professionals need to be aware of this condition which may occur in their community.

**Keywords:** Glycosuria, Isolated, Nepal, Renal

## 1. Background

Isolated renal glycosuria is a rare disease characterized by glycosuria in the absence of any other kidney diseases while the blood glucose is at a normal level or lower than normal levels [1]. Glycosuria indicates the presence of glucose in a detectable amount in urine, and a majority of the glycosuria is due to diabetes mellitus. Glycosuria can also appear in other conditions such as pregnancy, Fanconi syndrome, Lowe's syndrome, Wilson's disease, usage of certain drugs, and heavy metal poisoning [2].

Kidney plays a major role in the glucose homeostasis. In the normal state, proximal tubule reabsorbs all the glucose filtered by the glomeruli until the renal threshold for glucose (180 g per day) is reached [3]. In the majority of the inherited form of isolated renal glycosuria, also called familial renal glycosuria, mutations in the SLC5A2 gene coding for the glucose transporter, sodium-glucose co-transporter 2 (SGLT2) are

responsible. This transporter is responsible for the reabsorption of the bulk of filtered glucose [4]. Due to the defect in the transporter, the glucose appears in the urine.

The cases of familial renal glycosuria are asymptomatic, and if any the symptoms are usually mild [1, 3, 4]. Thus, physicians may fail to diagnose this condition, and misdiagnosis can occur. In one of the cases, the disorder was misdiagnosed and treated for diabetes mellitus that lead to hospitalization of the patient [5]. The incidence of this disorder is very low, and in Nepal, till date, no such case has been reported. Herein we describe the first case of renal glycosuria in a healthy woman.

## 2. Case Presentation

A 38 years old female visited our outpatient clinic with the symptoms of tiredness and fatigue. Patient reported relief of tiredness by drinking tea with added sugar. On clinical examination, there were no other abnormal signs and

symptoms. There was no history of previous diseases and the patient was not under any supplements or medications. To evaluate if there were any abnormality, a number of blood and urine tests were performed. On repeated blood screening, no abnormal laboratory findings, except for the glucose (3+) in urine were seen. The height and weight were 5.3 feet and 52 kg respectively at the time of the investigation. The detailed investigations are discussed below.

### 2.1. Blood Biochemistry

The fasting and postprandial blood sugar were 51.0 mg/dl and 65.0 mg/dl respectively. The markers of kidney function, serum urea and creatinine were 18.0 mg/dl and 0.6 mg/dl respectively. The serum total bilirubin and direct bilirubin were 0.7 mg/dl and 0.2 mg/dl respectively. The serum concentrations of liver enzymes, alanine aminotransferase (ALT), aspartate aminotransferase (AST) and alkaline phosphatase (ALP) were 21.0 IU/L, 26.0 IU/L and 176.2 IU/L respectively. Thus, blood biochemistry revealed no abnormal glucose metabolism and hepatic and renal dysfunctions.

### 2.2. Haematological Parameters

The haemoglobin level was 13.5 gm/dl (reference range in female 11-16 gm/dl). The total leucocytes count was 9000/mm<sup>3</sup>, with neutrophil 68%, lymphocyte 24%, eosinophil 2% and monocyte 6%. The platelet count was 273,000/mm<sup>3</sup>. These data suggest woman is non-anaemic, and no blood disorders were present.

### 2.3. Urine Analysis

The examination of urine revealed no other pathologies except the presence of glucose (3+). The urine colour was light yellow with clear transparency and acidic pH. There was no detectable albumin, and pus cells (0-1 HPF), epithelial cells (0-1 HPF), RBC (nil), cast (nil) and crystal (nil) were all in normal levels.

## 3. Discussion

The isolated renal glycosuria is a very rare disease, and we report this condition in a Nepalese female aged 38 years. Other abnormalities were ruled out because no abnormal glucose metabolism and signs of proximal tubular dysfunction were seen. At the time of presentation, the patient had no specific complaints except tiredness and because of this it often remains undetected. The woman was taking a regular diet and performing her routine activities and the patient was normal weight (BMI of 20.3 kg/m<sup>2</sup>). Several previous cases have revealed that no specific symptoms are associated with this disease [6, 7]. A recently published study in Israeli defence force showed that renal glycosuria is associated with lower body weight, and elevated blood pressure, which may be due to loss of glucose in urine [8]. The patient experience of improvement in symptoms after taking tea with sugar may be related to the increase in blood glucose level by the sugar and caffeine present in tea.

In the normal circumstances, the proximal tubules reabsorb all the glucose through SGLTs, so glucose does not appear in

the urine. There are six members of the SGLT family, and approximately 90% of glucose is reabsorbed by SGLT2, a high-capacity low-affinity glucose transporter [9]. In the current case, we ruled out any kidney disease including proximal tubular dysfunction by measuring urinary albumin and urine microscopy. The woman had normal liver function as indicated by liver enzymes and bilirubin concentrations. The woman had normal haemoglobin levels, and there were no indications of infection and inflammation. The renal threshold for glucose is decreased in individuals with renal glycosuria due to a defect in SGLT2 transporter [10]. The main cause of this defect is a mutation in the SLC5A2 gene, and to date, around 86 mutations in this gene have been found to be linked with renal glycosuria [11]. Because of the resource constraint, we could not identify the mutation in the current case.

Further genetic investigation in this patient is warranted to identify the mutation present in the disease. In addition, screening of other family members is recommended. Because the cases of renal glycosuria are often asymptomatic, this disease may be circulating in the community and physicians need to be aware of this disease. This is important because it may be falsely treated as type 2 diabetes if physicians or health care professionals are not properly trained and informed of this condition [5]. Early diagnosis of this disease may help prevent unnecessary economic burden and the risk of taking inappropriate medications. Future study needs to be performed for screening glycosuria in this ethnicity of Nepal.

## 4. Conclusion

In summary, we reported the first case of isolated renal glycosuria in the Nepalese population. The condition is often asymptomatic; therefore, physicians need to be aware of this condition as it may be misdiagnosed as glucosuria of type 2 diabetes. Further genetic analysis in the patient is recommended.

## List of Abbreviations

ALP:	Alkaline phosphatase
ALT:	Alanine aminotransferase
AST:	Aspartate aminotransferase
BMI:	Body mass index
HPF:	High power field
RBC:	Red blood cell
SGLT2:	Sodium-glucose cotransporter-2

## Declarations

### Ethics Approval and Consent to Participate

Not required.

### Consent for Publication

Written consent to publish the information was taken from the participant.

**Availability of Data and Material**

Available on request.

**Competing Interests**

The authors declare that they have no conflicting interests.

**Authors' Contributions**

UK, NDP and SK generalized the concept. UK collected and analysed the samples. UK and SK analysed the results. SK drafted the report. All authors read and approved the final version of manuscript.

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