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# Fanconi-Bickel Syndrome as A Cause for Proximal Renal Tubular Acidosis and Cataract

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**Abstract**

Fanconi-Bickel syndrome is a rare autosomal recessive disorder characterized by hepatorenal glycogen accumulation, proximal renal tubular dysfunction and impaired utilization of glucose and galactose. We report a case of Fanconi-Bickel syndrome (FBS) in an 8-month-old boy from Nepal who presented with dehydration fever, developmental delay, cataract, growth retardation and laboratory evidence of proximal renal tubular dysfunction.

**Keywords:** fanconi-bickel syndrome; proximal renal tubular; acidosis and cataract

## Introduction

Fanconi-Bickel syndrome (FBS) (OMIM #227810): also known as glycogen storage disease type XI (GSD XI):is a rare, autosomal recessive disorder of carbohydrate metabolism. It is caused by homozygous or compound heterozygous mutation in the SLC2A2 gene, which encodes the glucose transporter GLUT2. This leads to impaired glucose and galactose utilization, glycogen accumulation in the liver and kidneys, and proximal renal tubular dysfunction.

## Case Report

An 8-month-old male infant from Nuwakot was brought to the pediatric emergency department in an intubated state. He had been experiencing a fever for two months and multiple episodes of abnormal body movements over the past day. The fever was typically low-grade and almost daily. He had also failed to gain weight. Born full-term via spontaneous vertex delivery with birth weight of 2.8 kg the infant had no significant antenatal, natal, or postnatal issues. He was exclusively breastfed until 3 months of age and has been on formula along with other complementary feeds. He has been vaccinated according to the national schedule, with a positive BCG scar. His development was normal until five months of age, when he could sit with support and transfer objects, but he gradually lost these abilities and now can only roll over. He was born to consanguineous parents, and his 4-year-old brother is healthy.

For this condition, the child had been admitted to a hospital in Kathmandu, where he received multiple courses of antibiotics. He was later sent to a nutritional rehabilitation center but was referred back to the hospital due to persistent fever. During the second hospital stay, his consciousness level decreased, prompting the family to take him home, losing all hope for the infant. While at home, he developed generalized seizures and was taken back to the hospital, where he was intubated for low GCS and referred to our hospital for PICU care.

Upon examination, he showed no dysmorphic features. His weight was 5 kg, length 66 cm, indicating moderate wasting and severe stunting, and his occipitofrontal circumference was 43 cm, normal for his age. He had no pallor or icterus. His liver was palpable 3 cm below the right costal margin, and his spleen was not palpable.

During evaluation in previous hospital, his urine analysis showed glucose 3+, protein 1+, urine reducing substance 3+, urinary acetone negative. He had blood glucose upto 250mg. He also had polyuria during hospital stay. His HbA1c was 6gm%. Venous blood gas showed metabolic acidosis (anion gap couldnot be calculated) with respiratory alkalosis.

In emergency, His random blood sugar was 401mg/dl. His liver enzymes were slightly elevated.

Infant was started on insulin infusion but after 6hours of infusion insulin could be stopped due to normalization of blood sugar. However, acidosis was not corrected.

Infant was started on bicarbonate correction and gradually his condition improved along with anticonvulsant.

He was mechanically ventilated for 2 days. After extubation, his GCS was 15/15, with normal-sized, reactive pupils, though there was doubtful cataract.

He exhibited generalized hypotonia, absent reflexes, and hand tremors.

At our center, his blood gas revealed normal anion gap metabolic acidosis and hypokalemia. Rest of the baseline investigations were normal. USG liver and kidneys normal. Xray of long bone was normal. CECT head revealed Benign enlargement of extraxial CSF spaces in bilateral frontoparietal region. Otherwise, normal findings. Eye evaluation confirmed bilateral cataract.

Considering inborn error of metabolism with metabolic crisis involving kidney and brain, whole exome sequencing was sent which detected a homozygous c.1355T>C (p. Leu452Pro) variant in Exon 10 of SLC2A2 gene confirming a diagnosis of Fanconi-Bickel syndrome. Parental seggregation study was advised but could not be done due to financial issues. The patient was treated was Vitamin D analogs and sodium bicarbonate supplementation and advised for restriction of simple sugar.

## Discussion

At the presentation to our emergency, the cause of low GCS on him was probably due to severe metabolic acidosis. As the infant also had very high blood sugar, infantile diabetes was considered as the working diagnosis. However, with the rapid normalization of blood sugar, normal HbA1 C and Normal anion gap metabolic acidosis refute this diagnosis. As the infant also had failure to thrive with normal anion gap metabolic acidosis, renal tubular acidosis was suspected. However, complete evaluation to distinguish proximal and distal RTA could not be done as the infant was already on bicarbonate correction. However, on the background of absence of nephrocalcinosis, hypokalemia and glycosuria, Proximal RTA with Fanconi syndrome was strong possibility. He also had cataract so galactosemia was also considered in differential diagnosis. However, patient was fed on milk from the very early age and despite this he never has documented hypoglycemic episodes, transaminitis, seizure or severe developmental delay which makes classical Galactosemia less like. However, non-classic type of galactosemia due to Galactokinase deficiency with similar presentation has been reported [1]. Also considering cataract and proximal RTA, Lowe's syndrome was also considered in differential diagnosis. Hence, WES was sent and confirmed the diagnosis of Fanconi-Bickel syndrome.

There are few case reports of cataract as clinical manifestation of FBS [1]. However, cataract was not observed in other bigger case series [3,4]. The exact mechanisms linking Fanconi-Biedl syndrome and cataract formation are not fully understood, but impaired metabolism of sugars like galactose and glucose may play a role in lens opacification.

FBS is a rare clinical syndrome caused by recessively inherited homozygous or compound heterozygous pathogenic or likely pathogenic variants in SLC2A2 encoding GLUT2 (SLC2A2). The exact incidence and prevalence are not known but many pathogenic variants in SLC2A2 from different ethnic groups have been identified, of which around half are novel 3,5,6]. There are case report and literatures on clinical presentation of neonatal diabetes with SLC2A2 mutations that should be considered in consanguineous families [7]. In our case also, since there has been SLC2A2 mutation, that could be the reason for presentation of hyperglycemia.

There is the largest case series among 11 patients on FBS from China which reported SLC2A2 variation spectrum [4]. Our patient has homogyzygous missense mutation in SLC2A2 gene. The age at diagnosis for our patient is quite early compared to other cases reported. This probably is due to easy availability of genetic testing in recent days. The early diagnosis of this condition helps in better growth and developmental outcome while late diagnosis complicates the picture and increases the mortality usually because of respiratory infections or liver failure [8,9].

## Conclusion

FBS should be considered in a child presenting with hyperglycemia, proximal RTA and cataract.

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