Atypical hemolytic-uremic syndrome results from complement dysregulation, which can arise from mutations within complement factor H, B and I. This explanation is based on the knowledge that native pathway dysregulation and its consequences can be catastrophic if not timely addressed. The pathogenesis of this syndrome is ill-defined, and its significance is still considered to be unknown. This is, as far as we know, the first case ever described of this apparently disease-causing mutation in our country.

Althought this mutation has been reported in other countries with association to this disease(1,2), its significance is still considered to be unknown. This is, as far as we know, the first case ever described of this apparently disease-causing mutation in our country and is still very much unknown. While detecting a new mutation in CFI gene in a patient affected by aSHU. This mutation was described in her family members. Genetic analysis was performed on her family members. Gene sequencing, revealed the patient to be heterozygous for a new variant c.1071T>G (p.IIe357Met) but not in her father. Her mother is 82 years old and has no history of kidney disease. Severe hemolytic anemia (Hb 3.9 g/dL), a normal C4 complement level and negative antinuclear antibody. Plasmapheresis was initiated with no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response and terminal chronic kidney disease was assumed. She began peri-dialysis after a period of 30 days on hemodialysis. Later, genetic testing for mutations in the alternative complement pathway (CFH, MCP, CFI, CFB, C3, THBD) by gene sequencing, revealed the patient to be heterozygous for a new variant c.1071T>G (p.IIe357Met) but not in her father. Her mother is 82 years old and has no history of kidney disease. Severe hemolytic anemia (Hb 3.9 g/dL), a normal C4 complement level and negative antinuclear antibody. Plasmapheresis was initiated with no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response.

**RESULTS:**
The same mutation was detected in her mother (heterozygous for variant c.1071T>G (p.IIe357Met) but not in her father. Her mother is 82 years old and has no history of kidney disease. Severe hemolytic anemia (Hb 3.9 g/dL), a normal C4 complement level and negative antinuclear antibody. Plasmapheresis was initiated with no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response. Kidney biopsy revealed crescentic glomerulonephritis and vasculitis, and she was started on IV cyclophosphamide. She maintained no clinical response.

**INTRODUCTION AND AIMS:**
Fanconi-Bickel Syndrome (FBS) is a rare renal disease characterized by hepato-renal glycogen accumulation and so for it is also known as Glycogenosis XI. Glycogen accumulation seems to be central in the liver and kidney dysfunction. So far, no causative treatment are available for FBS and patients can survive only by following a strict electrolytes replacement therapy. Here, we treat one patient with an innovative therapeutic approach aiming to target renal glycogen accumulation.

**METHODS:**
One FBS patient was treated off-label for three months with a commercially available drug on the top of the electrolyte replacement therapy. Serum level of phosphate, uric acid and bicarbonate reabsorption threshold were studied as a primary endpoint. The abundance of NapiIIa in urinary exosomes and glycogen content in the urinary sediment were used as innovative endpoints. All the parameters were studied at baseline, day 7, 25 and 83 of treatment and after 7 and 30 days of drug washout.

**RESULTS:**
The treatment induced an improvement of serum phosphate and uric acid and a normalization of serum potassium level. When subjected to a bicarbonate load, the patient showed a shift to the left of the filtrate/excretion bicarbonate curve. The urine sediment showed an increase excretion of renal tubular cells, most of which were mainly from proximal tubule as showed by Napi2a staining. Glycogen content of this sediment was 3 times more abundant than in healthy subjects. The treatment induced a reduction in glycogen in the urinary sediment, while after one-month wash-out it went back to baseline level. Increasing serum phosphate level paralleled the increase Napi2a abundance in the urinary exosomes and so confirmed better tubular reabsorption.

**CONCLUSIONS:**
We have shown here a case report in which an innovative treatment results safe and efficacious for FBS. The limited usage of a single case is the major limitation, but this can serve as background for a more extensive trial.