**GP287**  
**FANCONI BICKEL SYNDROME AND RENAL TUBULAR DYSFUNCTION**  
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**Introduction**  
Fanconi Bickel Syndrome (FBS) is a rare autosomal recessively inherited inborn error of metabolism due to impaired utilization of glucose and galactose. The mutations of the responsible gene SLC2A2 cause defects in glucose transport protein-2. Clinical findings are severe growth retardation, polyuria, polydipsia, and hypophosphatemic rickets. Laboratory findings are proximal renal tubular dysfunction, fasting hypoglycemia and postprandial hyperglycemia leading to diabetes, in some cases even in the neonatal period, due to hepatorenal glycogen accumulation.

**Method**  
Data of 10 patients from 8 families diagnosed as FBS with molecular analysis were reviewed retrospectively for renal involvement.

**Results**  
3(30%) were female and 7(70%) were male. All had parental consanguinity. Mean age at diagnosis was 10.8±8.5 months. Mean current age was 7.3±6 years. One patient died at 1 year of age. Height measurements of 7 patients were below 3rd percentile and weight measurements of all patients were below 3rd percentile. 6 patients (60%) had hepatomegaly. Hypertransaminasemia, hypertriglyceridemia and metabolic acidosis with normal anion gap were observed in all patients. 9(90%) patients had hypophosphatemia, hypokalemia and 6(60%) patients had hypotnatremia. All patients had glycosuria and proteinuria. Tubular phosphate reabsorption levels of 4 out of 8 patients were below 78%. Mean HbA1C levels were 5.6±0.6, oral glucose tolerance test was performed on 6 patients over three years of age. Plasma glucose levels at 120 minutes were higher than 140 mg/dL in 4 (66,7%), higher than 200 mg/dL in 2 patients (33,3%) despite none of them had overt diabetes. 8 patients (80%) showed severe manifestations of rickets. All patients had homozygous SLC2A2 gene mutations 6 had p.G162Rfs*17 (c.482_483insC), 1 had p.H192R(c.573A>G), 1 had p.R301X(c.901C>T), 1 had p.D344Y(c.1030G>T), 1 had p.H192R(c.575A>G), 1 had p.HbA1C is a key point of thinking FBS in the differential diagnosis.

**Discussion**  
FBS is a well-defined and a very rare inherited metabolic disease characterized with hepatic and renal involvement. Here, we report our single center experience of 10 patients with FBS, due to the high frequency of parental consanguinity in our region. Different from the other glycogen storage diseases and disorders of carbohydrate metabolism such as galactosemia and hereditary fructose intolerance, hyperglycemia is an important factor contributing to early proximal renal dysfunction in FBS. Six out of our 10 patients had impaired glucose tolerance. In conclusion the early occurrence of renal tubular acidosis, especially under the age of one years with elevated levels of HbA1C is a key point of thinking FBS in the differential diagnosis.

**References**  