Clinical, biochemical data and molecular analysis of a patient affected of Glut2 deficiency (Fanconi Bickel Syndrome)

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Fanconi Bickel Syndrome is a rare autosomal recessive disorder of carbohydrate metabolism with accumulation of glycogen in liver and kidney (Hepato-renal glycogenosis) or GSD XI, caused by homozygous (%75) mutation in SLC2A2 gene, a (solute carrier family 2) encodes Glut2, a low affinity facilitative glucose transporter in hepatocytes, beta cells of pancreas, enterocytes, proximal renal tubular cells. Glut2 also participates with a cotransporter for transport of glucose+Na in the small intestine and kidney.
More than 144 cases have been reported so far. Diagnosis usually made late due to diagnostic challenges. Signs and symptoms begin in the first few months of life, most present failure to thrive, glucose and galactose intolerance, fasting hypoglycemia, postprandial hyperglycemia, polyuria, rickets, short stature and hepatorenomegaly. Treatment is symptomatique, directed toward stabilization of glucose homeostasis and RTA treatment.

Case Study: 22 year old male: birth wt: 2.75kg, neonatal hyperbilirubinemia, severe galactosuria and slow growth, referred at 3 month; wt: 4kg, wide anterior fontanelle and hepatomegaly.
..BS;111,125mg/dl, normal anion gap, ABG; PH=7.22, HCo3=12.2meq/L, urinalysis; PH =7, ketone ++ high urine P/Cr, aminoaciduria, glucosuria, mild proteinuria (proximal renal tubular acidosis) and galactoseuria via TLC, Ca=9.5,P=2.7mg/d, SGOT=36, SGPT=41IU, Alkaline phosphatase:1485 IU, Galactose -1-phosphate -Uridylyl transferase; 1941 U, sufficient UDP-galactose.4.epimerase. Hand Xray: rickets, sonography: mild hepatomegaly with normal echo, bilateral kidney enlargement. the specific triad and A homozygous pathogen variant: c.1358_1359del, p.C453fs on SLC2A2 confirmed the diagnosis,
Discussion

We reported a case of Glu2 deficiency referred at 3 months because of SGA, neonatal hyperbilirubinemia, galactosuria and poor growth. On physical exams, weight 4.3 Kg, wide fontanel and hepatomegaly were discovered. A galactose restricted diet was started.

Galactosuria plus renal Fanconi syndrome, dysglycemia with normal galactose utilization enzymes, rickets and being symptomatic in early infantile with hepatorenomegaly helped us to suspect the diagnosis of hepato-renal GSD soon and to start symptomatic treatment. Whole Exome Sequencing confirmed the diagnosis.