

Segregation of a novel homozygous 6 nucleotide deletion in GLUT2 gene in a Fanconi–Bickel syndrome family

• Fanconi–Bickel syndrome (FBS) is a rare autosomal recessive disorder characterized by hepatorenal glycogen accumulation, proximal renal tubular dysfunction, impaired utilization of glucose and galactose, rickets, and severe short stature. It has been shown to be caused by mutations in GLUT2 gene, a member of the facilitative glucose transporter family. Here, we report an Iranian family with 2 affected siblings. The clinical findings in the patient include developmental delay, failure to thrive, hepatomegaly, enlarged kidneys and rickets. A novel 6 nucleotide deletion (c.1061_1066del6, p.V355_S356del2) is shown to be segregated with the disease in this family.

