Fanconi-Bickel Syndrome; FBS

Alternative Name
Hepatorenal Glycogenosis with Renal Fanconi Syndrome
Hepatic Glycogenosis with Fanconi Nephropathy
Hepatic Glycogenosis with Amino Aciduria and Glucosuria
Fanconi Syndrome with Intestinal Malabsorption and Galactose Intolerance
Pseudo-Phlorizin Diabetes
Glycogenosis, Fanconi Type
Glycogen Storage Disease XI

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic diseases Metabolic disorders; Other disorders of carbohydrate metabolism

Incidence per 100,000 Live Births
NA

OMIM Number
227810

Mode of Inheritance
Autosomal recessive

Gene Map Locus
3q26.2

Description
Fanconi Bickel disease is a glycogen storage disease (GSD) type XI. It is a rare inherited disorder which is transmitted in an autosomal recessive pattern. The exact prevalence is unknown. Affected patients have rickets, aminoaciduria, phosphaturia, growth failure, hepatomegaly, and fasting hypoglycemia. Diagnosis can be established through clinical manifestations, radiological findings revealing rickets, and from characteristic laboratory findings. Diagnosis can be confirmed by enzyme study or mutation analysis. Affected patients do not respond to glucagon, and suffer impaired galactose metabolism. Treatment is supportive through; frequent feeds by the use of slowly absorbed carbohydrates, supplementation of vitamin D and phosphate and galactose-restricted diabetic diet.

Molecular Genetics
Several mutations in the SLC2A2 gene, previously known as GLUT2, had been found to cause GSD XI. SLC2A2 gene, is located on the long arm of chromosome 3. This gene contains 11 exons and spans approximately 30Kb. The encoded protein facilitates bidirectional glucose transport.

Epidemiology in the Arab World
Saudi Arabia
Moammar et al., (2010) reviewed all patients diagnosed with inborn errors of metabolism (IEM) from 1983 to 2008 at Saudi Aramco medical facilities in the Eastern province of Saudi Arabia. During the study period, 165530 Saudi infants were born, of whom a total of 248 newborns were diagnosed with 55 IEM. Affected patients were evaluated based on clinical manifestations or family history of similar illness and/or unexplained neonatal deaths. Almost all patients were born to consanguineous parents. GSD was found in 17/248 patients. The diagnosis was confirmed in all cases of GSD by measuring the enzyme activity in leukocytes, cultured fibroblasts or liver biopsy. Among GSD cases in this cohort, a single case was found to have GSD type XI. The estimated incidence of GSD XI in this cohort was 1 in 100,000 live births.
The authors concluded that data obtained from this study underestimate the true figures of various IEM in the region. Therefore, there is an urgent need for centralized newborn screening program that utilizes tandem mass spectrometry, and offers genetic counseling for these families.

References

Moammar H, Cheriyan G, Mathew R, Al-Sannaa N. Incidence and patterns of inborn errors of metabolism in the Eastern Province of Saudi Arabia, 1983-2008. Ann Saudi Med. 2010 Jul-Aug;30(4):271-7. PMID:20622343

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2088

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