Glycogen storage disease type-I among pediatric patients in Upper Egypt


Abstract:

The aim of this study was to identify the relative frequency of pediatric patients with Glycogen storage disease type-I (GSD-I) attending outpatient's pediatric clinics or inpatients pediatric departments of Assiut, Sohag and Qena university hospitals, Upper Egypt. This case control study was carried out on 40 pediatric patients, and 40 healthy age and sex matched subjects as a control. Plasma level of lactate, biotinidase and uric acid were determined by using commercially available assay kit in both patient and control groups. Liver biopsy for histopathological examination and glucose-6-phosphatase (G6Pase) assay for patients only when needed. Plasma level of lactate, biotinidase and uric acid were significantly higher of GSD-I patients than control. The definite diagnosis of GSD-I patients was by assay of G6Pase in the biopsied liver tissue homogenates which showed statistically significant lower level when compared with the control group. The relative frequency of GSD-I in Upper Egypt is 5/100,000. It has to be kept in mind whenever failure to thrive, hepatomegaly, fasting hypoglycemia and raised aminotransferases are present. Elevated plasma biotinidase is considered better positive than negative in prediction of GSD-I with higher sensitivity and low false negative rate.

Keywords:

GSD type I, Glucose-6-phosphatase, Biotinidase

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