Glycogen storage disease type IA but not glycogen storage disease type IB is associated to an increased risk for metabolic syndrome: Possible role of microsomal glucose 6-phosphate accumulation

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Introduction: Glycogen Storage Disease type I (GSDI) is an inborn error of carbohydrate metabolism caused by mutations of either the G6PC gene (GSDIa) or the SLC37A4 gene (GSD1b). It has been shown that Glucose 6-Phosphate (G6P) availability modulates 11β-Hydroxysteroid Dehydrogenase type 1 (11βHSD1) activity, an ER-bound enzyme catalyzing the conversion of cortisone in cortisol. A possible role of 11βHSD1 in the development of the Metabolic Syndrome (MS) has been reported.

Objectives: The aim of the current study was to evaluate the prevalence of MS and the adrenal cortex function and in GSDIa and GSD1b patients.

Methods: Seventeen GSDI (10 GSDIa and 7 GSDIb) patients were enrolled. The prevalence of MS and Insulin-Resistance (IR) was evaluated. Adrenal cortex function and biochemical markers of metabolic control were also analyzed.

Results: GSDIa patients showed higher HOMA-IR (7.36±2.13 vs. 1.90±0.13, p=0.00 vs. 1.97±0.40, p=0.05) and cortisol serum levels (165.10 ng/mL ± 15.13 vs. 127.70 ng/mL ± 7.00, p=0.01 vs. 83.46 ng/mL ± 21.52, p=0.00) than controls and GSDIb patients. GSDIb showed decreased cortisol serum levels compared to controls (83.46 ng/mL ± 21.52 vs. 138.43±14.39, p=0.04).

Conclusions: Our data showed the presence of increased risk of IR and MS in GSDIa patients. We also found impaired cortisol metabolism, with opposite features in GSDIa and GSDIb patients. 11βHSD1 regulation may explain the differences between GSDIa and GSDIb patients. We suggest a routine metabolic assessment in the management of GSDI patients.

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Key points for nursing severe EV71 HFMD children patients

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Hand-Foot-Mouth-Disease (HFMD) caused by EV71 infection is a common childhood disease in recent years in Shenzhen. While most cases are self-limited with mainly skin rash, severe cases are presented in a small proportion of EV71 infected subjects. Clinically, these cases manifested with brain stem encephalitis and even death. Therefore, early diagnosis of severe cases is critical for successful treatment and consequently to reduce mortality. During 2012 to 2014, over than 2310 cases of EV71 HFMD have been hospitalized in our hospital. Among them, 267 cases were diagnosed as severe HFMD. The treatment success rate is significantly higher than the average of whole country. The clinical manifestations including skin rash, fever, vital signs, and consciousness were the key point for intensive monitoring. Clinically, continued high fever (>39°C), increased blood pressure (systolic blood pressure >118 MMHG), small and hidden skin rash, facial or lip Angle jitter were important clinical manifestations for early diagnosis and/or prediction of severe cases, of which required early intervention.

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