1094

THE STUDY OF THE GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY IN CROATIA

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Conflict of Interest: None declared
Acknowledgement: None declared
Founding Source: None declared
Encore Abstract: Not applicable

Aim: To determine prevalence, clinical manifestations and genetic analysis of the glucose-6-phosphate dehydrogenase (G6PD) deficiency in Croatia.

Materials and methods: The fluorescent spot test was used to screen 2726 high school students to screen blood samples of 513 male children which had neonatal hyperbilirubinemia of the unclear cause. Fluorescence reading was performed at the beginning and in the 3rd, 6th, 10th and 25th minutes of incubation. All positive samples were quantitatively measured using spectrophotometric method. Genetic analysis was performed on 24 unrelated subjects.
Results: G6PD deficiency was found in 12 (0.44%) of the all school students. The incidence was 0.75% in males, and 0.14% in females. 20 samples (3,9%) of the children with neonatal hyperbilirubinemia were G6PD deficient. The acute haemolytic anaemia was observed after consumption of fava beans. Molecular testing revealed mutations: Cosenza 9(37.5%), Mediterranean 4 (16.6%), Seattle 3(12.5%), Union 3(12.5%) and Cassano 1(4.2%) and novel variant Split 1(4.2%). Three samples remained uncharacterized. All G6PD Mediterranean mutations had concomitant silent C→T transition at the position 1311.
Conclusions: The prevalence of G6PD deficiency in the Croatian population is 0.44%. The prevalence rate among newborns with neonatal jaundice is significantly higher. A novel G6PD mutation was identified. The results of G6PD mutations are unusual for Mediterranean basin in that G6PD Cosenza, rather than G6PD Mediterranean, is the predominant variant. The fluorescent spot test for moderate enzyme deficiency is reliable in the case of early fluorescence reading.