BMC Res Notes. 2011 Oct 24;4:436.

Molecular characterization of glucose-6-phosphate dehydrogenase deficiency in Jeddah, Kingdom of Saudi Arabia.

Al-Jaouni SK, Jarullah J, Azhar E, Moradkhani K.

Hematology Research Lab, King Fahd Medical Research Centre, Faculty of Medicine, King Abdulaziz University, P,O, Box 80215, Jeddah 21589, Kingdom of Saudi Arabia. dr.jaouni@gmail.com.

Abstract

BACKGROUND:

The development of polymerase chain reaction (PCR)-based methods for the detection of known mutations has facilitated detecting specific red blood cell (RBC) enzyme deficiencies. We carried out a study on glucose-6-phosphate dehydrogenase (G6PD) deficient subjects in Jeddah to evaluate the molecular characteristics of this enzyme deficiency and the frequency of nucleotide1311 and IVS-XI-93 polymorphisms in the glucose-6-phosphate dehydrogenase gene.

RESULTS:

A total of 1584 unrelated Saudis (984 neonates and 600 adults) were screened for glucose-6-phosphate dehydrogenase deficiency. The prevalence of glucose-6-phosphate dehydrogenase deficiency was 6.9% (n = 110). G6PD Mediterranean mutation was observed in 98 (89.1%) cases, G6PD Aures in 11 (10.0%) cases, and G6PD Chatham in 1 (0.9%) case. None of the samples showed G6PD A^- mutation. Samples from 29 deficient subjects (25 males and 4 females) were examined for polymorphism. The association of two polymorphisms of exon/intron 11 (c.1311T/IVS-XI-93C) was observed in 14 (42.4%) of 33 chromosomes studied. This association was found in 9 (31.0%) carriers of G6PD Mediterranean and in 4 (13.8%) carriers of G6PD Aures.

CONCLUSIONS:

The majority of mutations were G6PD Mediterranean, followed by G6PD Aures and < 1% G6PD Chatham. We conclude that 1311T is a frequent polymorphism in subjects with G6PD Mediterranean and Aures variants in Jeddah