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Frequency of Mediterranean mutation among a group of Saudi G6PD patients in Western region-Jeddah

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Abstract

Glucose-6-phosphate dehydrogenase deficiency (G6PD), a common human enzymatic defects characterized by extreme molecular and biochemical heterogeneity is found to have a variable frequency in different regions. The molecular basis of polymorphic variants in Saudi Arabia have yet to be fully addressed to. Accordingly, a study wasdesigned to determine the frequency of G6PD gene mutations in G6PD deficient cases. From forty-seven unrelated G6PD-deficient subjects, DNA was extracted individually from peripheral blood samples and exons 6 and 7 of the G6PD gene were amplified by PCR. Mutation analysis was carried out by using conformation sensitive gel electrophoresis (CSGE), followed by direct DNA sequencing. The results showed definite altered CSGE patterns. Two mutations were resolved in exon 6 of G6PD gene; Mediterranean mutation and Sibari mutation, not previously reported so far; while no mutation was detected in exon 7. The frequency of exons 6 mutations responsible for G6PD deficiency (Mediterranean type) is reported for the first time from this region, with a figure of 50.1%. The absence of other mutations in exon 7 causing G6PD deficiency points to the low genetic diversity in the studied population.

Author Keywords

CSGE; G6PD; Hemolytic anemia; Mediterranean mutation; Molecular variants; PCR