Bahrain Medical Bulletin, Vol. 26, No. 4, December 2004

Molecular Homogeneity of G6PD Deficiency

Nabeel Al Momen* Sheikha S Al Arrayed**
Ahmed Al Alawi A*

Objectives: To study the molecular basis of G6PD deficiency in the Kingdom of Bahrain. Further emphasis will be presented on the genetic polymorphism at nucleotide 1311 for both normal and deficient subjects.

Methods: DNA extraction was done for 83 G6PD-deficient subjects and 80 normal controls. A combination of PCR-RFLP and PCR-DGGE procedures were employed to uncover the sequence variations at nt 563 (C/T) (G6PD Mediterranean) and at nt 1311 (C/T) polymorphism in both subjects with deficient and normal G6PD activity.

Results: Ninety-one percent (93/102) of the X chromosomes from G6PD deficient patients had nt 563 (C-T) mutation (G6PD Med), whereas ~9% of the X chromosomes from G6PD deficient subjects might have other G6PD variant(s) (or normal X chromosomes in heterozygote females). Niney-six percent (89/93) of the G6PD Med-bearing X chromosomes showed thymine (T) at nucleotide position 1311. In contrast, 70% (82/117) of the normal X chromosomes showed cytosine (C) at nucleotide position 1311, while it was thymine (T) in 30% (35/117) of the normal X chromosomes.

Conclusion: The vast majority (91%) of X chromosomes from G6PD-deficient subjects in Bahrain are harboring nt 563 (C-T) mutation (G6PD Mediterranean). The G6PD Med variant in Bahrain is in tight linkage disequilibrium with thymine (T) at nt 1311. These data, collectively, revealed high moleculer homogeneity of G6PD deficiency in Bahrain.

Further studies are needed to uncover factor(s) contributing to heterogeneous phenotypic expression of the disease in Bahrain.