

The Viangchan-G6PD Mutation in Vietnamese-Kinh

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ABSTRACT As the most common missense mutation cause of G6PD deficiency in Vietnamese, Viangchan [Val²⁰¹Met] mutation can be used as the marker for development of the molecular diagnostic to diagnose this disease. To confirm Viangchan as the marker for diagnosis a case/control study is carried out. This study confirmed the role of Viangchan mutation in G6PD deficiency in Vietnamese and indicate the necessary of development of new molecular method for diagnosis of the diseases replace the fluorescence test recently as their limitation in identify G6PD patients. An AMRS-PCR was designed to screen Viangchan mutation in a 318 cases and 210 controls sample set. The result showed that Viangchan mutation occupied 26.7% in the population in this study, while in the G6PD deficient population it occupied 24%. The association analysis showed that the risk allele A in this mutation is strongly associated with G6PD deficiency in Vietnamese Kinh population (OR = 42.8; 95% CI [13.5-135.5]; p<0.0001). The genotypes which contained risk alleles were also strongly associated with the disease (OR=22.4; 95% CI [6.96-71.99]; p<0.0001). This study confirmed that the Viangchan is the most common causative mutation for G6PD deficiency in Vietnamese Kinh. Thus this mutation can be used as the indicator for diagnosis of G6PD in Vietnamese Kinh.