Molecular Variants of Red Cell Glucose-6-Phosphate Dehydrogenase Deficiency in Central Java, Indonesia

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Hum Hered 1995;45:346-350 (DOI: 10.1159/000154303)

Key Words : Population genetics, Mediterranean mutation, Mongoloid mutation, Silent mutation, Mutagenesis, Polymerase chain reaction

Abstract
One hundred and sixty-nine Javanese males were screened for the presence of red cell glucose-6-phosphate dehydrogenase (G6PD) variants by a dye decoloration screening test and starch gel electrophoresis. The frequency of G6PD deficiency was 14%. Three non-deficient electrophoretic variants with mobilities of 95, 105 and 107% of GdB+ were encountered. Sixteen G6PD-deficient subjects were further investigated for the presence of mutations at nt95 A G, nt487 G A, nt493 A G, nt563 C T, nt1024 C T, nt1376 G T, nt1388 G A and the silent mutation (nt1311 C T) of the G6PD gene by natural or artificially created amplified restriction sites. They were identified by the polymerase chain reaction and electrophoresis of restriction-digested products. Five subjects had the Mediterranean mutation (nt563 C T), but only one had simultaneous presence of nt1311(T). The next common mutations were 1376(T) in three subjects and 487(A) in two subjects. Five of the sixteen subjects had the nt 1311(T) mutation giving an overall frequency of 0.31. The other four mutations were absent in this population sample.