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GLUCOSE-6-PHOSPHATE DEHYDROGENASE MUTATIONS AMONG CANTONESE REVEALED BY POLYMERASE CHAIN REACTION USING DRIED BLOOD SPOTS

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Glucose-6-phosphate dehydrogenase (G6PD) deficiency is very common in south China, especially in Guangdong province. Seven different types of G6PD gene mutations have been found in Cantonese, but the mutation frequencies are not clear. Since 1990, neonatal screening for G6PD deficiency has been conducted in Guangzhou city, Guangdong province, and a high incidence of 3.6% is found. Dried blood spots collected on neonatal screening paper were used in this study. PCR products were amplified directly from blood spots followed by digestion with a restriction enzyme that recognize the mutant and the amplificati created sites. 169 samples of male G6PD-deficient newborns were analyzed. The results showed that 72 (42.6%) were G→T mutation at nucleotide 1376 (G6PD 1376 G→T), 35 (20.7%) were G6PD 1388 G→A, 30 (17.7%) were G6PD 95 A→G, 6 (3.6%) were G6PD 392 G→T, 3 (1.8%) were G6PD 1024 G→T, and 23 (13.6%) were not one of the 5 common mutations. Our results indicate that G6PD 1376 G→T is the most common mutation in Cantonese, and the former 3 mutations account for more than 70% of G6PD deficiency cases, as reported in similar studies in Taiwan. But the frequency of G6PD 95 A→G mutation is much higher among Catonese, and in the 169 samples studied, we did not detect any G6PD 493 A→G and G6PD 487 G→A mutations which have been reported in Taiwan.

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Denaturing gradient gel electrophoresis (DGGE): Rapid detection of common Glucose-6-phosphate Dehydrogenase (G6PD) variations and screening for new mutations

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The ability to detect variations is fundamentally important for molecular diagnosis of genetic diseases. Of the many techniques available, DGGE when used in conjunction with PCR, could achieve 100% detection.

We describe the use of DGGE to detect the most common Chinese G6PD variants: at: G->T at nt 1376, G->A at 1388 (both in exon 12), A->G at nt 95 in exon 02 and the DNA polymorphism at 13bp upstream in intron 11. In each case, the mutant alleles resolve well from the normal allele(s). The distinct hetero-duplex bands are characteristic of a particular genotype suggesting that this feature is very useful for identifying heterozygous carriers and at risk families. When DGGE is extended to cover the other exons, it can scan the gene for new variation(s) and together with direct sequencing, leads to the identification of new variation/mutation(s). We identified a mutation at nt 871 which had not been reported in Hong Kong. Since it can rapidly screen many unknown samples, this approach would be useful for determining the frequencies of these G6PD variations in other ethnic populations.

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