Journal of Biomedical & Laboratory Sciences
Volume 5, Number 2, April, 1993 : S10

Analysis of Common Glucos-6-Phosphate Dehydrogenase Mutations in Chinese by Polymerase Chain Reaction Using Dried Blood Spots Collected on Filter Paper

YM Hung[1]*, CC Yang[1], SH Chiang[1], SJ Wu[1], TK Tang[3] and KJ Hsiao[1,2]

Department of Medical Research[1], Veterans General Hospital Taipei, Institute of Genetics[2], National Yang-Ming Medical College; Institute of Biomedical Sciences[3], Academia Sinica; Taipei, Taiwan, R.O.C.

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common known enzymopathy affecting 400 million people worldwide. There is a high frequency (~2%) of G6PD deficiency in Taiwan and the mutations of 1376 G→A, 1388 G→A, 493 A→G, and 1024 C→T have been reported to account for approximate 80% of G6PD mutant alleles in Chinese. A method to detect these G6PD mutations from whole blood by polymerase chain reaction (PCR) with mismatch primers was reported (Blood 1992; 80: 1079). Dried blood spots collected on filter paper have been used in neonatal screening for detecting G6PD deficiency and other congenital metabolic diseases in Taiwan. Because the dried blood spots collected on filter paper are easy to be transported and stored, a method to amplify DNA from it by PCR was developed in this study. Dried blood spots specimens from forty-nine G6PD deficient patients detected by our neonatal screening program were collected. These four common G6PD mutations were analyzed by PCR with mismatch primers which were designed as previous described (Blood, 1992). Among the 49 G6PD patients, twenty-two "1376" (44.9%), six "1388" (12.2%), six "493" (12.2%), and four "1024" (8.2%) mutations were identified. These mutations frequencies are similar to other studies reported in Taiwan. This easy and non-radioactive method provides a way to confirm the positive G6PD deficient cases detected by the neonatal screening test using the same screening dried blood spots specimens. And the detecting G6PD mutations by PCR with mismatch primers could also be applied to detect heterozygotes of G6PD deficiency for genetic counseling. In addition, this method provides a way to collect samples for large scale epidemiological study of the G6PD mutations in different populations.

(14)