◎P-56

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

D.J.Tang[1], X.Q.Ma[1]*, C.Y.Song[1], B.H.Lin[1], S.J. Wu[2], S.H. Chiang[2], K.J. Hsiao[2,3]
[1]Neonatal Screening Centre,Guangzhou Maternal & Neonatal Hospital,China;[2]Dept.of Med.Res. Veterans General Hospital-Taipei; [3]Inst.of Genetics,National YangMing Univ.,Taipei,Taiwan

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 amplification created sites. 169 samples of male G6PD deficient newborns were analyzed. The results showed that 72 (42.6%) were G→T mutation at necleotide 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.

1996;P56