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Application of Dried Blood Spots Collected on Filter Paper for Screening of Maternal β-Thalassemia Carrier

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Because of relative high incidence (1-3%) of β-thalassemia carrier in Taiwan, a mass screening program directing to prospective prevention of homozygous β-thalassemia is strongly indicated. Since HbA_2 is increased in β-thalassemia carrier, a method to determine HbA_2% in the dried blood spots collected on filter paper was developed by our laboratory in 1991. The HbA2 eluated from the blood spot was determined by microchromatography.

From March to Sept. 1992, We conducted a pilot maternal β-thalassemia screening program in Taitung County and Nantun County. Total of 2,073 pregnant women were screened using the filter paper blood collecting technique, Out of those screened, 93 cases (4.5%) were positive ($HbA_2\% \geq 3.2\%$). 88 cases (94.6%) of the positive cases were recalled successfully. 38 of them were confirmed to be β-thalassemia carrier. The screening HbA_2% values of confirmed carriers were between 3.4% and 5.4%. The range of MCV and MCH of confirmed carriers were reported from 58 to 87 fl and 18.5 to 31 pg, respectively. The incidence of β-thalassemia was estimated to be around 1.9%. In order to confirm the screening results, DNA of the dried blood spots collected from the β-thalassemia carrier were amplified by polymerase chain reaction (PCR) and then hybridized with allele specific oligonucleotide (ASO) probes for detecting IVS-II654, 41/42 frameshift, codon 17 and TATA-28 mutations, wich were reported to be the common β-thalassemia mutations in southern Chinese. Among the 38 carrier cases analyzed, 34 cases (89.5%) were found having one of these four mutation types. In this study, a public health and medical services network was incorporated in the screening system. The results indicate that such a network may work well even in rural areas of Taiwan.

(142)