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SCREENING FOR ß-THALASSEMIA CARRIER USING DRIED BLOOD SPOTS COLLECTED ON FILTER PAPER

Hsiao KJ[1,2], Chen CY[1], Wu SJ[2], Yang ML[3].
Institute of Genetics[1], National Yang-Ming Medical College; Depts. of Medical Research[2] and Obst. & Gyn[3]., Veterans General Hospital-Taipei; Taipei, Taiwan, Republic of China.

Because of relative high incidence of ß-thalassemia carrier in Taiwan, a mass screening program directed to prospective prevention of homozygous ß-thalassemia was strongly indicated. Since Hb A_2 is increased in ß-thalassemia carrier, a method to determine Hb A_2% in the dried blood spots on filter paper was studied. The Hb A_2 eluted from the blood spot was determined by microchromatography. The linearity (R=0.996), precision (C.V. of within run: 5-8 %, run to run: 9-12 %), stability (7 days at 25° C, 80 days at -20° C), and recovery (91-95%) of this method revealed that filter paper offered a convenient and reliable method in transferring blood for HbA_2% determination. The level of Hb A_2% in whole blood and dried blood spots collected from 57 habitants in an aboriginal area (Hsiulin District, Hualien County) and 489 pregnant women from the out patient department of Veterans General Hospital, Taipei, were determined. The results from whole blood and dried blood spots were found in good correlation. Hb A_2% in those subjects screened showed a clear cutting value of 3.5%, which was then set to detect ß-thalassemia carrier. The carrier rate was determined to be 1.6% for the both populations. In order to confirm the screening results, the ß globin gene of the dried blood spots collected from the ß-thalassemia carrier was amplified by polymerase chain reaction and then hybridized with allele specific oligonucleotide probes for detecting IVS-2 654, 41/42 frameshift, codon 17 and TATA-28 mutations. These four mutations were found to cover over 87% of the ß-thalassemia carriers detected by us. This study presented an approach for mass screening of ß-thalassemia carrier by detecting Hb A_2% in dried blood spots. The method developed could be easily incorporated into the dried blood spot maternal alfafetoprotein screening program which has been on trial in the rural areas in Taiwan since 1988.