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ⓞ 137 ß-THALASSEMIA CARRIER SCREENING IN HSIULIN DISTRICT OF HUALIEN COUNTY.

C.Y. Chen*[1], K.F. Wu[3], I.F. Lou[3], B.H.May[4] K.J. Hsiao[1,2,3]. Institutes of Genetics[1] and Biochemistry[2], National Yang-Ming Medical College; Department of Medical Research[3], Veterans General Hospital-Taipei; Department of Gastroenterology[4], Mennonite Christian Hospital-Hualien; Taiwan, R.O.C.

In order to study the ß-thalassemia carrier rate in Tai Ya aborigine region, 434 cases were collected from approximate 1000 adults (age older than 35 years) living in the Hsiulin District of Hualien County and their ß-thalassemia carrier status were screened. Their blood samples were collected in EDTA and measured for the mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH) values using Coulter Counter (model S) in the same day. Hemoglobin A_2% was determined by DE-52 microchromatography (within run C.V.=5-8%; run to run C.V.= 6-13%) if MCH and/or MCH values of the sample was below normal reference range(MCV=80-100 fl; MCH =27-32 pg). Seventeen cases were found with MCV and/or MCH values lower than the normal reference range. Seven of the seventeen cases had hemoglobin A_2% (range:3.8-8.2) higher than normal (reference range:0.9-3.5%) . Hemoglobin A_2% of fifty random control cases with normal MCV and MCH were found in the range of 1.2-3.0 %. From these data the ß-thalassemia carrier rate in Tai Ya aborigine region was estimated to be around 1.6%, which is similar to that reported in Guangxi (1.52%), Fujian (1.83%) and Silk Road region (1.62%). The ß-globin gene mutations in these seven ß-thalassemia carrier were studied by polymerase chain reaction(PCR) amplification of their ß-globin gene from their blood genomic DNA and indentified with allele specific oligonucleotide probes (ASO). Four of them found to be 41/42 frameshift, two of them are IVS-2 654-RNA processing mutant. The results indicate that the 41/42 frameshift is the major ß-thalassemia mutant type in this region.