

COMMON GENETIC VARIATION IN ENDOTHELIN RECEPTOR TYPE A IS ASSOCIATED WITH SERUM HEMOGLOBIN LEVEL: TAICHUNG COMMUNITY HEALTH STUDY FOR ELDER (TCHS-E)

在 EDNRA 基因上的常見變異會影響血清中血紅素之濃度

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Background: Hemoglobin is the iron-containing oxygen-transport metalloprotein in the red blood cells, where it releases the oxygen to burn nutrients to provide energy to power the functions of the organism, and collects the resultant carbon dioxide to bring it back to the respiratory organs to be dispensed from the organism. In humans, endothelin receptor type A (*EDNRA*) gene is located on chromosome 4. Its product, endothelin receptor A, is expressed in vascular smooth muscle cells, and its activation by Endothelin-1 (ET-1), a vasoconstrictor, leads to vasoconstriction. The purpose of this study was to determine if single nucleotide polymorphism (SNP) of *EDNRA*, rs5335 is associated with hemoglobin in Taiwanese elders.

Methods: SNP rs5335 of *EDNRA* of a total of 471 unrelated elders (251 males and 221 females) were genotyped. This SNP have two alleles, C and G, resulting in three genotypes, C homozygotes (CC), heterozygotes (CG), and G homozygotes (GG). Linkage disequilibrium (LD) was analyzed for this SNP. Serum hemoglobin concentration was analyzed by a biochemical autoanalyzer (Beckman Cou, Fullerton, CA, USA) and low hemoglobin was defined as hemoglobin <14 mg/dl for male and <12 mg/dl for female (121 elders as low

hemoglobin and 350 elders as normal hemoglobin).

Results: The minor allele frequency for rs5335 was C with a proportion of 0.4223. After adjusting for age and gender, our study indicates that SNP rs5335 C/G genotype was significantly associated with decreased hemoglobin level ($\beta = -0.27$ mg/dL, $p < 0.05$). In addition, the adjusted odds ratios of low hemoglobin were 2.35 (95% CI: 1.35-4.10) among elders with SNP rs5335 C/G genotypes compared with elders with C/C genotype.

Conclusion: We conclude that polymorphism rs5335 in the *EDNRA* gene affects hemoglobin, indicating rs5335 appear to be a susceptibility biomarker of hemoglobin. But further study may be required.