Mass Screening of Carnitine Palmitoyltransferase II Deficiency ; Its Significance as an Early Diagnosis of Sport-Related Hematuria and Rhabdomyolysis

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Abstract : Sport hematuria is recognized as a transient and benign disorder. However, reports have been accumulated that hard physical trainings, exhausting sports, military exercises, and severe infections resulted in rhabdomyolysis and, if not appropriately managed, in acute renal failure. The reports also demonstrated those patients deficient in carnitine palmitoyltransferase II (CPT II) could not generate sufficient amount of energy through β-oxidation of long-chain fatty acids, which resulted in the repetitive non-traumatic rhabdomyolysis. In this study, we tried to establish methods to genetically screen out potential patients with CPT II deficiency from athlete students quickly and easily. Three hundred and sixty-eight sport students were enrolled in this study with informed consents. Before and after wearing summer training camps, they were subjected to a dipstick urinalysis and urinary secretions of protein, creatinine, and myoglobin. Twenty-eight students secreted high amount of myoglobin. Four out of the 28 agreed with the genetic diagnosis of CPT II deficiency. Eight pairs of PCR primers were designed to cover the coding region. Each PCR-amplified gene product was subjected to a direct DNA sequencing. No students carried any mutation related to the disorder.

Key words : **Sport hematuria, Rhabdomyolysis, Carnitine palmitoyltransferase II deficiency, Denaturing high performance liquid chromatography, Direct DNA sequencing, SNPs**