中文題目：*HSD*3β 基因多型性在原發性皮質醛酮症病人扮演的角色

英文題目：*HSD*3β Gene Polymorphisms and Primary Aldosteronism

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Objects：The *HSD3*β pathway plays a pivotal role in aldosterone synthesis. This study aims to determine the total contributions of *HSD3*β and *HSD3*β2 genes to primary aldosteronism (PA) using a tagSNP approach.

Methods：In a case-control cohort, 688 consecutive ethnically matched unrelated individuals comprising 362 PA and 326 essential hypertension were recruited. Nineteen tag single nucleotide polymorphisms across the *HSD3*β1, *HSD3*β2 and CYP11b2 were genotyped. Expression of HSD*3*β mRNA and immunoblot analysis to the specimens of aldosterone-producing adenoma (APA) were compared with non-functional incidentaloma.

Results：The AG heterozygotes (OR= 1.92, 95%CI, 1.13-3.32, p= 0.018, corresponding a population attributable risk fraction (PAF), 6.7%) of rs12410453 and CC homozygotes (OR= 2.21, 95%CI, 1.28-3.95, p= 0.006, PAF 30.7%) of rs6203 were related to PA. The expression of *HSD3*β1 mRNA, *HSD3*β2 mRNA and *HSD3*β protein increased in APA, in comparison with incidentaloma. The interaction between rs6203 and rs12410453 polymorphisms had synergistic effect on the increased plasma aldosterone to renin ratio. The haplotypes in a LD block containing rs6203 were significantly associated with serum potassium level (OR, 1.24; 95% CI, 1.02-1.50).

Conclusions：We identify risk-conferring genetic variations in HSD3β gene influences susceptibility to PA. Concomitant presence of rs6203 CC and rs12410453 GA genotypes synergistically increased the predisposition to PA. Correlation between allele of the H*SD3*β gene and raised ARR likely serve a role in PA management.