Kunlun He et al., J Clin Exp Pathol 2017, 7:2 (Suppl)
DOI: 10.4172/2161-0681-C1-034

Mutation in *APOA5* gene associated with hypertriglyceridemia

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It is well accepted that the serum lipid level is modulated by genetic and environmental factors. Therefore, identification of the genetic variations involved in lipid metabolism could provide a clue to search for novel pathway in lipid regulation and thereby new therapeutic or preventive methods for coronary artery disease, and further improve the prognosis of heart failure and other cardiovascular disease. We extensively resequenced of our candidate genes and evaluated of rare variant accumulation to identify additional genetic variation responsible for increasing susceptibility to human hyperlipidemia. This study included 638 Chinese patients who were admitted to the Department of Cardiology, Chinese PLA General Hospital (Beijing, China) with chronic heart failure between January 2011 and January 2013. In total, 392 adult patients with hyperlipidemia and 246 population-based controls without hyperlipidemia were included in this study. In order to make the best possible to identify putatively damaging SNVs, five protein prediction algorithms (LRT score, MutationTaster, PolyPhen-2 HumDiv, PolyPhen-2HumVar and SIFT) were applied in non-synonymous SNVs. To explore the potential biological effects of the associated SNPs, we tested whether they were overlapped or correlated with the expression quantitative trait locus from four databases in peripheral blood cells. To mine the potential epistasis effects of the associated SNPs, we exhaustively searched all pairs among the variants. This high depth of resequencing study based on target genes repeatedly verified of two common mutation in *APOA5* gene associated with hypertriglyceridemia (11-116661392, 11-116662579). Besides, 3 rare variants in *APOA5* were also found to be related with the increase of triglycerides level. We detect the interaction effect across all pathways and discovered a stronger epistasis effect between gene *CNDP1* and *APOA5*, which both of them came from metabolic process and explained 7.55% genetic variance. We found common and rare variants associated with high triglyceride levels through the high depth of resequencing strategy. Much stronger associated signals are needed to excavate with large sample size and multicenter studies. Meanwhile, we also need studies of muti-omics to detect pathogenic mechanism of pathogenic variants.

Biography

Kunlun He has completed his Medical School degree from the Third Military Medical University, PhD from Chinese PLA Medical School, and Postdoctoral studies from College of Physicians and Surgeons of Columbia University. He is the Vice President of Chinese PLA General Hospital, and the Professor of Department of Cardiology. In recent years, he focuses on Translational Medicine of Cardiovascular disease. He has published more than 158 peer reviewed papers, achieved the first class awards of Beijing Science and Technology, and also he has been serving as an Editorial Board Member of three medical journals.

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