Extended Abstract Journal of Molecular and Cellular Biochemistry 2021
Vol.5 No.3

Efficient genome-wide association studies and post- GWAS integrative analyses for human cancer and neurodegenerative diseases

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Abstract

It is evident that in etiologies of human complex diseases, genetic factors play some important roles. Genome-wide association study (GWAS) is a standard technique to identify heritable genetic basis of complex diseases. In relation with GWAS, there exist some challenges in selecting input samples completely randomly, to biologically describe GWAS results, to translate them into clinical benefits and to compare germ line variants achieved from GWAS with somatic mutations in creating, development and treatment of human complex diseases. Likelihood-based statistical methods are robust in estimating linkage disequilibrium when factors like non-randomness and population structures exist. Then the results of GWAS can be used for post-GWAS analyses to predict multiple biological components like genes, non-coding RNAs and transcription factor binding sites in association with complex diseases. An integrative analysis seeks to pool information from multiple GWAS results, somatic mutations and genetic drug targets of human complex disorders and the results of such analysis can provide new insight into the genetic and treatments of complex diseases.

This presentation is prepared from the viewpoint that the robust statistical methods can be applied to arrive at valuable results from GWAS and that primarily genetic information derived from GWAS is subject to further post-GWAS analysis to provide more biologically informative results in relation with genetics of human complex diseases that can be applied to real time clinical applications. Then the results of such analyses can be used to discuss and compare human cancers and neurodegenerative diseases from a genetic perspective.

We concluded that in spite of the differences between human cancers and neurodegenerative diseases, the roles of germ line and somatic mutations in creating, developments and treatments of those two kinds of human complex diseases are similar.

This work is partly presented at International Conference on Biotechnology, Biomarkers & Systems Biology March 04-05, 2019 at Amsterdam, Netherlands