Abstract of Presentation

Note: This paper should be typed in "Times New Roman" of 12pt.

Presentation Title(Should be no more than 20 words):

Analysis of genome-wide data in common diseases in Japan

Abstract :

Methods in studies to identify disease-related genes have changed dramatically from candidate gene approach to genome-wide approach. In the former, biochemistry and molecular biology played principal roles; however, in the latter, statistics and genetics play those roles. Among a few categories in the latter approach, genome-wide association study (GWAS) is a powerful method to identify associations between genetic variations and traits. In GWAS, more than 100,000 SNP genotypes are obtained from a subject, and both genotype and phenotype data from many subjects are analyzed statistically. This approach was first reported from RIKEN in 2002 and has been accepted as a powerful method in medicine. RIKEN has identified many disease-associated genetic variations by applying both newly generated programs and the programs available from other researchers to large-size data. Recently we have extended the targets of the study from qualitative traits like diseases to quantitative traits. Such traits include physical traits like height, biochemical traits like serum urate concentration and hematological traits like white blood cell count. The combinations of the associations between genetic variations and diseases with those between genetic variations and the quantitative clinical data are likely to construct more accurate and robust algorithm to predict the phenotypes of individuals and to predict the impact of medical interventions targeted at the products of the associated variations. Although the quantitative traits so far used are the transcriptomics data as well as laboratory data obtained in routine clinical practice, other traits such as proteomics and metabolomics data will be treated quantitatively just as the clinical laboratory data. Integration of the results from various levels of human biological process will lead to the more accurate prediction of the phenotypes of individuals and outcomes of medical interventions.