



Korean Genome Analysis Project: Past, Present, and Future

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As an international cooperation, Human Genome Project had completed the human genome sequencing in 2003 and this great achievement made it possible the integrated view of cell, organ, and human by the systemic whole-genome study of DNA (mainly variations), transcripts, and proteins. During the Project NIH announced the significance of SNP (single nucleotide polymorphism) in disease and the next year in 1998 The SNP Consortium was organized to detect SNP in a large scale. In the same year Nakamura's group in Japan had sequenced whole genes to discover SNPs in Japanese and enrolled more than 100,000 Japanese SNPs in DB.

Center for Genome Science in Korea NIH launched four years ago has aims to realize the result of Human Genome Project by focusing on 1) the collection and management of Korean samples useful for the genomic study such as DNA, urine, and serum, 2) the collection and database of the epidemiological and clinical information of those subjects, and 3) the identification of Korean genetic variations and their relationship to diseases. At present more than 40,000 Korean samples were collected and the clinical data has been analyzed to identify the risk factors for diseases, especially 10,000 Korean among those has been examined as a prospective cohort starting from 2001. Korean Genome Analysis Project carried out in the Institute aims to collect the genetic variations in the Korean population and identify the variations related to the diseases, mostly complex traits such as diabetes, hypertension, asthma, osteoporosis etc. About 700 genes were sequenced in the 24 Korean individuals and more than 10,000 SNPs were identified so far. The association study of those genes has being investigated to the complex diseases in a collaboration with 12 Disease Genome Research Centers based on university hospital. However, the complexity of multifactorial diseases makes it difficult to clarify the genetic risk factors by conventional regression analysis and several pits and falls and future strategy will be discussed.