S5-1

Genetic Determinants of Lipid Metabolism and Clinical Applications

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Serum lipid concentrations are one of the major risk factors for CHD. LDL, HDL cholesterol levels are influenced by both genetic and environmental factors. Until now, major mutation has been described in lipoprotein, apoE, apo B, as well as ABCA1, CETP gene. Apo E genotype was world widely examined and E4 allele is associated with elevations in LDL cholesterol and incidence of CHD. 219K polymorphism of ABCA1 gene is associated with increasing HDL cholesterol. But the frequency of several mutation at these candidate gene loci in our cardiovascular genome study are displayed strong ethnic difference from that of Caucasians. Family-based linkage anlysis is well known as an informative approach and provides an opportunity to search for new gene loci. We collected complex families composed of CAD individuals and analyzed mathematical models of lipid traits. And then, linkage analysis was performed by used genetic markers. The results showed tentative linkage between LDL cholesterol and Apo AI-CIII-AIV gene cluster. Currently, SNP haplotype or combined genotype analysis is reported to be more proper method than each SNPs. In conclusion, genetic study related to lipid metabolism is one of the important approaches to prevent of CHD incidence and provide the accurate therapy. In the futher study, it is necessary that gene-gene and gene-environmental interaction should be considered.

S5-2

Identification of clinically useful genes and SNPs of skeletal diseases in Korea

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The rapid increase of the elderly people over 65 years old in Korea will result in the estimated number of 14% of total population in 2022. One of the prominent features of this aging or aged society from the medical aspects means a rapid increase of skeletal diseases including osteoporosis and arthritis. Therefore, it is crucial to prepare some preventive means to figure out health problems related to skeletal diseases in this coming aged society. With the aim of overcoming this problem, the skeletal diseases genome research center (SDGRC) was launched in Kyungpook University Hospital from June 1, 2001. Our goal is to identify genes and SNPs which are useful for early diagnosis and treatment of skeletal diseases. For the first three years, our project focuses on identification of genes and SNPs for diseases. For the next three years, we will characterize the functions of these genes and SNPs. For the final phase, we will concentrate on clinical application of our research works. The project is composed of five different parts interrelating with each other. Two parts of the project focus on collecting DNA and cells from patients with osteoporosis or arthritis. Other three parts of project deal with the identification of important genes and SNPs which are related to the mechanism of bone formation, cartilage maintenance and bone resorption. The SDGRC requires a wide range of involvement of scientists with materials and core facilities and the project is running with interdisciplinary collaboration. The successful performance of our project will provide the opportunity for better health and better quality of life for the elderly people. The outcome of this ongoing project will be presented.