Community-Based Cohort Study for Genome Epidemiology in Cardiovascular Disease

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Cardiovascular disease (CVD) is estimated to cause approximately a fourth of all deaths in Korea. Ischemic heart disease and cerebral infarction, which are related to atherosclerosis, have been dramatically increasing in recent years. CVD is a typical example of complex disorder, and consists of diverse diseases which are correlated with each other. Those diseases have various risk factors; each risk factor is related to develop various diseases. A line of studies have reported that cardiovascular disease incorporates with more than 200 risk factors and physiological markers.

Risk factors of cardiovascular diseases can be categorized as 1) biological characteristics, such as gender, age, infection, and genetic factor, 2) behavioral factor, including smoking, drinking, dietary pattern, physical activity, and socio-psychological factor, and 3) socio-geographical environment. Those factors could affect physiological marker, such as blood pressure, glucose, lipid, insulin, which causes the intermediate outcome, involving hypertension, diabetes, and atherosclerosis. These will consequently lead to ischemic heart disease or cerebrovascular disease. Pathophysiology of atherosclerosis is mainly explained by 1) injury of blood vessels, 2) inflammatory reaction, and 3) oxidation response mechanism. Along this line, gene polymorphisms associated with these three mechanisms, such as coagulation factor, plasminogen activator inhibitor, apolipoprotein, lipoprotein lipase, glucokinase, homocysteine, angiotensinogen, angiotensin converting enzyme, is noted.

The advance in molecular biology made it possible to evaluate more accurately the gene polymorphism, and to assess the intermediary process of diseases. This also contributed to the incorporation with epidemiologic studies. Through this genome epidemiological study, people could understand the relationship between genetic and environment interaction in the context of the cause and mechanism of cardiovascular disease. This in turns led to suggest the specified guidelines as well as to present empirical basis on prevention and management for cardiovascular disease by identifying high susceptible population.

For genome epidemiological study in cardiovascular disease, our research teams have developed community-based cohort with 5,600 subjects in rural areas, including Gyeonggi-do, Gyeongsangbuk-do, and Jeollabuk-do, from 2004 to 2006 year. In this study, we assess lifestyle pattern, and any risk factor of environmental exposure, which are associated with cardiovascular disease. In addition, we are going to perform the clinical examinations to detect high blood pressure, diabetes, any disorders on lipid, and atherosclerosis. Our team has established the repository of biological specimens, such as genomic DNA, serum, plasma, urine to carry out genomic, proteomic, and metabolomic analysis. Starting from 2007, the re-examination studies will be carried out to estimate the incidence rates of high blood pressure, and diabetes, any changed characteristics of blood lipid, and atherosclerosis.

This cohort study should 1) involve accurate laboratory tests to characterize various diseases of CVD in detail, 2) standardize and upgrade the quality of surveys, 3) maintain the good quality in the subsequent repeated survey, and, 4) ensure the high participation rate of target population in re-examination surveys. Moreover, the close cooperation among geneticians, molecular biologists and epidemiologists is an essential step to examine the gene-environmental interaction.