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Genome-Wide Association Studies of the Korea Association REsource (KARE) Consortium

Kyung-Won Hong[1], Hyung-Lae Kim[2]* and Bermseok Oh[1]*

[1]Department of Biomedical Engineering, School of Medicine, Kyung Hee University, Seoul 130-702, Korea, [2]Department of Biochemistry, School of Medicine, Ewha Womans University, Seoul 158-710, Korea

Abstract

During the last decade, large community cohorts have been established by the Korea National Institutes of Health (KNIH), and enormous epidemiological and clinical data have been accumulated. Using these information and samples in the cohorts, KNIH set out to do a large-scale genome-wide association study (GWAS) in 2007, and the Korea Association REsource (KARE) consortium was launched to analyze the data to identify the underlying genetic risk factors of diseases and diverse health indexes, such as blood pressure, obesity, bone density, and blood biochemical traits. The consortium consisted of 6 research divisions, formed by 25 principal investigators in 19 organizations, including 18 universities, 2 institutes, and 1 company. Each division focused on one of the following subjects: the identification of genetic factors, the statistical analysis of gene-gene interactions, the genetic epidemiology of gene-environment interactions, copy number variation, the bioinformatics related to a GWAS, and a GWAS of nutrigenomics. In this special issue, the study results of the KARE consortium are provided as 9 articles. We hope that this special issue might encourage the genomics community to share data and scientists, including clinicians, to analyze the valuable Korean data of KARE.

Keywords: genome-wide association study, KARE, genetic risk factor

Main Text

Most common diseases are influenced by multiple genetic factors as well as environmental factors. However, each single genetic effect might contribute a small part to the phenotype. Therefore, large sample sizes and genome-wide analysis are necessary to identify multiple genetic factors. Since 2001, the Korea National Institute of Health (KNIH) has established cohorts nationwide and accumulated epidemiological and clinical information on a large scale, up to 300,000 subjects. Using these samples, KNIH set to produce genotype data in 2007 and proceeded to perform a genome-wide association study (GWAS). In order to make the most of the massive phenotype and genotype data set, the extramural research consortium was organized to host scientists interested in these data. The Korea Association REsource (KARE) consortium consists of 6 research divisions, formed by 25 principal investigators in 19 organizations, including 18 universities, 2 institutes, and 1 company. Each division focused on one of following subjects: the identification of genetic factors, the statistical analysis of gene-gene interactions, the genetic epidemiology of gene-environment interactions, copy number variation, bioinformatics related to GWAS, and a GWAS of nutrigenomics.

In this issue, 9 papers were selected from the consortium. Hong *et al.* (2010) investigated the feasibility of using imputed SNPs for a GWAS with blood pressure traits to determine whether they could improve the previous results that were analyzed without imputation (Cho *et al.*, 2009). Park *et al.* (2010) performed a GWAS for blood pressure with 3 different phenotype models: young hypertensive cases versus elderly normotensive controls, the upper 25% versus the lower 25% of SBP distribution, and SBP and DBP as continuous traits. Kim *et al.* (2010c) conducted a GWAS for hematological parameters using copy number variations. Kwon and Kim (2010) analyzed a GWAS for 3 erythrocyte traits, such as hemoglobin, hematocrit, and red blood cells. Doo and Kim (2010) conducted an association study between an ADIPOQ gene SNP (rs182052) and obesity in Korean women. Kim *et al.* (2010b) compared the Affymetrix SNP array 5.0 and oligoarray platforms for defining copy number variation. Oh *et al.* (2010) applied a joint identification approach to large-scale GWA data in order to identify genetic variants of obesity for the Korean population. Kim *et al.* (2010a) applied a structural equation model to a GWAS for the following two purposes: to model a complex relationship between a genetic network and traits as risk factors and to achieve

*Corresponding author: E-mail ohbs@khu.ac.kr
Tel +82-2-961-0617, Fax +82-2-961-5515
E-mail: hyung@ewha.ac.kr
Tel +82-2-2650-5727, Fax +82-2-2652-7846
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a better understanding of the biological mechanism through not only a single gene but also many genes involved in the pathway of the trait. Kim *et al.* (2010d) conducted targeted resequencing of loci identified by the KARE GWAS to detect potential casual and/or rare variants for complex traits, such as type 2 diabetes and triglycerides.

During the last 2 years of the consortium, there have been more than 15 meetings and gatherings to present and discuss results of the KARE data analysis. Up to 100 participants were involved in the discussions, establishing a new paradigm of science in genomics in Korea. The events would not have been possible had KNIH decided not to make the valuable data open to the consortium. As members of the consortium, we appreciate the members of the Center for Genome Science in KNIH, the Ansung and Ansan epidemiological cohorts, and DNALink for producing and managing such enormous and valuable resources. Lastly, we hope to apply these investigations to better serve the health of the participants in the cohorts.

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