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## Preface

## BioBank Japan project: Epidemiological study



## Preface

Biobank Japan project was started as a leading project of Ministry of Education, Culture, Sports, Science and Technology (MEXT), Japan with the aim of providing evidence for the implementation of personalized medicine. In the first five years from fiscal 2003 to 2007, this project constructed a large, patient-based biobank, referred to as the Biobank Japan, consisted of 200,000 patients as the basic infrastructure of genetic research for common diseases. This activity was accomplished through the collection of clinical information and biological samples related to 47 target diseases and with the cooperation of 12 Japanese medical institutes. In the second five year period from fiscal 2008 to 2012, this project conducted genome-wide association studies in various diseases and identified many genetic variations associated with disease susceptibility and drug responses. As of the end of 2015, this project has published 296 papers in representative academic journals including 9 papers in *Nature* and 49 papers in *Nature Genetics*. All the publication in this project is opened to the public through the project website (<https://biobankjp.org/work/public.html>). In the third period from fiscal 2013 to 2017, this project is expanding the Biobank Japan by enrolling new participants with 38 target diseases, starting sequence-based genomic analyses, and constructing a tissue bank. In addition, this project started the collaboration with clinical research groups in Japan from 2014. Currently, this project stores DNA, plasma, and frozen cancer tissues collected through the clinical trials performed in Japan Clinical Oncology Group (JCOG), Japan Children's Cancer Group (JCCG) and National Hospital Organization (NHO).

Precision medicine is a newly emerged concept in the State of the Union speech of US President Barack Obama in 2015. The concept of precision medicine is the same as that of personalized medicine. According to the definition of US National Institutes of Health, precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. This means that the main objectives of precision/personalized medicine are the therapeutic optimization and the decrease in the incidence of common (multifactorial) diseases that various genetic, environmental, and lifestyle factors have influenced to their occurrence, progression and prognosis in a complexed manner. Therefore, to clarify the interactions between genetic, environmental, and lifestyle factors have an important role for the implementation of personalized/precision medicine. Rapid advancement of human

genetic research have identified many susceptibility genes or loci associated with common diseases, however, there is scarce information about the clinical utility of identified genetic factors on the treatment and prevention of common diseases. Moreover, the information concerning environmental or lifestyle factors that modify the genetic risk of common diseases is limited.

Although Biobank Japan project have developed genetic information of genome-wide genotyping data in 200,000 patients, there have been no report on the clinical information corresponding to these patients because this project was initially designed by focusing on the use of genetic research only. In 2009, Biobank Japan project launched a Clinical Data Analysis Working Group organized by Guest Editors and started the survival survey of approximately 150,000 patients from 2011. This survival survey was successfully accomplished with the strong cooperation of 12 medical institutions and MEXT, and resulted in the high follow-up rate (97.0%) with the mean follow-up period of 7.7 years. Moreover, Biobank Japan project collected various clinical information from medical records in 200,000 patients annually from the registration to fiscal 2012. Hence, Clinical Data Analysis Working Group started data cleansing of this huge clinical data from 2013 and constructed a large clinical database consisting of up to 10 year consecutive clinical information including disease status, laboratory examination, and drug information etc. Using this large clinical database, Clinical Data Analysis Working Group reported epidemiological aspects of Biobank Japan samples in this supplement. I hope the manuscripts published in this supplement will guide to the further understanding of Biobank Japan samples and promote further analysis toward the implementation of precision/personalized medicine.

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Michiaki Kubo, MD, PhD, On behalf of Guest Editors  
RIKEN Center for Integrative Medical Sciences, Yokohama City,  
Kanagawa, 230-0045, Japan

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