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Ã¢â?¬Å?The interest of companies in biobanks has significantly increased but investments have not yet been realisedÃ¢â?¬Â•

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BioBank Japan, a hospital-based multi-institutional biobank that has collected DNA, serum and medical record information from ~260,000 patients affected by one of 51 pre-defined diseases, is now launching a new phase to accelerate precision medicine and health in Japan. In an address on May 24 in Tokyo, Professor Yoichiro Kamatani, a member of the Steering Committee at BioBank Japan, presented the new phase and future direction of this important biobank for the future health of Japan. In an interaction with BioSpectrum Asia, Prof. Kamatani reveals more details about the work being done at BioBank Japan.



Since its inception in 2003, BioBank Japan has achieved many milestones. Could you please elaborate on those?

I would like to introduce a work of our colleagues published in 2002 which was the preliminary but the first successful report of genome-wide association studies (GWAS) in the world. This group founded BioBank Japan (BBJ) and formed the initial core group. Biobank Japan soon reported novel genetic factors including KCNQ1 for type 2 diabetes gene or CD244 for rheumatoid arthritis. Detecting genes that larger groups in the US and Europe had not reported at that time demonstrated early on that analysing Japanese or Asian populations was an effective gene detection strategy due to differences in allele frequencies and genetic structures of diseases among populations. These efforts were followed by our participation in the Asian Genetic Epidemiology Network (AGEN) where the integrative analysis of genomic data in East Asia was performed, and have achieved some initial results in our collaborations in Asia including the identification of 17 newly associated variants for kidney function.

Around the time when international genome-wide meta-analyses began to be conducted by various disease groups, BBJ often represented one of a few non-European groups in many studies. Recent years have shown that integrated analysis of samples with genetic diversity enhances the detection of genetic susceptibility variants more than the analysis of a single population. BBJ significantly contributed to recognising this at an early stage.

BBJ successfully completed genotyping of 180,000 individuals in 2015. From this data, we were able to conduct GWAS on a biobank scale. As a result, we identified 200 loci associated with Body Mass Index, 1407 loci associated with clinical test values, 88 loci related to type 2 diabetes, 40 loci associated with myocardial infarction, and 150 loci associated with atrial fibrillation. Further, our continuing collaborative effort further contributed to the identification of 287 loci for schizophrenia, 12,111 loci for human height, and 89 loci for stroke by international consortia.

Moreover, BBJ demonstrated, through mutual comparison with the UK Biobank, that polygenic risk score (PRS), which is a risk score calculated from genetic data weighted by GWAS results, has issues with transferability between races. All of these instances have served to validate the efficacy of genome analysis in non-European populations, which we believe backs up the fact that biobanks outside of the US and Europe are now being launched one after another and succeeding in securing funding.

Most recently, our research has found that in situations where there are concurrent rare genetic variants and helicobacter pylori infections, which are particularly common in Asians, there is a significantly strong increased risk of developing gastric cancer. This indicates that Asians who carry these rare variants should actively seek eradication of helicobacter pylori.

What are the key objectives behind launching the new genomics-multi omics project at BioBank Japan? What are the phase-wide plans?

BBJ has been one of the few large-scale biobanks outside of US and Europe, and through its GWAS, we have identified a large number of disease susceptibility genetic factors that are not detected in those countries. These efforts of BBJ, essential for achieving racial equality in genomic medicine, have been applauded on various occasions. On the other hand, biobanks are now being asked to contribute to the construction of precision medicine not only through genomic data but also by harnessing environmental factors that change throughout life and multi-omics data such as proteomics and metabolomics. BBJ has already collected a large-scale and longitudinal series of blood samples to achieve this. Our goal is to conduct research that forms the foundation for achieving race-equitable precision medicine through advancing this analysis. Firstly, we are aiming to prepare the blood omics data of all participants in BBJ and will continue to work towards preparing longitudinal data.

For the same purpose, we aim to collect real-time medical records of BBJ participants in their respective hospitals. Additionally, we are advancing our efforts in information collected through wearable devices.

Will both the private and public sectors participate in this project, with respect to the industry and academia in Japan?

Currently, BBJ is operated entirely with public funds. We have started discussions aimed at obtaining funding from companies such as pharmaceutical firms, but we are still on the way through the process. We are in a position to accept funding.

Will you be focusing on other Asian countries as well in this project?

While the participants in BBJ are limited to Japanese individuals, from the outset, it has been supporting genome projects in Malaysia and Thailand. Even now, we are in contact with the Korea Biobank, Taiwan Biobank, and China Kadoorie Biobank.

Furthermore, the genome analysis results from BBJ are made public and are used in the analysis of these other biobanks.

What challenges might you encounter while executing this project?

Japan, with its long history as a single nation, has a fundamental national character that places importance on lineage. Genome data includes sensitive information about this, so it was not initially easy to proceed with the construction of a large-scale genome biobank in a way that would satisfy the government and the people. Despite imposing numerous constraints on ourselves, BBJ made extraordinary efforts for the advancement of global genome research, and as a result, BBJ has been able to construct one of the world's leading non-European biobanks, which is a source of pride for us. Each country in Asia is expected to face challenges in advancing population genomics research due to its unique cultural backgrounds. However, the fact that several countries have overcome these challenges and achieved significant results should serve as a reference for others.

On the other hand, there are still high barriers to continuing expanding the scale of the biobank and advancing the social application of genome research in Japan due to the same reason, and we are still working hard to overcome them.

What are the current challenges facing the healthcare sector in Japan? How are the government, industry and academia addressing them?

Previously, health-related authorities were not enthusiastic about the application of genome data to health and medical care, but this has now been resolved, and national projects for genome medicine for cancer and rare diseases are progressing. On the other hand, the focus of BBJ is on complex diseases. The application of these diseases to health and medical care has not yet reached a consensus, and we continue to advance this from both the presentation of research results and explanations to the authorities. This effort is not limited to BBJ but is also jointly pursued by other domestic biobanks in Japan, such as the Tohoku Medical Megabank and the National Center Biobank Network.

While the interest of pharmaceutical companies and laboratory testing companies in biobanks has significantly increased compared to before, the large-scale investments made by global mega-pharmaceutical companies have not yet been realised. However, interest is consistently shown, and we continue to work towards facilitating the development of biobanks with the participation of corporations.

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