



Special Issue: Genomic Era

## Brief Review

# Genomic Era

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The last decade can be called the genomic era. In 2003, it was announced that the whole genome sequence had been determined, and the subsequent HapMap project and the 1000 Genome project detected many genetic polymorphisms. Technological progress enabled us to detect a large number of genetic polymorphisms in a very short time, and the genome-wide association study (GWAS) method was established. Nowadays, one million single-nucleotide polymorphisms (SNPs) can be typed on one chip and hundreds of samples can be typed in only a few days. Moreover, new-generation sequencers have recently been developed and soon the whole genome sequence of a person will be read in a day for \$1,000. Such dramatic advances of polymorphism typing and DNA sequencing technology revealed the pathogenesis of many diseases from a genetic point of view.

Many GWAS against a variety of common diseases have been performed, and tens of susceptibility genes for each disease have been detected. As representatives of such studies for inflammatory diseases, rheumatoid arthritis (RA) and primary biliary cirrhosis (PBC) are dealt with in this special issue. Dr. Chikashi Terao at Kyoto University is a

core member of the GARNET consortium, which consists of three large Japanese RA genetic research laboratories (RIKEN, Tokyo Women's Medical University, and Kyoto University) and participated in a worldwide GWAS meta-analysis. He will overview the GWAS against RA. Dr. Minoru Nakamura at Nagasaki University is a leader of PBC research in Japan and conducted a GWAS against PBC. He will discuss the pathogenesis of PBC from the perspective of the genome.

The genomic sequence has long been believed not to change, except in exceptional types of cell such as T and B lymphocytes. However, sequence technology revealed that genomic aberration is commonly seen when a cell divides. The DNA sequence of iPS cells tends to be considered to be completely the same as that of the founder cells, but this is not always the case. Dr. Akira Watanabe at the Center for iPS Cell Research and Application (CiRA) of Kyoto University, who is a leader in the Sequencing Core Facility of Dr. Shinya Yamanaka's Laboratory, has tackled this issue for the standpoint of iPS cell quality control, and will describe genome integrity in detail. In the future, genomic aberration in somatic cells might become evident as



a cause of disease development.

Another important viewpoint of the genetic effect on disease development is epigenetic regulation of genes. Genetic polymorphisms of regulatory elements may contribute to the level of gene expression, but the effects of the genetic products are regulated strictly by histone modification. Dr. Takashi Minami at the University of Tokyo has studied epigenetic histone modification in endothelium

genome-widely and will investigate pathogenic mechanisms of vascular diseases.

All the authors in this special issue are experts in the field of genomic and epigenomic research and I am sure that these articles will help the readers understand the genomic/epigenomic aspects of inflammation and regeneration.