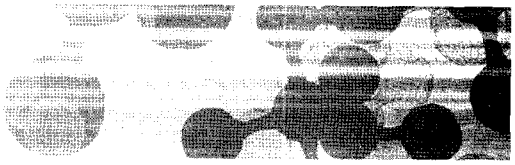

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Clinical implication of structural variation in human genome

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Genomic variability can be present in many forms, including single nucleotide polymorphisms (SNPs), variable number of tandem repeats such as minisatellite and microsatellites, presence/absence of transposable elements, and structural alterations of chromosome. So far, SNPs were thought to be the predominant form of genomic variation and to account for much normal phenotypic variation. Therefore, thousands of research data regarding the association between SNP genotypes and disease susceptibility have been published annually. However, recently new concept of genomic variation has been suggested named copy number variation (CNV).

Why is it important to study CNV? Like SNP, CNV may account for some of the differences in individuals' susceptibility to disease or explain why people has different reaction to specific drug or environmental stimuli. Quantitative trait of human disease may be associated with copy number differences of certain genomic loci. It has been suggested that many of the identified sites of structural variation encompass genes that are not essential for viability by could be described as 'environmental sensor genes' associated with drug detoxification, immune response, etc. SNPs alone may not predict all phenotypes, and a better understanding and further research into this newly appreciated type of genetic variability is required to properly address inter-individual variations including drug efficacy and toxicity. With the advent of higher resolution genome-wide assays such as microarray-based CGH (array-CGH), CNV associated disease interpretation has rapidly developed. Using array-CGH based copy number analysis data, we can get more idea about cancers, idiopathic genetic diseases, psychiatric diseases, and metabolic disorders.

Taken together, it is expected that CNV analysis, accompanying with SNP analysis, will be critically important to understand the nature of many complex diseases and also important to prepare new genome medicine in a near future.