S13-5 Current status of research in clinical genomics, and guidelines for PGx

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GWAS (genome-wide association study) was first introduced by RIKEN and now widely used for the identification of genetic causes of many diseases in the world. We have performed GWAS using about 500,000 SNP genotypes from each of about 147,000 subjects with 10 different diseases to identify associations between genomes and laboratory data. As a result, 60 associations for 8 hematological traits and 29 associations for 12 biochemical traits were identified ($P < 5 \times 10^{-8}$). Of them, 33 hematological associations and 13 biochemical associations were novel. In the future research, we should perform studies in which both qualitative and quantitative traits are included. When individual germline information is included in clinical laboratory tests, special attentions should be paid from the ethics point of view. However, PGx has a quite different characteristic from other germline tests such as those for monogenic diseases. Therefore, the ethical problem is much smaller for PGx tests than other germline tests. Based on such consideration, 3 Japanese medical associations collaborated to release “Guideline for PGx tests in clinical practice” in March 2009. Five Japanese medical and pharmacological associations are now collaborating to establish a guideline in which PGx for clinical research, drug development and non-medical areas is included.