Workshop Executive Summary

The Office of Research Infrastructure Programs in the Division of Program Coordination, Planning, and Strategic Initiatives convened on September 10-11, 2015 a Symposium titled "Linking Disease Model Phenotypes to Human Conditions" at the Fishers Lane Conference Center, Rockville, MD. Over 120 participants, including 58 NIH representative from 16 Institutes and Centers, attended the meeting. Among participants were foreign scientists, representatives from other government agencies, such as FDA and US Army, and the biotech and pharma industry. The purpose of the Symposium was to discuss the current status of phenomics and its role in closing the gap that exists between biomedical research and clinical medical practice. Twenty-five speakers from the US, Germany and Great Britain presented their research and provided advice to the NIH and other participants. The meeting started with greetings from the NIH officials and a key note presentation by Peter Robinson (Max Planck Institute for Molecular Genetics, Germany) entitled "Deep Phenotyping for Translational Research and Precision Medicine". Day 1 of the symposium covered the following sessions: "The Current Status of the Human Clinical Phenotype Ontology and Terminology, and Associated Data Annotation and Use", "Cross-Species Phenotype Analysis and Ontology", "Large Scale High Throughput Analysis of Disease Model Phenotyping Data and Annotation of Gene Function" and "Linking Disease-Relevant Phenotypes with Physiologically Relevant Molecular Pathways and Networks". Each of the sessions was followed by a round table discussion. The speakers and participants agreed that lack of alignment of phenotypes between model species and humans has been a historic impediment to understanding disease processes. Future progress depends upon integration of clinical, biological, and genomic data, as well as development of tools for identification and analysis of specific and amendable disease-causing molecular phenotypes of various diseases. Day 2 of the symposium consisted of the following sessions: "Clinical and Experimental Biology Data Integration Emerging Field of Precision Medicine" and "Informatics Tools for Phenotypic Analysis and Data Sharing". After several round table discussions, the meeting closed with the listing of a set of recommendations. From the broad active discussions during the symposium, it became evident that with the high-throughput DNA sequencing methods made available in recent years, there is a wealth of information from human genome studies of patient cohorts with human diseases/conditions with the need of identifying the relevant effector genes. The discussion at the symposium brought to light that integration of this data with detailed disease descriptions, phenotype information from appropriate animal models, and identification of environmental conditions can provide much better candidates for disease gene effectors for use in precision medicine. Identified bottlenecks for this process include the lack of a community-wide, standardized, and machine-readable language for describing phenotypes and their genomic and environmental contexts, as well as algorithms for integration among key areas of biology and medicine.