Summary of NHGRI's Strategic Planning Session
World Congress of Psychiatric Genetics (WCPG) Meeting
Anaheim Marriott
October 26, 2019

The National Human Genome Research Institute (NHGRI) and the National Institute of Mental Health (NIMH) co-hosted a strategic planning event at the end of Education day during the 2019 WCPG annual meeting. Education day consisted of presentations on the use of technologies such as machine learning, epigenomics, transcriptomics, nuclear architecture and single cell approaches in psychiatric genetics, followed by strategic planning proposals for the Psychiatric Genetics Consortium (PGC). NHGRI introduced their strategic planning goals and timeline as well as a comparison of NHGRI, National Institutes of Health (NIH) and genomics research. NIMH provided an overview of key elements of their strategic thinking, which included genes to function, platforms for gene discovery, the need for participants with diverse ancestry, and the approach of bridging levels of analysis to understand complexity. NHGRI summarized their mission as well as the scope and philosophy of the current NHGRI award portfolio, including investigator-initiated research. The five focus areas for NHGRI strategic planning were presented, as well as challenges in Basic Genomics and Technology and the Genomics of Disease. NHGRI solicited feedback from the audience.

A number of high-level ideas were raised. Better support and utilization of crosscutting, generalizable studies were requested. For example, metabolism and microbiome affect many phenotypes across domains, yet studies and results are often siloed by disease. Broader phenotyping and sharing would increase the value of these studies to the community. Better integration of clinical biomarker studies is needed; today they are often siloed by technology (genomic v. imaging) and by disease, limiting their scientific impact. Including ancestrally diverse participants should be the norm, rather than special diversity-focused studies. Better guidance on finding the appropriate institute for applications is needed; the available guidance can be difficult to find and are inconsistent across sources. Better communication between NIH program and review on research priorities would help applicants. Sometimes institutes communicate priority ideas to applicants, yet reviewers assign poor scores because these areas are not deemed to be important by the reviewers.

A number of technology needs were raised. While it was noted that there are technology development meetings, there is also a need for a venue for communication between technology users and technology developers. Standard biosamples for studying cell-cell variation and person-person variation are needed; this could be met with a repository for standard cells/tissues for distribution, with common data available, that could be utilized across diseases/technologies. Higher-throughput scRNA-seq is needed to support studies such as the perturbation of all genes and perturbation of combinations of genes, given that this is a common readout for those types of studies. Better multi-omic, integrative technologies and approaches are needed, especially for single cells. Better methods and resources for networks/pathways analysis are required. While the existing resources are simplistic, this has been an important approach in advancing psychiatric genetics.

Disease needs were also raised. Better biomarkers of clinical trajectories are needed to track progression, treatment and outcomes. Studies on crosscutting elements affecting many diseases, such as metabolism and the microbiome, are needed. Disease studies with broad phenotyping using standardized terms are needed to connect phenotypes across diseases and domains. Studies of gene expression and eQTL across all age groups would be foundational; psychiatric studies have revealed the importance of developmental stage-specific gene expression. Approaches are needed to integrate epigenetic data across systems, such as large cohorts, tissues/organs, cell-based systems, and iPSC from patients; current studies are typically not performed across systems, and inferences across systems can be criticized as unjustified. Finally, disease studies must take into account that disease presentation is not always uniform across populations. Thus, techniques are needed to identify phenotypes across populations. Finally, a need for a program to educate clinicians in the principles of genetic medicine was recognized.