

USP Precision Medicine & Genomics Virtual Roundtable April 23rd, 2020

Executive Summary

Unlike the traditional one-size-fits-all, population-based approach to disease treatment and prevention, precision medicine (PM) takes into account individual differences in patients' genes, environments, and lifestyles when developing strategies for the prevention and treatment of disease. Until recently, the promise of PM was oriented toward the future, but the emergence and widespread distribution of multiple new technologies (e.g., next generation sequencing, machine learning) and new biomarkers (e.g., DNA, RNA, protein, and extracellular biomarkers) from easily accessible mediums (e.g., plasma, urine, and saliva) have enabled the development of PM into a valued tool in the practice of medicine. However, challenges to the growth of PM remain before the full potential can be reached. To address these challenges, USP Global-Biologics brought together experts in PM for a (virtual) roundtable discussion on April 23rd, 2020.

Challenges

Challenges that need to be overcome to enable acceptance of the clinical utility and widespread use of PM include education and standardization. Physicians are presented with a variety of diagnostic options and may wonder how to use them and how to avoid mistakes in the interpretation of results. Much of the confusion can be traced to a lack of experience in identifying quality data that is clinically actionable. Heterogeneity in the presentation of PM results compounds these challenges. The data may exist as a mixture of diagnosis codes, clinical notes, images, and molecular data collected from different sources, making interpretation difficult. There are few shared standards for preparing, analyzing, or sharing data. Additionally, there is significant bias inherent in the data itself due to a lack of inclusion of population diversity in test development.

Solutions

The panelists agreed that there are many challenges facing PM and discussed strategies for addressing these issues. For example, education programs designed to inform primary care physicians who work in community settings could augment their knowledge of how to manage their patients' PM data. Also, presenting the data in ways more amenable to utilization by clinicians would contribute to adoption and contribution to patient care. Additionally, an explicit focus on linking analytical results and clinical utility would spur PM adoption. This can be accomplished by bringing together data scientists, life scientists, and clinicians to collectively identify the most relevant issues in a clinical setting such that information gained from novel PM technologies can be optimally adapted for clinical use. Finally, a standardized and transparent informed consent process presented as a narrative and in multiple languages may increase participation by underrepresented minorities, thereby diversifying the sampled population and improving care for individual patients.

Conclusions and Next Steps

The goal of the roundtable was for subject matter experts to discuss the state of the art of precision medicine and current challenges to translating genomic data into clinical decisions. The participants proposed some potential strategies to enhance the consistency and interpretability of genomics data to ultimately support more widespread adoption of genomics-based medicine. A rich exchange of ideas was obtained, which will form the basis for continuing discussions with stakeholders, and to explore how USP can facilitate the robust



adoption of personalized medicine through the development of best practices, standards, and educational programs.

For more information about the Virtual Roundtable on Precision Medicine & Genomics, please contact Sarita Acharya, Science & Standards Liaison, Science-Global Biologics, Rockville, MD at sarita.acharya@usp.org.