Preface

Precision Medicine Exits the Hype Cycle and Enters into Productive Clinical Use

Ryan J. Schmidt, MD, PhD
Editor

The notion of precision medicine broadly refers to the administration of a patient-specific treatment based on unique features of an individual or their disease that are identified in the laboratory. This concept is not new to medicine and has existed for many years to allow for the administration of compatible blood products and antibiotics selected based on antibiotic resistance testing. The modern incarnation of precision medicine leverages our ability to determine the molecular cause of genetic disorders and apply rational therapies based on specific disease-causing genetic variants.

Technological advances in molecular genetics have rapidly increased our understanding of constitutional genetic disorders. Broad-based clinical molecular diagnostic testing has now become standard in many disease areas. The results of this testing now have the potential to extend beyond providing a diagnosis and serve as a basis for precision therapies that target specific genetic alterations. In addition, clinical molecular diagnostic testing can serve as a starting point for additional research that increases our understanding of genetic disease. Collectively, these components comprise the foundation for a comprehensive precision medicine program. The combination of diagnosis, treatment, and research capabilities has the potential to interact in a synergistic manner when thoughtfully implemented together at a single institution.

As the precision medicine enterprise matures, it faces a variety of barriers that currently limit its widespread use. Now that the technical challenges that surround developing and implementing a precision therapy are being overcome, the financial and regulatory components of this model of care must also be built.

Precision medicine has begun to exit the hype cycle and enter into productive clinical use. As we celebrate the treatment of the first patients with precision therapies, we must consider what it will take to diagnose and treat the last patients with the most

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technically challenging genetic variants. In addition, we must address the needs to effectively scale up the precision medicine enterprise and overcome the practical barriers that will inhibit the widespread application of these complementary diagnostic and therapeutic approaches.

Ryan J. Schmidt, MD, PhD
Clinical Genomics Laboratory
Center for Personalized Medicine
Department of Pathology and
Laboratory Medicine
Children's Hospital Los Angeles
Keck School of Medicine of USC
2100 West 3rd Street, Suite 300
Los Angeles, CA 90057, USA

E-mail address:
rschmidt@chla.usc.edu