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EDITORIAL

The road ahead: less travelled and more arduous than initially envisioned

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While some have argued that physicians have always provided 'personalized care' to individual patients, the capacity to transition from genomic discoveries to clinical decisions has raised the spectre of a potential shift in how we assess risk and treat disease. Although some currently use the terms 'personalized medicine' and 'molecular guided therapy' synonymously, it is worth noting that what has really changed are the methods that have been proposed to 'personalize' or 'individualize' care. Overall, the change appears to be more quantitative than qualitative, partly because we can envision a day in which full genomes are sequenced and crossed against comprehensively annotated databases. However, we are still far from that point and need to be sanguine in defining realistic short-term goals while accounting for the pleotropic effects of the transitions ahead. The distant promises have to be tempered by the realities of change, change that comes slower and less predictably than anticipated.

The near future will certainly be filled with exciting new discoveries and developments using the tools of genomics. The challenge will be translating these into effective strategies that can be useful in the clinic and the public health forum. Particularly daunting will be the education of health-care professions and the public at large. The concept of risk and how to convey it is already shifting before our eyes as we catalog more and more genetic variation with less than full penetrance. We have not yet explored how to effectively communicate genetic risk for complex diseases nor have we settled on how to provide recommendations based on information that continues to evolve.

The accelerated use of genomic technologies has increased our collective ability to resolve variability in populations and begin to map variation against disease outcomes. It has unearthed a paradox that we will face for generations to come: how to apply the knowledge we learn from grouping individuals to decide what to do for an individual. This has posed a substantive challenge, both in the immediate setting and the futuristic visions of genomic medicine. In particular, how we begin to introduce the emerging knowledge of genomic discovery into practical and durable paradigms for clinical care and public health decisions must artfully address the problems of entrenched systems and approaches to health-care. While we can imagine the development of novel healthcare strategies, the impact of these challenges crosses a diverse spectrum of stakeholders, from consumers, to government, regulatory authorities, healthcare providers, insurers, health professionals, the technology and pharmaceutical industry, to name a few.

Our ability to rapidly acquire information about individual people and their disease has already created a major disconnect between the science of discovery and the uncertainty of exactly how the information will be translated into new paradigms. Moreover, it is difficult to predict how the proposed changes will have a significant impact on ethical and societal issues. These latter issues must be addressed in parallel to realize the full benefit of what we

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will harness from genomic studies. If we race too far ahead in discovery and fail to adequately consider the implementation steps that follow a suitable validation process, the gap between the practical utility of new approaches and the promise of new discoveries widens. It is clear that new technologies will pump out genomic information faster, cheaper and with less error, but exactly how these will impact our lives, both in the clinic and beyond will remain challenging. While some will argue that the intangible cost of delay will always be unknown, it is the price we often have to bear in science, and defining the optimal path forward as the complexity increases is paramount.

This special edition of *Human Genetics* addresses some of the current challenges in implementing genomic medicine and the promise of personalized healthcare. It is not authoritative nor comprehensive, but instead it is focused on nexus points, which represent some of the tough decisions and systemic changes ahead. As long as we proceed with a perspective that keeps watch over the larger issues as well as the specifics of any given genomic data set, we will be in a better position to embrace the changes and fully take advantage of technological advances in genomics for our collective and individual benefit.