Introducing “Genomics and Precision Health”

W. Gregory Feero, MD, PhD

The US health care system has seen significant changes over the last decade resulting from diverse factors including the widespread adoption of electronic health records, the opioid crisis, and the roll-out of the Affordable Care Act. Such changes have buffeted the day-to-day work of physicians and other health care practitioners, leaving many finding it challenging to address demands placed on them. Looking forward in 2017, it seems that the future will hold more turbulence. In this milieu, it is easily possible to overlook that medicine has been undergoing a much more gradual and deeper transformation. This shift is inexorably moving medicine from an endeavor in which care for individual patients is driven by trial and error informed by studies designed to measure population outcomes to one in which care is selected based on a deep understanding of health and disease attributes unique to each individual. Accelerated by the completion of the Human Genome Project, this transformation has been variably called genomic medicine, genomic health care, personalized medicine, and precision medicine. The extent to which it ultimately alters medicine remains unclear.

The exact definition of precision medicine is a moving target; however, the National Institutes of Health currently states that precision medicine “is an emerging approach for disease treatment and prevention that takes into account individual variability in environment, lifestyle and genes for each person.” The National Academies of Science, Engineering, and Medicine recently renamed the “Roundtable on Translating Genomic-Based Research for Health” to “Roundtable on Genomics and Precision Health.” Including both genomics and precision health in the title conveys that while genomics and precision health substantially overlap, they are not synonymous. Genomic science and technology encompasses domains well away from health care including agriculture, zoology, and evolutionary biology. Similarly, aspects of precision health, such as the use of data on behavior from wearable sensors to guide care, may not rely on any genomic data. Likewise, health instead of medicine acknowledges that the former term is more encompassing than the traditional medical model of disease-based care. In the not-too-distant future, the coupling of sound science with in-depth understanding of an individual’s characteristics, environmental exposures, and behaviors to achieve good health will probably simply be called quality care.

Public and private organizations across the globe are investing heavily in developing precision health. At the US federal level, there are several initiatives including All of Us (formerly known as the Precision Medicine Initiative or PMI), the Cancer Moonshot, and the Million Veterans Program. The enactment of the 21st Century Cures Act in December of 2016 further establishes federal commitment to precision health programs, ensuring billions in funding. Major private health care systems, including Kaiser Permanente in California and Geisinger in Pennsylvania and New Jersey, have large ongoing initiatives to collect, study, and make use of genomic and other types of data from populations in their system catchment areas. Internationally, the UK’s National Health Service has embarked on the 100 000 Genomes Project, a bold multiyear initiative to transform UK biomedical research and clinical care to a precision health-oriented model. Similarly, China, Iceland, and Japan have major, long-term precision health initiatives. The cumulative effect these activities will have on biomedicine over the coming decades will be substantial.

For most clinicians, the tangible benefits of the global push into precision health have been slowly accumulating. Probably the most high-profile effects have been in cancer care, for which some aspect of molecular genetic or biochemical testing often accompanies the diagnostic evaluation of tumors, selection of treatments, and provision of prognosis. Similarly, diagnosis of monogenic and syndromic conditions has been transformed by technologies that allow rapid and precise interrogation of biochemical and genetic abnormalities in patients. Further, therapies are now available that can target the deleterious effects of specific variants for a limited number of conditions such as cystic fibrosis. In reproductive health, advances in variant detection technology have facilitated expanded carrier screening, cell-free DNA aneuploidy screening, and preimplantation genetic diagnosis. In some medical specialties, incorporation of genomic and other forms of precision health data into care is routine.

To leverage advances in precision health, nongeneticist physicians must gain an adequate understanding of emerging technologies. This is no easy task because the technologies can be complex, are in continual evolution, and, given their newness, few evidence-based guidelines exist to guide prudent clinical application. Additionally, for many clinicians, their last in-depth exposure to genetics was in medical school which, assuming subsequent completion of a residency, is a relative eternity in genomic science. This substantial knowledge asymmetry between purveyors of precision
health technologies and non-geneticist clinicians has created the potential for well-intentioned misapplication of tests to guide prevention, diagnosis, and treatments, with unknown downstream consequences. In the United States, efforts to help clinicians with this educational gap date back decades—from projects such as the Genetics in Primary Care Initiative and the work of the National Coalition for Health Professional Education in Genetics to the ongoing work of the National Human Genome Research Institute’s Inter-Society Coordinating Committee for Practitioner Education in Genomics.11-13

The article by Evans and colleagues14 in this issue of JAMA marks the launch of a new series, JAMA Insights: Genomics and Precision Health, consisting of brief educational articles intended to help non-geneticist clinicians overcome knowledge barriers. The articles will cover a wide range of topics and will include cutting-edge precision health technologies as well as those that are in common use by some medical specialists. Each article will be structured to describe the intended application of the technology, how the technology works, important considerations regarding clinical use, costs, and the current state of evidence supporting use of the technology for the stated application. The articles will be accompanied by online figures, supplemental content, and podcasts by the authors. In the first article, Evans and colleagues address an application using genome sequencing in healthy individuals to guide subsequent care. This promising precision health approach raises a number of challenging issues including the quality of currently available testing processes, appropriate interpretation of results, how information about the results should be used, and the resulting value of the application vs other preventive interventions. Despite a lack of clarity surrounding these issues, the use of genome sequencing in healthy individuals is occurring in the United States in the context of clinical and research programs.

The eventual success of precision health depends on well-informed decisions made by patients and the health care professionals who provide their treatment. The editors invite feedback regarding the JAMA Insights: Genomics and Precision Health series and hope that the JAMA readership finds these articles accessible and enjoyable and, most importantly, that reading them contributes to better health for patients.

ARTICLE INFORMATION

Author Affiliations: Maine-Dartmouth Family Medicine Residency, Augusta, Maine; Associate Editor, JAMA.

Corresponding Author: W. Gregory Feero, MD, PhD, Maine-Dartmouth Family Medicine Residency, 15 E Chestnut St, Augusta, ME 04330 (wfeero@mainegeneral.org).

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REFERENCES


