

Editorials: *Controversies in Family Medicine*

Is “Precision Medicine” Ready to Use in Primary Care Practice?

Yes: It Offers Patients More Individualized Ways of Managing Their Health

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Medicine has entered a transformational era in which prevention, screening, and therapy are increasingly based on a precise understanding of attributes of health and disease in the individual patient. Ignoring this puts patients at risk from avoidable, ineffective, or hazardous treatments and from over- or underscreening for chronic conditions. Additionally, failure to advocate for access to “precision medicine” may result in increased health disparities in areas such as prenatal genetic screening. Several misconceptions regarding precision medicine perpetuate debate about its readiness for use in primary care.

First, precision (sometimes referred to as personalized or genomic) medicine may be falsely equated with highly predictive genetic testing to assess risk of common conditions, such as diabetes mellitus, cardiovascular disease, and dementia, which currently does not exist. However, precision medicine has evolved to encompass more than using genetic testing to tailor care and ideally would integrate multiple streams of information, including behaviors, physical characteristics, and multiple genetic and nongenetic biomarkers, as well as patient preferences, to optimize care. The National Institutes of Health defines precision medicine as “an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person”.¹

Clinicians now have the ability to offer patients care that is tailored to their unique biology in ways that were not possible a few decades ago. Consider, for example,



This is one in a series of pro/con editorials discussing controversial issues in family medicine.

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how well-woman care has changed. Decisions regarding expanded carrier screening, which now can include genetic testing for risk of more than 100 conditions²; lipid management, which is now based on the use of formal individualized risk models³; cervical cancer screening, which is predicated in part on genotypic testing for human papillomavirus⁴; and breast cancer screening, which is based on family history risk assessment, genetic test results, and shared decision making,⁵ are individualized to each person’s family history, genetic test results, behavior, and personal choice and attributes. Care plans tailored to individual patients can then be determined. A generation ago, the annual Papanicolaou smear was a rigidly adhered to component of health maintenance for women. Currently, a more precise molecular understanding of cervical cancer biology has led to better risk stratification for individual persons. As a result, younger women and those lacking the genetic markers for human papillomavirus types 16 and 18 are now spared from unnecessary invasive screening.

Second, primary care physicians may view precision medicine as conflicting with population-centered care and evidence-based medicine. In fact, these areas are synergistic. Ideally, precision medicine should be predicated on appropriate adoption of interventions based on evidence of improved outcomes for individual persons, at reasonable incremental costs to society.⁶ Evidence

supporting such interventions may be generated in large population studies or rigorously conducted trials that focus on specific participants. Sometimes the best interventions may be population based, such as improving exercise through enhancing the “built environment” in cities. Other times, it could be the use of an expensive, yet cost-effective, targeted chemotherapeutic agent selected on the basis of variations found in a patient’s tumor sequence.⁷ Improved health of a population is, after all, a summation of the health of individual persons.

Finally, primary care physicians may feel that precision medicine is too complex, expensive, and inaccessible to disadvantaged populations that they often serve. However, precision medicine approaches are increasingly becoming incorporated into evidence-based guidelines that reduce some of the complexity and uncertainty, as well as insurance coverage issues for patients. For example, reflexive testing for incidental colon and endometrial cancer for evidence of hereditary nonpolyposis colorectal cancer (Lynch syndrome) is now routinely performed by pathology laboratories, reducing the need for physicians to memorize and apply complex clinical diagnostic guidelines.^{8,9} Similarly, human immunodeficiency virus and hepatitis C genotyping to tailor care is now an accepted part of treatment protocols, and is widely covered by public and private insurers. At this time, the cost of the most comprehensive human genetic testing—sequencing the entire protein coding regions of the human genome (including interpretation)—is roughly equivalent to three contrast-enhanced abdominal computed tomography scans and is covered by insurers in certain diagnostic situations, such as unexplained developmental delay.¹⁰ The utility of having this information for broader segments of the population is unclear, but this may become more evident in the future.

The question that primary care physicians should be considering is not whether precision medicine is ready for “prime time” in their practices. Rather, they should be considering how they will adjust their practice patterns to the changing landscape of medicine to maximize patient benefit while minimizing potential harms, including costs.

EDITOR’S NOTE: What is precision medicine? Some define it narrowly as “genomic medicine,” or as tailoring diagnosis and treatment based on a patient’s genetic information. If so, then like Drs. Prasad and Obley, I think that it has largely not yet proved its potential to improve clinical outcomes. But some, like Dr. Feero, define it broadly as taking into account all relevant variables about an individual patient, and using this information to tailor treatment. If so, then that seems like a long-standing principle of care, with good evidence of improved outcomes. The degree of controversy over precision medicine’s value hinges on the definition used—genomic/molecular medicine vs. individualized care (with or without genetic information). We hope that these pro/con editorials shed light on a topic of heightened interest in this day of home DNA kits, and counter some of the potential hype of unfulfilled genetic solutions for age-old clinical problems.—Jay Siwek, MD, Editor, *American Family Physician*

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Author disclosure: No relevant financial affiliations.

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