Genomics in Primary Care Practice

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KEYWORDS

• Primary care • Preventive care • Family health history • Ancestry • Genomic • Value

KEY POINTS

- Family health history is a proxy for genomic testing, and can inform and guide preventive care plans.
- Family health history can be a useful tool to discuss risk and engage patients in preventive care plans.
- Both family health history and DNA analysis provide probabilistic risk, not deterministic risk.
- Both bloodline ancestry and shared environmental factors are important predictors for many disease states.
- Education at all levels is needed to stay current and to provide efficient genomically informed, value-based care.
- Genetic counselors can be valuable partners and resources to interpret and educate patients regarding genomic risk and risk associated with family health history.

INTRODUCTION

Since the completion of the Human Genome Project in 2003, clinicians and patients alike have anticipated the promise of genomics. We have hoped that genomic information would provide answers to several age-old questions: What causes disease? Why are some people afflicted over others? Why do some people respond to certain therapies over others? Particularly for primary care clinicians, it was hoped that genomics would provide additional information that would allow us to more precisely predict and prevent disease. Instead, we have observed that progress has been slow in the realm of disease prediction. While new discoveries linking genomics and disease are published almost weekly, the use of genomics in standard clinical practice, in most fields other than oncology, has lagged behind. For various reasons including

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outstanding questions of disease association versus prediction, unproven clinical utility, cost of clinical trials, genomic testing and analytics, limited reimbursement, and a lag in clinician education, genomics has been slow to reach its potential promise in primary care medicine.

Compounding the questions surrounding the fundamental clinical utility of genomic information for risk prediction is the change we are currently facing with health care reform (eg, the Patient Protection and Affordable Care Act). We have reached a new era in health care: one in which costs will be heavily scrutinized, quality and impact on clinical outcomes will need to be proved, and value has become the new buzzword. The specific problem we are facing is that in the United States, health care spending continues to increase and is expected to reach 18.4% of the gross domestic product by 2017.[1] Yet despite our high levels of health care spending, when compared with other developed nations, the United States consistently performs poorly in measures of access, outcomes, and quality of care. We have reached an era in which we are increasingly being held accountable for following evidence-based standards of care and for which care coordination will be critical. How does genomics fit into this model, and what will its role be?

This article aims to discuss the current role of genomics for risk assessment in primary care practice and how it might interface with value-based health care initiatives. The US Preventive Services Task Force (USPSTF) has issued some evidence-based recommendations for personalized prevention based on family history or genetic testing.[2] In the United States, the Evaluating Genomic Applications in Practice and Prevention Working Group, an independent expert group similar to the USPSTF, makes recommendations about clinical applications of genomic tests, based on systematic reviews of evidence.[3] Through use of a case example, these recommendations are critically explored with the goal of highlighting their applicability and utility within the primary care setting.

Case discussion: the effects of gene variants on disease risk are complex, so the predictive value of genomic profiles for individuals is limited

Mrs Jones is a 50-year-old woman with no significant medical history, who has just moved from another state and presents for a new patient annual preventive care/physical examination visit with Dr Brown, a primary care physician. She brings with her the results of a personal genomic test that she received as a 50th birthday gift. Of most interest to her, these results indicate that she is at increased risk for coronary heart disease and decreased risk for colon cancer. Mrs Jones has been dreading her screening colonoscopy, which she knows is recommended at age 50. She asks Dr Brown what she should do about the elevated risk for coronary heart disease and whether she can forgo screening colonoscopy based on the personal genomic test results (**Box 1**).

This case highlights 2 important features: first, the use of genomics in the adult primary care setting is sometimes driven by patients; second, clinicians need to understand and be able to educate patients about the limitations of genomic testing. The cost of personal genomic testing has fallen dramatically in the last 5 years, and is now as low as US$99. As a result, it is reasonable to assume that with a push in consumer advertising[4] the uptake and interest in such tests will increase. Although the medical community has considered that such information might be psychologically harmful to patients, harm has not been shown in several studies.[5,6] In fact, studies have shown that consumers have properly understood their results,[6,7] but it is unclear as to whether such information has influenced long-term change in behavior. Regardless, it is clear that there is interest among laypeople to have access to genomic