The sequencing of the human genome has led to faster and more efficient approaches to genomic analysis and the emergence of direct-to-consumer genetic tests. As a result, some have envisioned that an individual’s genome soon will be an integral part of the medical record, along with family history, lifestyle and other personal descriptors. Before that goal is realized, however, there are a number of obstacles to be overcome, in terms of technical and clinical utility, cost-effectiveness, regulation and ethical considerations.

First, the clinical utility of genome-wide association studies has not been adequately demonstrated for most common diseases. In general, research will need to expand to include populations other than Europeans and to address rare genetic variants rather than—or in addition to—the common ones currently assessed. As recently as August, researchers identified several technical challenges facing next-generation sequencing.

Also, personal genomics does not take into account nongenomic data, such as diet, exercise, epigenetics or exposures to environmental stimuli. Indeed, the few studies that have sought to assess whether access to genetic information has an impact on personal health behaviors have shown little change in lifestyle behaviors after reporting of genetic test results.

Finally, formal evaluations of genomic tests have not demonstrated a widespread clinical utility. The California Technology Assessment Forum evaluated four genetic tests and found that two of them failed to meet criteria of safety, efficacy and improvement in health outcomes. One expert recently told a Government Accountability Office investigator that “the most accurate way for these companies to predict disease risks would be for them to charge consumers $500 for DNA and family medical history information, throw out the DNA, and make predictions based solely on the family history information.” After a year-long investigation, the GAO demonstrated that companies such as 23andMe, Navigenics, Pathway Genomics and Decode Genetics offered misleading and inconsistent risk estimates to their clients. The GAO accused 10 of the 15 companies it investigated of engaging in “deceptive marketing, misinformation and questionable practices.”

It is also imperative that the costs of not only DNA sequencing but also of the entire genetic testing enterprise decrease substantially before the technology is widely adopted. Infrastructure is needed to integrate genetic information with other health data. According to genomic pioneer Craig Venter, “We don’t have the computational infrastructure to compare even thousands of genotypes and phenotypes with each other;” let alone the vastly larger dataset envisioned for healthcare settings. We also will need exceptionally good information aggregators and filters to sift through the enormous amounts of data generated by whole genome sequencing.

If useful information can be gained at a reasonable cost, then healthcare providers will need training to interpret these data, and payers will need to devise new ways of reimbursing clinicians for discussing genetic information with patients. Moreover, knowledge of supposed genetic risks could drive up other healthcare costs, inciting a “cascade effect” of unwarranted follow-up diagnostics and interventions. Thus, even if a genome can be sequenced for $1,000, it probably will not be clinically useful unless thousands of additional dollars are spent in interpretation.

The regulatory environment of personal genomics is in flux. It is unclear whether the Food and Drug Administration will approve laboratory-developed tests, as evidenced by the controversy sparked in May of this year, when Walgreens announced its proposal to sell Pathway Genomics test kits directly to consumers; the drugstore eventually dropped the proposal.

Furthermore, there is concern that access to genetic information may lead to discrimination, and that the Genetic Information Non-discrimination Act of 2008, while a step in the right direction, still has loopholes, for example in life insurance and pre-existing conditions. States such as New York and California have sent letters to direct-to-consumer genetic testing companies requiring that all genetic tests be requested by physicians, aligning with recent American Medical Association recommendations that genetic testing be conducted only under the supervision of a “qualified healthcare professional.”

The limited clinical utility demonstrated by personal genomics raises a number of important ethical considerations. At the heart of the matter is a tension between an individual’s right to know his or her own genetic information and the information deliverer’s obligation to do no harm. It is imperative, then, to clarify the responsibilities direct-to-consumer companies have to their clients.

This relationship is in between a physician-patient relationship, in which the physician has a duty to “do no harm,” and a fee-for-service market transaction, in which the consumer is assumed to have informed himself of the relative risks and benefits and therefore has the freedom to buy the service. In either relationship, the service provider must be a qualified professional, and it is uncertain—especially in light of the GAO findings—whether the direct-to-consumer companies’ employees meet basic requirements of genetic or healthcare counseling.

Finally, as with all genetic testing, regulators will need to consider how to protect the relatives of the individuals undergoing testing. The results of one’s own genetic exploration necessarily have implications for the members of his or her family. Direct-to-consumer companies, and indeed all professionals involved in the disclosure of genetic information, will need to address whether a “duty to warn” exists in this context as it does in the clinical setting.

Rapid advances in genetic analysis and sequencing technologies have generated enthusiasm for the translation of genomic discoveries into the healthcare setting, ushering in what some have termed the “age of personalized medicine.” Weaknesses exist, however, in the regulatory frameworks and cost-benefit models surrounding the new technology and the impact it will have on patient care. Questions about effectiveness, cost, legality and ethics will need to be answered before we truly welcome this new age.