Published in final edited form as:
JAMA. 2008 December 10; 300(22): 2669–2671. doi:10.1001/jama.2008.803.

Raiding the Medical Commons: An Unwelcome Side Effect of Direct-to-Consumer Personal Genome Testing

Amy L. McGuire[1,*] and **Wylie Burke**[2]

[1] Center for Medical Ethics and Health Policy, Baylor College of Medicine, Houston TX

[2] Department of Medical History and Ethics, University of Washington, Seattle WA

It is now possible for individuals to learn about their genetic susceptibility to dozens of common and complex disorders, such as coronary artery disease, diabetes, obesity, prostate cancer, and Alzheimer's disease, without ever seeing a physician. Direct-to-consumer personal genome testing companies, such as 23andMe, Navigenics, and deCODEme hope to empower consumers to take control of their health by providing tailored assessments of genetic risk based on reported associations between genomic variation and susceptibility to disease.

Several states limit or forbid this practice as a violation of state law that requires the appropriate involvement of a licensed physician when providing medical diagnostic information (1). Personal genome testing companies claim that their services are for informational and educational purposes only. They warn consumers that the information should not be used for diagnosis, treatment, or health ascertainment purposes and direct them to their physician if they have questions or concerns about their health status (2–3).

Because of uncertainty about the validity and clinical utility of test results, Hunter and colleagues advise physicians to discourage patients from pursuing personal genome testing and to respond to test results with general statements about their limited predictive value (4). While this response is consistent with current knowledge, a recent survey of online social networking users suggests that at least some potential consumers would expect their physician to help them interpret test results and believe that physicians have a professional obligation to do so (ALM, unpublished data). This expectation has important implications for primary care physicians, even pediatricians, because many direct-to-consumer personal genome testing companies allow testing of children as well as adults.

Primary care physicians already spend much of their time helping patients understand and manage health risks. Assessment of cardiac risk factors, occupational exposures and other health indicators allow physicians to identify health risks and counsel patients accordingly. Physicians are also accustomed to talking with patients about health information disclosed on the internet or through other media outlets. At the same time, primary care physicians have limited time with patients, face many competing demands (5), and are poorly reimbursed for time spent counseling patients about preventive care. Patient concerns about direct-to-consumer test results have the potential to exacerbate these problems and strain already limited health care resources.

Please address correspondence to: Amy L. McGuire, JD, PhD, Assistant Professor of Medicine and Medical Ethics Center for Medical Ethics, and Health Policy, Baylor College of Medicine, One Baylor Plaza, MS 420, Houston, Texas 77030, Phone: 713-798-2029, Fax: 713-798-5678, amcguire@bcm.edu.

Raiding the Medical Commons

The clinical value, if any, of most direct-to-consumer personal genome tests remains unproven. A statistically significant association between a particular genomic variant and a disease does not necessarily mean that the presence of that variant in a given individual is clinically meaningful. Many of the variants discovered in genome-wide association studies (GWAS) are associated with only marginal increases in risk, with odds ratios often 1.5 or less (4). The usefulness of this information for clinical decision making is unclear.

To the extent that personal genome test results lack clinical value, the time spent discussing them with patients detracts from time spent on other activities more relevant to the health of the patient. Worse, it could result in a cascade effect, in which ambiguous, incidental, or false positive results lead to further work-up that creates anxiety, cost, and potential harm (6). If a patient asks about an increased risk for cancer or heart disease, even if the increase is small, a physician may feel obligated to offer tests such as CT scans, colonoscopy, or other procedures. These studies will increase cost, may result in more false positive test results, and could expose the patient to unnecessary and potentially harmful side effects, such as radiation exposure (7). A labeling effect may also occur, leading healthy patients to view themselves (or their children) as impaired based on abnormal test findings (6). Without specific, proven actions to improve health based on test results, the potential for net harm from unnecessary or unproven health care is significant. It also represents a drain on the health care system, limiting the resources available for other established health care interventions. Similar to other screening tests-or procedures of questionable clinical value that have been marketed direct-to-consumer, , such as whole body CT scans(8), ordering follow-up tests and providing treatment on the basis of direct-to-consumer personal genome tests of indeterminate clinical value constitutes a raid on the medical commons (9). As Cassel and Brennan have argued, physicians' professional obligations include providing leadership to reduce the waste of health care resources (10); this role must include efforts to limit the unnecessary use of those resources that could flow from the marketing of genomic profiling to consumers.

Of course, the hope is that as knowledge increases, some genomic information will prove clinically useful. Research is needed to evaluate the predictive value of genomic tests and their potential to improve the use of clinically effective interventions. Tests shown by such research to improve health outcomes should be incorporated into clinical practice, with health insurance coverage of testing and any indicated follow-up.

Charting a Responsible Approach

Several initiatives can help to avoid inappropriate expenditure of health care resources and at the same time prepare for the medical integration of clinically meaningful genomic test results. The first, as called for by the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS), is increased pre-market scrutiny of genetic tests (11). Currently, most genetic tests like other laboratory tests enter the marketplace without any prior regulatory review. One result is the availability over the internet of genetic tests that lack robust evidence for claims about risk, let alone the potential that testing might improve health outcome (12). Pre-market review could assure that all genetic tests offered to consumers provide valid information.

However, pre-market review cannot prevent a drain on the medical commons. Many gene-disease associations are well validated, and arguably, companies have a right to offer such tests to consumers for informational or educational purposes (2–3), even if their clinical utility is unknown. High quality information sources are therefore needed to minimize premature incursions of this technology into patient care.

Before genomic risk information can be considered as a useful clinical tool, several questions need to be answered: What is the level of risk identified by a positive test result and what modifying factors affect risk? What clinical actions are justified by this risk information? And what are the health outcomes when this risk information is incorporated into health care? Rigorous answers require transparency; sources of evidence should be specified, and any inferences from the evidence need to be justified.

For most of the tests now included in personal genomic profiles, attempts to answer these questions will reveal only limited information: the risks are small and data on health benefit, or even on test use, are lacking. To establish clinical utility, promoters will need to provide convincing evidence that the test meets established standards for screening, such as reliable identification of asymptomatic individuals, improvement in health outcome with early treatment, and acceptability of the testing and treatment program (13). Over time, evidence for clinical utility is likely to emerge for some test uses and not others.

Creating the kind of information sources that consumers and primary care physicians need-reliable, unbiased, regularly up-dated, and readily available-takes resources, as does the research required to establish the clinical utility of new genetic and genomic tests. Yet assessment of medical innovation is dramatically underfunded. Emanual and colleagues estimate that fewer than 0.05% of US health care funding is devoted to technology assessment (14), and in a review of published literature, Khoury and colleagues found that no more than 3% of genomic studies addressed questions related to clinical integration and health outcome (12).

If genomic research is to achieve its promise, investments in health outcomes research, health technology assessment, clinical practice guidelines, and information tools will need to grow. In the meantime, strategies are needed to provide physicians and consumers with timely and complete information about what we do and do not (yet) know about new genomic products.

Companies providing personal genome testing to consumers are not an unbiased source of information. Yet it would be reasonable to require such companies to assist in the process of creating useful information tools. In addition to pre-market review to assure that tests offered to consumers meet validity standards, test developers might also be required to provide information about any research that has assessed health outcomes after testing (specifying the interventions used for individuals who tested positive, as well as populations and study designs utilized) and about any adverse effects of testing and follow-up care. Companies may utilize a scientific advisory board to determine which scientific findings to include in their service (2–3). Information about the research relied upon, as well as its potential limitations, should be shared with consumers. Also, some direct-to-consumer personal genome companies are leveraging consumer data and using it, along with phenotypic information collected with consent, to study genotype-phenotype associations (2). In order for their research to be interpretable it must conform to best practices of measurement and study design, including attention to biases in sample selection and limitations of self-reported phenotype. Although these companies are not subject to federal regulation for the protection of human subjects of research, their studies should be conducted according to accepted scientific and ethical standards. These standards may be difficult for companies to accomplish, but ad hoc, underpowered, or biased analyses cannot provide an adequate substitute. Finally, results of these studies should be provided in the form of peer-reviewed publications when possible.

For most tests now being offered to consumers, there are simply no data on the outcomes of testing. A clear statement about the absence of such data would be an important aid to physicians tasked with counseling their patients about personal genome testing, and should help to protect the medical commons from unnecessary health care based on test results. To

make interpretation of the lack of evidence easier, mandated language might be considered for example, a statement to accompany test results might say “Outcomes of testing have not been studied. These tests may have no effect on health, or may have beneficial or harmful effects.” Because patients often make decisions on the basis of unconscious associations and assumptions rather than conscious deliberation(15), physicians counseling patients about personal genome testing should assess individual expectations, elicit underlying motivations, and correct false assumptions.

We expect that test manufacturers will be eager to disclose robust evidence that supports health benefits from test use. As the evidence accumulates, formal health technology assessment will be worth pursuing, as a first step toward evidence-based use of personal genomics in health care.

Acknowledgments

Funded by the Greenwall Foundation Faculty Scholars Program in Bioethics, NIH R01HG004333, and NIH P50HG003374. The authors have no competing interests to disclose.

References

1. Genetics and Public Policy Center. Survey of Direct-to-Consumer Testing Statutes and Regulations. Jun2007 [Accessed October 27, 2008]. Available at: www.dnapolicy.org/resources/DTCStateLawChart.pdf
2. 23andMe. [Accessed August 20, 2008]. Available at: https://www.23andme.com/
3. Navigenics. [Accessed August 20, 2008]. Available at: http://www.navigenics.com/
4. Hunter DJ, Khoury MJ, Drazen JM. Letting the Genome out of the Bottle --Will We Get Our Wish? N Engl J Med 2008;358(2):105–107. [PubMed: 18184955]
5. Yarnall KS, Pollak KI, Østbye T, Krause KM, Michener JL. Primary care: Is There Enough Time for Prevention? Am J Public Health 2003;93(4):635–641. [PubMed: 12660210]
6. Deyo RA. Cascade Effects of Medical Technology. Annu Rev Public Health 2002;23:23–44. [PubMed: 11910053]
7. Brenner DJ, Hall EJ. Computed Tomography An Increasing Source of Radiation Exposure. N Engl J Med 2007;357(22):2277–2284. [PubMed: 18046031]
8. Fenton JJ, Deyo RA. Patient Self-Referral for Radiologic Screening Tests: Clinical and Ethical Concerns. J Am Board Fam Pract 2003;16:494–501. [PubMed: 14963076]
9. Hiatt HH. Protecting the Medical Commons: Who is Responsible? N Engl J Med 1975;293(5):235–41. [PubMed: 1143304]
10. Cassel CK, Breenan TE. Managing Medical Resources: Return to the Commons? JAMA 2007;297 (22):2518–2521. [PubMed: 17565087]
11. Secretary’s Advisory Committee on Genetics, Health, and Society. U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services. Department of Health and Human Services; Apr. 2008 Available at http://www4.od.nih.gov/oba/sacghs/reports/SACGHS_oversight_report.pdf
12. Khoury MJ, Gwinn M, Yoon PW, Dowling N, Moore CA, Bradley L. The Continuum of Translation Research in Genomic Medicine: How Can We Accelerate the Appropriate Integration of Human Genome Discoveries into Health Care and Disease Prevention? Genet Med 2007;9(10):665–74. [PubMed: 18073579]
13. Wilson, JMG.; Jungner, F. Principles and Practice of Screening for Disease. World Health Organization; Geneva: 1968. (Public Health Papers No. 34)
14. Emanuel EJ, Fuchs VR, Garber AM. Essential Elements of a Technology and Outcomes Assessment Initiative. JAMA 2007;298(11):1323–1325. [PubMed: 17878424]
15. Wilson TD, Bar-Anan Y. The Unseen Mind. Science 2008;321:1046–1047. [PubMed: 18719269]