

The Informed Gatekeeper?: A Commentary on Genetic Tests, Marketing Pressure and the Role of Primary Care Physicians

Timothy A. Caulfield

I. Introduction

Many have predicted that the rapid advances occurring in the field of genetics will have a profound impact on our health care system. Though we have yet to see a large number of clinically useful, broadly applicable, genetic technologies,¹ there is little doubt that numerous new genetic tests are on the horizon. These tests will inevitably be brought to the public through the significant involvement of industry. While such involvement is both necessary and desirable—indeed, it is difficult to imagine how these technologies could be developed and disseminated without the participation of the private sector—the commercialization of these technologies has also been identified with an array of social, ethical and health policy concerns.

Of course, these concerns are not unique to the use of genetic testing technologies. From the utilization of pharmaceuticals to the ordering of diagnostic services, the issues linked to commercial pressure in health care have been with us a long time.² A recent and controversial study done at the University of British Columbia concluded that drug companies artificially create demand for prescription drugs by raising public fear about disease.³ Nevertheless, the complexity and ambiguous nature of the information generated by genetic testing, together with the broad social and ethical concerns

associated with these technologies, make the commercialization concerns in this context particularly challenging.

The goal of this brief paper is to highlight the essential role that primary care physicians can play in mediating the commercialization concerns. For most Canadians, the primary care physician remains the initial entry point into our health care system. Even private testing companies, such as Myriad Genetics, require samples to be sent by a physician. As such, primary care physicians are well placed to play a central role as patient educator.

A related concern is that market pressures will lead commercial testing companies to sell their services to an inappropriately broad sector of the population.

II. The Concerns

Much has been written about the potential adverse impact of market forces in this context.⁴ For example, commentators have hypothesized that commercialization pressure may lead, or has already led, to the implementation of genetic services before their efficacy and social ramification have been appropriately evaluated.⁵ It has been argued, for example, that financial interests and professional enthusiasm led to the premature commercialization of the tests for the APOE4 and the BRCA1/2 mutations.⁶

A related concern is that market pressures will lead commercial testing companies to sell their services to an inappropriately broad sector of the population (i.e., the broader the definition of “at-risk population” the bigger the market) and that industry-generated interest in testing may cause unnecessary anxiety in

individuals who may not, but for the marketing, be interested in testing.⁷ As noted by Buchanan, *et al.*, “Given the public’s lack of understanding of genetics and genetic causation, and the seriousness of disease linked to defective genes, the road to profit can lie through stimulating public fears.”⁸

III. The Health Care Provider’s Role

These and other commercialization concerns have led a number of policy groups to recommend more regulatory oversight of the genetic testing industry,⁹ including increased monitoring of the advertising and marketing of genetic tests.¹⁰ For example, it has been suggested that federal agencies, such as the US FDA and Health Canada, take a more active role in the assessment of laboratory based genetic testing.

Though such regulatory measures would help to ensure the accuracy of marketing information and the quality of testing procedures, it seems unlikely that regulation alone could address all the identified concerns. Industry marketing strategies that are well within the parameters of a given regulatory scheme can, nonetheless, have a profound impact on the use of a health care product. The pharmaceutical industry, for instance, is highly regulated but many commercialization concerns remain (e.g., the over utilization of certain pharmaceuticals).

What else can be done to address these issues? Because so many of the commercialization issues are tied to the impact of marketing pressures on the use and interpretation of genetic tests, health care providers can play a central role in mitigating many of these concerns. Though the provision of genetic testing services will ideally involve a variety of health care providers, including specialists in genetic counselling, family physicians remain the initial point of contact for patients wishing to access health care services. It seems likely, therefore, that they will bear the brunt of patient inquiries about genetic tests. Indeed, for a number of the privately available tests, such as those provided by Myriad Genetics, the primary care physician may be the only required “gatekeeper.”

Assessing and Critiquing Marketing Material

How can primary care physicians help to address the issues associated with the commercialization process?

First, they can, on behalf of their patients, assess the claims and recommendations of industry. As noted above, a private genetic testing company has a natural and understandable desire to expand the size of its market. This can lead to genetic testing recommendations that are much more inclusive than those produced by independent organizations. For example, Myriad Genetics, the company which holds the patent on the BRCA1/2 test, will test anyone so long as the request comes through a physician.¹¹ Groups independent from industry, however, have been more conservative in the assessment of the test’s clinical value.¹² Indeed, the uncertainties regarding the utility of this test “have led advisory bodies including the American Society for Clinical Oncology to recommend that testing be restricted to women from high-risk breast or ovarian cancer families.”¹³

It has also been noted that the information provided by testing companies may stress the “seriousness” of a disorder in order to encourage people to get tested. For example, one study found that pamphlets for cystic fibrosis testing provided by commercial companies differed from university and hospital pamphlets in that they “minimize positive descriptions of CF.”¹⁴

In order for primary care physicians to effectively critique industry marketing material, they will need to be familiar with testing guidelines and recommendations from a broad set of stakeholders, particularly those produced by entities, such as professional organizations, independent of the genetic testing industry.

Mediating Genetic Enthusiasm

Second, physicians can help patients to understand the current limited clinical utility and possible social ramifications of genetic testing. Given the large amount of “genohype” which surrounds genetic technologies,¹⁵ many patients may be unaware of the complexity and often ambiguous nature of a testing result. For instance, despite the fact the test for the BRCA1/2 mutation was one of the first put into clinical use, there is still disagreement surrounding its value and the clinical significance of a positive test.¹⁶

While the initial uptake of genetic testing services has not been as robust as predicted,¹⁷ there is no doubt that much of the public has a strong interest in genetic testing.¹⁸ For example, one study found that 83% of men and 76% of women in the general public expressed the intention to be tested for breast or prostate cancer risk.¹⁹ Another study found that 82% of the general population was found to be interested in hereditary cancer risk testing.²⁰

Of course, the genetic industry is clearly not the sole source of this “genetic enthusiasm.” The understandable eagerness of the scientific community and the representations of genetics in the

media have undoubtedly played a large role in producing the hype which surrounds genetic testing technologies (Andrews, 1999; Caulfield, 2000). That said, we should not forget that marketing pressures come in many forms; press releases on new genetic discoveries, advertising directly to health care professionals and informal lobbying by scientist involved in the genetic testing companies are but a few examples of potential sources of industry pressure. To a large degree, the public, the media and, even, independent scientists all hear the same optimistic story. In such a climate, primary care physicians should be a source of a more tempered portrayal. Such an approach may have an impact on patients' decisions about genetic testing. As noted by Holtzman in the context of BRCA1/2 testing: "When women at risk of breast cancer learned about the predictive uncertainty of testing from sources independent of the companies offering them, they were much less eager to have tests."²¹

However, in a world where the notion of patient autonomy seems to have evolved into a perceived right of access, addressing patient enthusiasm for genetic testing may prove a significant challenge. In one study, for instance, it was found that 95% of the women (first degree relatives of women with breast cancer) thought they should be able to get testing despite a physician's recommendation to the contrary.²² The goal for health care providers in this context, however, should not be to actively discourage use but to ensure that autonomous decision making and non-directive counselling is not endangered by the "aggressive marketing of genetic tests."²³ As noted by Biesecker and Marteau:

In a milieu in which marketing materials promote testing and providers have incentives to encourage patients to undergo testing, non-coercive, personal decision-making about genetic testing may well be compromised.²⁴

IV. Complicating Factors

Private Testing

As waiting lists for access to genetic services grow, and as opportunities to privately purchase testing services

increase,²⁵ the responsibilities of primary care physicians will grow even more complex. Patients who either do not meet established criteria for publicly funded testing or who are on a long waiting list may wish to purchase a genetic testing service privately. In such circumstances, primary care physicians may be the only health care provider involved in the ordering and interpretation of the test,²⁶ thus heightening the significance of their counselling role.

However, in a world where the notion of patient autonomy seems to have evolved into a perceived right of access, addressing patient enthusiasm for genetic testing may prove a significant challenge.

Legal Pressures

Medical legal concerns will also play a role in this context. For example, Canadian law places onerous informed consent obligations on physicians. In fact, one could argue that many of the suggestions made in this paper are already part of a physician's disclosure obligations.²⁷ Physicians have a legal obligation to provide patients with information that a reasonable person in the patient's position would want to know. This likely includes information about the efficacy of genetic testing, alternatives and social, ethical and legal issues.

However, the law may also exacerbate a number of the commercialization problems by encouraging physicians to utilize genetic services. As testing becomes more common and as patients' expectations rise, physicians may feel the need to practice defensive medicine. That is, physicians may succumb to the demands of patients or may believe that they should simply recommend testing in order to avoid liability. Because of the high profile nature of wrongful birth law suits, defensive practices might be particularly problematic in relation to the use of prenatal diagnosis.²⁸ Given the invasive nature of genetic testing and PND in particular, physicians should strive to find more constructive ways to address liability concerns (e.g., enhancing communication skills).²⁹

V. Conclusion: The Need for Education and Training

In order for primary care physicians to play this important "informed gatekeeper" role, they must have the requisite education and communication skills. At the current time, however, it appears that many physicians are not equipped with the knowledge base or counselling skills required to provide patients with information about genetic testing services.³⁰ As such, programs are necessary to train both medical students and practising physicians.

Given the rapid pace of scientific advances in genetics, developing training programs in this area will undoubtedly



prove a daunting task - a fact recognized by the federal government. In March 2000, Health Canada established the Working Group on Public and Professional Educational Requirements Related to Genetic Testing of Late Onset Disease. One of the stated goals of the group is to "[i]dentify the educational/learning needs regarding genetic testing of late onset diseases among health professionals and the public." Recommendations from the group should be forthcoming in the near future.

There is no doubt that the suggestions outlined in this paper place an onerous obligation on primary care physicians to learn about emerging technologies and to spend the time necessary to communicate effectively with their patients about the risks and benefits of genetic testing. Some may feel that the time pressures faced by primary care physicians make fulfilling these obligations all but impossible. This is a credible concern. However, given the primary care physicians' current position in the health care system, it seems inevitable that they will have to play a significant role in this context.

Finally, I would like to emphasize that I am not placing blame on industry. The concerns outlined above are a natural and inevitable consequence of having the private sector involved in the provision of genetic testing services - involvement that is both required and unavoidable. That said, we need to recognize that commercial pressures can have a profound impact on how technologies are used and perceived by both the public and health care professionals. Developing strategies to provide an independent and balanced perspective seems only logical.

Timothy A. Caulfield is Research Director at the Health Law Institute, and Associate Professor, Faculty of Law and Faculty of Medicine and Dentistry, University of Alberta.

1. E. Wulfsberg, "The Impact of Genetic Testing on Primary Care: Where's the Beef?" (2000) 61 Am. Family Physician 971-978.
2. For example, see A. Wazana, "Physicians and the Pharmaceutical Industry: Is a Gift Ever Just a Gift?" (2000) 283 JAMA 373-380.
3. C. Tait, "Pharmaceutical Research Manipulated, University Study Claims" *Edmonton Journal* (18 December 2000) A3.
4. E. Kodish, "Commentary: Risks and Benefits, Testing and Screening, Cancer, Genes and Dollars" (1997) 25 J. of Law, Med. and Ethics 252-55; T. Caulfield and R. Gold, "Genetic Testing, Ethical Concerns and the Role of Patent Law" (2000) 57 Clinical Genetics 370-375; T. Caulfield, "The Commercialization of Human Genetics: a Discussion of Issues Relevant to Canadian Consumers"

- (1999) 21 J Consumer Pol'y 483-526.
5. N. Holtzman, "Are Genetic Tests Adequately Regulated?" (1999) 286 Science 409 at 409; H. Welch & W. Burke, "Uncertainties in Genetic Testing for Chronic Disease" (1998) 280 JAMA 1525; L.B. Andrews, "Past as Prologue: Sobering Thoughts on Genetic Enthusiasm" (1997) 27 Seton Hall L. Rev. 893; and Kodish, *supra* note 4.
6. Andrews, *ibid*.
7. B. Koenig, *et al.*, and the Breast Cancer Working Group of the Stanford Program in Genomics, Ethics and Society, "Genetic Testing for BRCA1 and BRCA2: Recommendations of the Stanford Program in Genomics, Ethics and Society" (1998) 7 J. of Women's Health 531; and Holtzman, *supra* note 5.
8. A. Buchanan, *et al.*, *.From Chance to Choice: Genetics and Justice* (Cambridge: Cambridge University Press, 2000) at 341.
9. Secretary's Advisory Committee on Genetic Testing (SACGT) (2000) *A Public Consultation on Oversight of Genetic Tests. Federal Register* (64 FR 67273) <<http://www4.od.nih.gov/oba/sacgt12-99.htm>> ; Holtzman, *supra* note 5.
10. T. Caulfield, *et al.*, "Providing Genetic Testing Through the Private Sector: A View From Canada" (2001) ISUMA: Can. J. Pl'y Research (in press); SAGT, *ibid*; and Koenig, *supra* note 7.
11. V. Brower "News: Testing, testing... testing?" (1997) 3 Nature Med 131-32; K. Birmingham, "News: Myriad's rationale for wider testing" (1997) 3 Nature Med 709; and O. Smith, "News: breast cancer susceptibility tests still valid, companies argue" (1997) 3 Nature Med 709.
12. Kodish, *supra* note 4.
13. Wulfsburg, *supra* note 1.
14. G. Loeben, T. Marteau & B. Wilfond, "Mixed messages: presentation of information in cystic fibrosis screening pamphlets" (1998) 63 Am. J. Hum Genet 1181-9. See also B. Biesecker & T. Marteau, "The future of genetic counselling: an international perspective" (1999) 22 Nat. Gen. 133-137
15. T. Caulfield, "Underwhelmed: Hyperbole, Regulatory Policy and the Genetic Revolution" (2000) 45 McGill L.J. 437-460; and Holtzman, *supra* note 5.
16. J. Peto, *et al.* "Prevalence of BRCA1 and BRCA2 Gene Mutations in Patients With Early-Onset Breast Cancer" (1999) 91 National Cancer Inst. 943; F. Collins, "BRCA1 - Lots of Mutations, Lots of Dilemmas" (1996) 334 New Eng. J. Med. 186; H. Healy, "BRCA Genes - Bookmaking, Fortune telling, and Medical Care" (1997) New Eng. J. Med. 1448; and
16. M. Singer & R. Cebul, "BRCA1: To Test or Not to Test, That is the Question" (1997) 7 Health Matrix 163.
17. B. Peshkin & C. Lerman, "Genetic counselling and hereditary breast cancer" (1999) 353 The Lancet 2176-77.
18. S. Martin, "Most Canadians Welcome Genetic Testing" (2000) 163 CMAJ 200.
19. C. Ulrich, *et al.* "Genetic testing for cancer risk: a population survey on attitudes and intention" (1999) 1 Community Genetics 213-222.
20. M. Andrykowski, *et al.* "Hereditary cancer risk notification and testing: how interested is the general population?" (1997) 15 J Clin Oncol. 2139-48.
21. Holtzman, *supra* note 5 at 409.

22. J. Benkendorf, *et al.*, "Patients' attitudes about autonomy and confidentiality in genetic testing for breast-ovarian cancer susceptibility" (1997) 73 *Am. J. Med. Genet.* 296-303.
23. *Supra* note 14 at 135.
24. *Ibid.*
25. B. Williams-Jones, "Re-Framing the Discussion: Commercial Testing in Canada" (1999) 7 *Health L. J.* 49.
26. *Supra* note 14.
27. N. Sharpe, "Reinventing the Wheel?: Informed Consent and Genetic Testing for Breast Cancer, Cystic Fibrosis, and Huntington Disease" (1997) 22 *Queen's L.J.* 389.
28. M. Renaud, *et al.*, "Canadian Physicians and Prenatal Diagnosis: Prudence and Ambivalence", Research Volume 13, Royal Commission on New Reproductive Technologies (Ottawa, 1993).
29. T. Caulfield, "Liability in the Genetic Era: Wrongful Birth and Wrongful Life Lawsuits" (2001) 23 *J. Soc'ty Obs. and Gynae. Canada* 143-147.
30. F. Giardiello, *et al.*, "The use and interpretation of commercial APC gene testing for familial adenomatous polyposis" (1997) 335 *New Engl J. Med.* 823; A. Hunter, *et al.*, "Physician knowledge and attitudes towards molecular genetic (DNA) testing of their patients" (1998) 53 *Clin Genet* 447; C. James, *et al.*, "Are practicing and future physicians prepared to obtain informed consent? The case of genetic testing for susceptibility to breast cancer" (1998) 1 *Community Genetics* 203-212.



