Commercialization of genetic technologies is expanding the horizons for the marketing and sales of genetic tests direct-to-consumers (DTCs). This study assesses the information provision and access requirements that are in place for genetic tests that are being advertised DTC over the Internet. Sets of key words specific to DTC genetic testing were entered into popular Internet search engines to generate a list of 24 companies engaging in DTC advertising. Company requirements for physician mediation, genetic counseling arrangements, and information provision were coded to develop categories for quantitative analysis within each variable. Results showed that companies offering risk assessment and diagnostic testing were most likely to require that testing be mediated by a clinician, and to recommend physician-arranged counseling. Companies offering enhancement testing were less likely to require physician mediation of services and more likely to provide long-distance genetic counseling. DTC advertisements often provided information on disease etiology; this was most common in the case of multifactorial diseases. The majority of companies cited outside sources to support the validity of claims about clinical utility of the tests being advertised; companies offering risk assessment tests most frequently cited all information sources. DTC advertising for genetic tests that lack independent professional oversight raises troubling questions about appropriate use and interpretation of these tests by consumers and carries implications for the standards of patient care. These implications are discussed in the context of a public healthcare system.

Introduction

The vast flow of genetic information from the Human Genome Project has both fueled and been fueled in large measure by private sector investment, and this has had implications for the growth of genetic applications in the healthcare industries, particularly in the area of genetic testing. While the impact of this push toward commercialization has been most strongly felt in the United States, the expansion of the global marketplace has blurred borders as consumers in many developed and developing countries now have access to a vast array of genetic tests over the Internet from national and international providers (Mykitiuk, 2004). The World Wide Web has opened a vast and subtle avenue for companies to launch direct-to-consumer (DTC) advertising campaigns for genetic testing (Gollust et al., 2003; Williams-Jones, 2003). The market for available tests has expanded to include not only patients, but also a much larger demographic: healthy “pre-patients.” Genetic tests refers to the analysis of DNA, RNA, chromosomes, and proteins to detect heritable disease-related DNA alterations (Holtzman and Watson, 1997). In the context of this paper, the term is used specifically in reference to those tests that are marketed as being relevant to health and well-being. “Health and well-being” encompasses all aspects of physical well-being, including state of health, therapy, reproduction, and nutrition. Traditionally, genetic testing was limited to those conditions for which a mutation in a single gene was causally associated with a particular disease, called single-gene disorders (Holtzman and Watson, 1997). However, most aspects of human health that have a genetic component are not affected by any particular gene in such a clear-cut fashion, and this is particularly true of most chronic diseases. Most diseases are influenced by multiple genes and gene–environment interactions, and therefore are considered to have multifactorial etiology.

A test’s usefulness in predicting susceptibility or confirming diagnosis of a disease or condition is referred to as its clinical utility. Factors that affect the clinical utility of a genetic test
include analytical and clinical validity. Analytical validity is a function of the test’s sensitivity and specificity. Clinical validity refers to the accuracy with which a test predicts a clinical outcome. This depends upon the test’s clinical sensitivity, clinical specificity, and positive predictive value (Holtzman and Watson, 1997). Factors such as genetic heterogeneity and gene penetrance often affect a test’s clinical validity and may limit the usefulness of genetic testing for disease susceptibility (Burke, 2002). Because of these limitations, the genetic basis of health and well-being is understood by geneticists, clinicians, and other experts in probabilistic terms (Welch and Burke, 1998). In a clinical context, the benefits and limitations of a genetic test must be communicated to the patient in a language and format that is understandable enough that the patient can make an informed choice about treatment, be it for immediate therapy or some future therapy. This task has long been recognized as a challenge to the medical profession due to the complex nature of genetic information.

Canadians have access to some genetic tests through the public healthcare system (e.g., for cystic fibrosis, Huntington’s disease, and, in some provinces, breast and ovarian cancer [BOC] susceptibility). Tests are often available through the requisition of a physician or specialist (e.g., clinical geneticist), who orders the tests, reviews, and discloses the results to the patient, and arranges for appropriate individual and/or family genetic counseling. However, there are gaps and inconsistencies in provincial health insurance schemes regarding coverage of genetic services, and stringent requirements for access, such that only a select group of high-risk patients qualify and only tests considered “medically necessary” are covered (Williams-Jones, 2006). Further, the test results remain a part of a person’s permanent health record (Mykittiuk, 2004). These factors may motivate some patients to seek private genetic testing services and may shape the impact of DTC advertising campaigns for genetic tests in Canada (Williams-Jones, 2003).

Researchers, physicians, and professional organizations have raised many concerns regarding DTC commercial marketing and sales of genetic tests (Caulfield, 1998; Gray and Olopade, 2003; Williams-Jones, 2003, 2006; American College of Medical Genetics, 2004; McCabe and McCabe, 2004; Mykittiuk, 2004; Wasson, 2006; Wolfberg, 2006). Consumers may misunderstand or be misled by advertising messages and then, without consulting their physicians, order genetic tests that are not indicated and/or not approved by professional organizations. Advertisements may promote a genetically reductionistic view of health by inappropriately emphasizing genetics over lifestyle and other factors. There may be inconsistencies in standards for the provision of genetic counseling, which may lead to inadequate counseling and misinterpretation of genetic test results. While genetic test results may help some people make better informed choices about their health, they may also lead to no benefit that is unique from that which can be accrued from standard alternatives such as the advice of a healthcare professional. In the worst-case scenario, misinterpreted genetic information may lead to poor choices and potentially adverse health impacts based on genetic deterministic interpretations. Without adequate counseling, patients may decide to opt for risky and controversial preventative measures such as prophylactic surgery that they may not have been inclined toward if adequate pre- and posttest genetic counseling had been provided. Even when companies require their private testing services to be utilized under the supervision of a physician, advertisements may induce patients to pressure their physicians to provide requisitions for genetic tests that the latter may otherwise deem to be inappropriate. Directly or indirectly, DTC advertising may exert pressures on the public healthcare system and drive up healthcare costs.

Based on Internet searches conducted for the purposes of this study, health-related genetic tests advertised in online DTC advertisements are classified into three broad categories for the purposes of this study: (1) diagnostic tests, that is, tests for single high penetrance genes linked to monogenic diseases (e.g., Tay-Sachs disease and Huntington’s disease); (2) risk assessment tests, that is, tests for one or two moderate-to-low penetrance genes associated with increased risk of developing specific polygenic diseases or conditions (e.g., breast cancer and Alzheimer’s disease); and (3) enhancement tests, that is, tests for many low penetrance genes that have ramifications on general aspects of health, nutrition, and/or treatment regimens (e.g., nutrigenetic or pharmacogenetic tests, and cardiovascular health profiles). While it may be desirable to have more specific categorizations of genetic tests for counseling purposes (e.g., McPherson, 2006), the aforementioned three categories are used as an analytical framework within this study since they reflect general differences in the clinical validity of tests and the nature and degree of the risks and benefits that are to be expected as a result of testing.

One of the main arguments used to support DTC advertising for genetic tests and other healthcare products is that they increase consumer awareness of therapeutic interventions, thus empowering individuals to take an active role in managing their health (Ledley, 2002). Further, proponents suggest that DTC advertisement and provision of genetic tests enables consumers to take control of their own health. The extent to which this may be true is a function of many factors, including the accuracy and pertinence of the information provided to consumers (Hull and Prasad, 2001). Genetic information needs to be interpreted and contextualized by clinicians and counselors in order to be meaningful to individuals. The public educational value of DTC advertising for genetic testing may be limited by the complexity of genetic information and complicated social contexts surrounding genetics (Gollust et al., 2002; Henneman et al., 2004; Weigmann, 2004). Such factors increase the likelihood that advertisements may promote misunderstandings among laypeople regarding disease etiologies. Thus, genetic information provided directly to consumers may be misinterpreted by them and applied inappropriately in their decisions regarding the management of their health and particularly from tests with

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1Probability that the test result will be positive when a given genetic mutation is present.

2Probability that the test result will be negative when a given genetic mutation is absent.

3Heterogeneity refers to the fact that the same genetic disease might result from the presence of any of several different variants of the same gene (allelic heterogeneity) or the presence of several different genes (locus heterogeneity) (Holtzman and Watson, 1997).

4Gene penetrance refers to the probability that disease will develop when a disease-related genotype is present. When gene penetrance is incomplete, a test’s positive predictive value is reduced. Penetrance is incomplete when other genetic or environmental factors must be present for a phenotype to appear (Holtzman and Watson, 1997).
low clinical validity (Human Genetics Commission, 2003; Williams-Jones, 2006). A small body of literature addressing the issue of DTC advertising for genetic tests will now be reviewed in brief.

**DTC Advertising Literature**

DTC advertising in the context of healthcare refers to the promotion—through newspaper, magazine, television, and/or Internet marketing—of health products that normally require a prescription or requisition (National Library of Medicine, 2006). The first DTC advertising campaign for genetic testing was launched in 2002 by Myriad Genetics through print and television media in four cities in the United States. The 5-month campaign promoted the BRCA1/2 gene test for susceptibility to BOC, and was directed at women aged 25–54 with a strong history of familial breast cancer in order to promote awareness of “recent advances in cancer prevention and early disease detection” (Myriad Genetics, September 12, 2002). However, the campaign drew immediate criticism when a full-page advertisement in a playbill was characterized as “manipulative, misleading, and misguided” not only due to its content but also because of the context in which it was shown, that is, during a play depicting a woman’s emotional struggle with ovarian cancer (Hull and Prasad, 2001; Gollust et al., 2002). Gollust et al. (2002) argued that the advertisement exploited the hopes and fears of women by implying that genetic testing can help dispel anxiety regarding the potential for developing BOC, and did not mentioning that only 5–10% of breast cancer cases are hereditary (National Cancer Institute, 2002). The playbill advertisement also did not urge women to discuss the possibility of genetic testing with their physicians, but instead simply provided the company’s Web site (Hull and Prasad, 2001; Gollust et al., 2002).

Myriad’s television and online advertisements have also been closely scrutinized in the context of a longer history of pharmaceutical DTC advertising, Williams-Jones (2006) contends that Myriad’s ads mirrored the optimistic tone of pharmaceutical advertising in that they neglected to present risk information and thus primarily served to spur consumer demand for genetic testing, limiting their utility as source of information. For example, while the television advertisements acknowledged that only 5–10% of breast cancers are hereditary, they did not mention that the BRCA mutation may only be present in 17–25% of those cases and that for the majority of women who are highly susceptible to developing breast cancer, such susceptibility is due to mutations in other genes (National Cancer Institute, 2002; Williams-Jones, 2006).

During the BRCA ad campaign, Mouchawar et al. (2005a) documented the impact of the advertising on health service utilization in the target cities. They reported that one “Managed Care Institution” experienced a 244% increase in referrals for BRCA testing compared to the precampaign period, while the proportion of referrals of high-risk women decreased from 69% to 48%; referrals in another institution appeared to be unaffected by the campaign (Mouchawar et al., 2005a). A post-ad campaign survey of consumers and healthcare providers in the target cities found no evidence to suggest an increased interest in BOC testing among women with a strong history of familial BOC (Centers for Disease Control and Prevention, 2004). The study found that healthcare providers in the target cities generally lacked the knowledge required to advise patients about inherited BOC and related genetic testing (Centers for Disease Control and Prevention, 2004). This was indicative of a twofold failure on part of (1) the BRCA ad campaign to specifically promote increased education and awareness within the high-risk target populations and (2) the healthcare system to appropriately address the resulting increase in demand for BOC testing (National Human Genome Research Institute, 2004).

Content analyses of BRCA advertisements that have been conducted in Canadian media have led to the conclusion that the value of the service was frequently misrepresented (Donelle et al., 2004). The portrayal of genetic risk was problematic for advertisements targeted to both high-risk (Ashkenazi Jew) and low-risk (general Canadian) populations. Statistics were presented in complex and contradictory ways, and there were inconsistent messages about the value of genetic testing to determine BOC susceptibility (Donelle et al., 2004). While the study was able to differentiate between advertising to high and low genetic risk populations, it was limited to analysis of a single type of risk assessment test.

A study of representations of genetic tests in DTC advertisements in U.S. print media outlets, including Jewish community newspapers, cancer-support magazines, and pregnancy magazines, found that several of the ads overstated the value of genetic testing for clinical care, and some provided misinformation, exaggerated risks of disease development, and distorted disease risk information (Gollust et al., 2002). The authors reported that the ads failed to include risk information to balance claims of effectiveness and suggested a deterministic relationship between genes and disease. Finally, the ads described complex, confusing, anxiety-producing genetic concepts for the consumer through the use of themes such as hope, fear, peace of mind, and an appeal to the desire to assert control over the potential health outcomes (Gollust et al., 2002).

Although the informational value of such ads may be limited, some evidence indicates that DTC advertising may not be as problematic as the aforementioned studies would imply. During the BRCA ad campaign, Mouchawar et al. (2005b) found that women generally did not report anxiety or confusion as a result of the Myriad DTC advertisement, except for women with a higher self-perceived risk of BOC and Hispanic women. Physicians also did not report a significant impact on the physician–patient relationship as a result of the campaign. In a multiethnic focus group study, Bates et al. (2004) have also found that potential consumers tended to adopt critical views of the messages provided in the context of a pharmacogenetic advertisement in which race was used as a proxy to qualify individuals as suitable candidates for a drug.

This study explores DTC advertising of genetic tests online: What requirements/provisions are in place regarding physician involvement and genetic counseling arrangements, and what kinds of information are provided to the consumer? How do trends differ based on the types of genetic tests companies offer? The first question can be answered by a systematic analysis of company Web sites featuring DTC

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5This trend was also reflected in other studies in which the majority of physicians surveyed lacked adequate genetic knowledge to appropriately guide patients in the use of genetic tests (Hunter et al., 1998; Menasha et al., 2000; Pichert et al., 2003).
advertisements. The second question can be answered by comparing the results across different genetic test categories.

Methods

Companies engaging in DTC advertising were classified and analyzed with respect to five categorical variables: type(s) of tests being offered, company requirements for physician involvement, genetic counseling arrangements, type of information provided to the consumer, and source(s) of information referenced on the Web site. A qualitative approach was used to arrive at descriptive “levels” for each of the categorical variables. Advertisements were assessed using these variables and compared across test types.

Sample

The project deployed a nonrandom sampling method using popular and publicly accessible Internet search engines such as Google and Yahoo! in order to simulate a consumer’s Internet search leading to company Web sites and advertisements. Companies engaging in DTC advertising for genetic tests were sought through the combined use of key words and phrases such as “genetic test,” “kit,” and “susceptibility.”
Snowball sampling was employed by using search results (such as news articles, commentaries, and nongovernmental organization reports) as “referrals” to companies and company Web sites, where possible. In addition to the use of search engines, several popular health Web sites, such as MedLine, WebMD Health, and others, were searched using key words such as “genetic test” in order to account for the potential for referral to companies from such sources through paid endorsements and pop-up advertisements.

All the companies identified through the search were included in the sample if their Web sites promoted genetic tests related to health and well-being, directly to consumers. Companies that were clearly oriented only toward healthcare professionals or researchers were excluded. A company was considered to be engaging in Internet DTC advertising if its Web site had an explicitly or implicitly consumer-friendly interface. An explicitly consumer-oriented Web site was defined as one that has a designated section of the Web site for “patients” or “consumers.” An implicitly consumer-friendly Web site included lay-level information, that is, informational passages that directly addressed the reader as a potential user of the testing services and provided the option to order home test kits. Company names were assigned codes for the purpose of analysis.

Data collection

Analysis was done iteratively with data collection from January 2005 to January 2006. Coding was used to arrive at descriptive levels within categorical variables. The six categorical variables and their corresponding descriptive levels were as follows: (1) types of test(s) being marketed (diagnostic, risk assessment, or enhancement); (2) target market (consumers only or consumers and clinicians); (3) stated requirements for physician supervision of testing (family physician or specialist; local contracted physician; company-employed physician; no physician referral required); (4) provisions for genetic counseling (e.g., the company indicated that it was the supervising physician’s responsibility to refer consumer for counseling; counseling was provided by a local contracted genetic counselor; company provided long-distance counseling; genetic counseling was not discussed on the Web site); (5) types of information available regarding disease (disease etiology; symptoms and onset; prevention; treatment and diagnosis; genetic epidemiology; no information is provided); (6) sources of information and reference (peer-reviewed studies; unpublished data; communication articles; company research only; links to ethics literature; links to professional organizations; links to patient organizations; no sources cited).

Advertisements were classified according to the test type(s) being offered. If a company offered more than one type of test, at least one example of each of those types of tests was included in the analysis. Categorical variables were analyzed overall and stratified according to the type of genetic test being offered. Each categorical variable was summarized quantitatively according to the descriptive levels.

The methodology assumed that the policies as stated on company Web sites were up-to-date and reflective of the practices of those companies. However, a company may have tailored the means of communication with the consumer, such that individual genetic testing experiences differed based on the type of test, as well as the location, needs, and preferences of the consumer. In addition, the fluidity of information on the Internet means that the analysis of the information provided on company Web sites represents a snapshot in time of company standards, policies, and representations of genetic testing services. For the duration of this study, a number of companies that were originally found to be engaging in DTC advertising changed the content of their Web site to shift the target of advertising to clinicians only, while others that had previously neglected to make a distinction between marketing to consumers and clinicians now have a designated section of their Web sites dedicated to each target market.

Results

The Internet-based searches resulted in a sample of 24 companies (n = 24). These companies were categorized according to the types of genetic tests being advertised: (1) companies offering diagnostic tests (n1 = 13), (2) companies offering risk assessment tests (n2 = 13), and (3) companies offering enhancement tests (n3 = 10). One out of three companies advertised more than one type of genetic test (e.g., both diagnostic and risk assessment tests), and therefore subgroups were not mutually exclusive (Table 1). Figure 1 provides a mutually exclusive breakdown of the sample according to the types of genetic tests offered. More than half of all companies engaging in DTC advertising also advertised to clinicians (Fig. 2).

Where DTC provision of services was coupled with DTC advertising, testing was made possible through the use of mail-in noninvasive home test kits that often required a cheek swab to be mailed back to the company for analysis. When testing required more invasive sampling techniques (e.g., a venous blood sample), mail-in test kits were provided for sample collection at a local clinical laboratory, and genetic services were provided under the supervision of a physician.

Clinician mediation/requisition requirements

Almost half of all companies stated a requirement for physician supervision/requisition of their services (Fig. 3). On-site physicians were specified for preimplantation genetic diagnosis or prenatal testing, since such procedures are normally initiated at fertility centers. The majority of companies
offering diagnostic and risk assessment tests (7 out of 10) indicated that the genetic testing process was to be mediated by a physician. Enhancement genetic tests were least likely to be supervised by a family physician or specialist, compared to risk assessment tests. In lieu of this requirement, companies advertising enhancement tests often cautioned consumers not to make diet or lifestyle changes or alter their pharmaceutical drug regimens based on the test results without first consulting their physicians. In two cases, companies offering enhancement genetic tests referred consumers to local company-contracted healthcare professionals through whom genetic testing would be arranged.

**Genetic counseling requirements**

Three quarters of the companies in the sample recommended, arranged for, or directly provided genetic counseling services, according to their Web sites (Fig. 4). Over 4 out of
10 company Web sites stated the need for face-to-face genetic counseling by an independent healthcare professional. These Web sites either stated that it was the supervising physician’s responsibility to arrange for counseling or indicated a willingness to refer the consumer to a local genetic counselor. One-third of company Web sites indicated or implied that they would directly provide genetic counseling services to the consumer, either on-site or (more commonly) over the telephone. One quarter of companies did not state a need for genetic counseling in the advertisements on their Web sites.

Half of the companies advertising diagnostic or risk assessment tests stated or implied the need for face-to-face genetic counseling or testing (Fig. 4). Long-distance counseling was most commonly arranged in the context of risk assessment or enhancement testing and without a requirement for physician involvement. Advertisements for enhancement tests were less likely to state the need for face-to-face counseling compared to DTC advertising for risk assessment or diagnostic tests. Some company Web sites advertising enhancement tests implied that long-distance genetic counseling would be provided by dieticians or other specialists hired by the company. One company indicated that a genetic counselor was not required to interpret its test results and provided basic information about genetics, implying this would render the consumer capable of fully understanding the results.

A great deal of variability was observed in recommendations for genetic counseling on company Web sites. No formal distinctions were made between the need for pre- and posttest genetic counseling. The timing of the counseling that was to be expected was implicit on Web sites advertising in vitro or prenatal testing; for example, an in vitro fertilization (IVF) clinic required that patients present proof that they have undergone genetic counseling before undergoing any preimplantation genetic diagnosis (PGD) or prenatal screening procedures. Another clinic made it clear that the counseling was provided at the clinic before any testing was done. When genetic testing services were being provided DTC, a 1-800 number and e-mail were provided for the consumer to contact company representatives. During this contact, it would be determined which genetic test (if any) was suitable for the individual. This counseling session or screening would revolve around a series of questions regarding family history and queries regarding the candidate’s reasons for testing. Even when physician supervision of testing was required, one company provided an online “quiz,” implied to be a self-screening tool for consumers to assess their suitability as a candidate for the BRCA test.

Information provision

Most companies offering genetic tests provided consumers with background information about the disease (Fig. 5). Six out of 10 companies overall discussed disease etiology, including 9 out of 10 companies that offered risk assessment tests. The etiology of a disease or condition was sometimes implied by a company’s use of pretest questionnaires and surveys. For example, nutrigenetic (enhancement) testing required consumers to fill out a questionnaire to provide specific information on the frequency of various lifestyle/eating habits. The answers provided on this questionnaire became the basis for the interpretation of the consumer’s genetic test results.

6Company EN5.
implying a contribution of the aforesaid environmental factors to disease etiology. Other commonly discussed aspects of disease were clinical symptoms and onset, as well as prevalence of a given disease within general and “high-risk” populations. Advertisements for risk assessment tests were more likely to provide information on clinical symptoms and onset of disease. One-fifth of companies provided no background information on the disease or condition being tested.

The sources of information provided to consumers through online advertisements varied widely (Fig. 6). Claims regarding the clinical validity of the tests being offered were supported by references that ranged from communication articles (short descriptions of current research findings in scientific journals) to peer-reviewed (mostly observational) studies, and unpublished company findings. Almost half of all companies cited published peer-reviewed studies; citations of such studies were most common among companies offering risk-assessment tests and least common for companies offering diagnostic tests. Rarely did such studies directly explore the link between the genetic test being offered and the disease of interest. Use of unpublished data was low overall but greatest among companies offering diagnostic testing. Over one-third of companies provided no references to outside sources of information, and this was most common in advertisements for diagnostic tests.

Almost one half of all advertisements provided links to professional or (more commonly) patient organizations (Fig. 6). Organizational links were indicated as gateways to sources of information and support—sometimes to lend credibility to selection criteria—but rarely used by the company to qualify patients for the genetic tests being advertised. Most advertisements for risk assessment tests (e.g., hereditary breast cancer) provided referrals to professional organizations, while companies offering diagnostic or enhancement tests were less likely to provide such links (Fig. 6). One company attempted to lend false credibility to its services through inappropriate use of an organizational link. In promoting the apolipoprotein E (ApoE) test for susceptibility to Alzheimer’s disease, the company provided a link to an article on the Web site of the American Psychological Association (APA) discussing the role of variations in ApoE on memory. However, the APA does not recommend the use ApoE testing as screening measure for Alzheimer’s disease, and the American College of Medical Genetics recommends against this practice (American College of Medical Genetics, 2005).

Discussion

Trends in company policies were reflective of differences in the professional consensus regarding the clinical validity of the tests being offered, regardless of the type of genetic testing services (diagnostic, risk assessment, or enhancement) being advertised. Genetic tests with professionally accepted clinical validity (e.g., cystic fibrosis, BRCA1/2) required physician involvement, in which case genetic counseling arrangements were regarded as being the responsibility of the supervising physician. Access to controversial risk assessment tests (e.g., ApoE) and enhancement tests (e.g., most nutrigenetic tests) generally did not require physician involvement, and the extent, scope, duration, and nature of counseling were left to the discretion of the company. In the latter case, genetic counseling was commonly offered over long distances, with the quality of counseling and qualifications of the counselors remaining in question. These data are indicative of a disturbing trend in which tests with the least clinical utility are

12Company EN2; Company DIRAEN2, South Africa; Company EN1, Canada.

13Company DIRA5.
provided with the least professional oversight and counseling services. This may lead consumers to overestimate or underestimate their risks of developing health conditions with complex etiologies.

While the majority of companies provided background information on the disease being tested and even basic genetics, this information was not always complete, pertinent, or accurate. Even accurate and pertinent information can lead to confusion when it is not incorporated as part of a diagnostic approach in which healthcare professionals play an active part. When the latter interaction is absent, it becomes more likely that a consumer may be insufficiently prepared to make informed decisions regarding the testing process or results due to inadequacy or inaccuracy of the information about the clinical utility of the genetic test(s) in question. Companies providing tests with little or no professionally recognized clinical utility were most likely to provide larger volumes of information, as well as misleading or irrelevant information. These included companies providing enhancement tests (e.g., nutrigenetic tests) and risk assessment tests with low clinical utility (e.g., ApoE for early onset Alzheimer’s disease susceptibility testing). While the vast majority of company Web sites contained a discreet “disclaimers” section urging precaution in the implementation of health measures based on test results, other more prominent emotive messages contradicted these disclaimers. The fact that companies offering diagnostic tests offered little to no information or references may be understood in light of the fact that such tests are most often mediated by a physician, who is assumed to be prepared to fill that information gap.

The assumption that consumers may turn to online sources of information is justifiable. A study at the Mayo Clinic found that almost half of all patients \((n = 157)\) attending a genetics clinic reported searching for genetic information online prior to their visit, and 4 out of 10 of these individuals reported feeling confused by the information they had found (Taylor et al., 2001). Major search engines such as Google and Yahoo! now generate results with government-funded organizations (such as the U.S. National Institutes of Health) and nonprofit entities at the top of the list, thus increasing the chances that consumers will initially turn to these sources instead of commercial Web sites. However, increased partnership of the industrial sector with nonprofit entities may diminish the perceived credibility of nonprofit organizations (Williams-Jones, 2006). Further, the likelihood of consumers encountering commercial promotions increases as the Internet becomes more widely accessible and the industry expands. This can be corroborated by the fact that almost all of the Web sites examined in this study are still active after 2 years of follow-up and despite the highly competitive nature of the industry, indicating a strong actual and potential market for DTC genetic services.

The messages sent by companies through genetic test advertisements are unlikely to be ignored. DTC advertising often portrays genetic testing as a service that provides empowering knowledge about one’s body—a clever framing strategy that promotes a sense of personal entitlement to that knowledge. Evidence of the impact of pharmaceutical DTC advertising on the patient–physician relationship implies that marketing-induced demand may be manifested in the form of increased patient requests for physicians to mediate testing services (Davis, 2000; Gilbody et al., 2005). However, some evidence implies that a more complex relationship may exist between exposure to advertising and uptake of the message. Consumers may adopt critical and reasoned approaches to evaluating the advertising claims in genetic tests (Bates et al., 2004), and some advertisement may have informational value (Mouchawar et al., 2005b). Depending on health service referral processes and the preexisting strength of the physician–patient relationships, advertisements may not necessarily lead to increased pressure on physicians (Mouchawar et al., 2005b).
The increasing prevalence of DTC advertising and DTC provision of tests with low clinical validity necessitate measures to protect consumers from exploitation. In the United States and United Kingdom, there have been calls for better regulatory oversight of genetic tests to ensure the quality of tests being offered to consumers (Secretary’s Advisory Committee on Genetic Testing, 2000; Human Genetics Commission, 2002; Barrett and Hall, 2003; Genetics and Public Policy Centre, 2006; Javitt, 2006; Javitt and Hudson, 2006). The U.S. Government Accountability Office warned that nutrigenetic tests are “medically unproven and so ambiguous” that they may mislead consumers about their risk of developing various multifactorial diseases (United States Government Accountability Office, 2006).

As the rapidly evolving state of technology hastens the introduction of genetic tests to the healthcare industry, it is crucial to reevaluate whether and how these genetic services fit in the present scheme of public healthcare systems, and to what extent they should be insured and integrated (Williams-Jones, 1999; Caulfield et al. 2001). As in the case of many other health services, a full “uncoupling” of financial and market forces from clinical testing and therapeutics may not be feasible nor desirable (Caulfield, 1999). A potential partnership with public health service institutions and the pharmaceutical industry may enhance overall access to genetic tests while helping to maintain the quality of care (Wasson, 2006). Wade and Wilfond (2006) argue that offering genetic counseling services for tests with low clinical utility may be consistent with the professional values underpinning the practice of genetic counseling if clients are well informed about the benefits and limitations of the results.

While DTC advertising may be a challenge to regulate particularly when services are advertised over the Internet, concerns can be partly addressed by promoting increased education amongst physicians and publics. Strong public interest in and perceived right of access to health services suggests the need for increased education in the area of genetics and disease etiology. Public agencies from the national level to local health regions must take the lead in public education and engagement and provide the necessary information resources to sensitize consumers to the issues and concerns surrounding genetic testing. Further research is required to assess the relationship between consumer/physician demand for genetic tests and the portrayal of the value and limitations of genetic testing in DTC/direct-to-physician advertisements.

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