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ORIGINAL RESEARCH

Risky Business: Risk Perception and the Use of Medical Services among Customers of DTC Personal Genetic Testing

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Abstract Direct-to-consumer genetic testing has generated speculation about how customers will interpret results and how these interpretations will influence healthcare use and behavior; however, few empirical data on these topics exist. We conducted an online survey of DTC customers of 23andMe, deCODEme, and Navigenics to begin to address these questions. Random samples of U.S. DTC customers were invited to participate. Survey topics included demographics, perceptions of two sample DTC results, and health behaviors following DTC testing. Of 3,167 DTC customers invited, 33% (n=1,048) completed the survey. Forty-three percent of respondents had sought additional information about a health condition tested; 28% had discussed their results with a healthcare professional; and 9% had followed up with additional lab tests. Sixteen percent of respondents had changed a medication or supplement regimen, and one-third said they were being more careful about their diet. Many of these health-related behaviors were significantly associated with responses to a question that asked how participants would perceive their colon cancer risk (as low, moderate, or high) if they received a test result showing an 11% lifetime risk, as compared to 5% risk in the general population. Respondents who would consider themselves to be at high risk for colon cancer were significantly more likely to have sought information about a disease (p=0.03), discussed results with a physician (p=0.05), changed their diet (p=0.02), and started exercising more (p=0.01). Participants' personal health contexts—including personal and family history of disease and quality of self-perceived health—were also associated with health-related behaviors after testing. Subjective interpretations of genetic risk data and personal context appear to be related to health behaviors among DTC customers. Sharing DTC test results with healthcare professionals may add perceived utility to the tests.

Keywords Direct-to-consumer · Genetic testing · Survey · Risk perception · Health behavior

Introduction

Direct-to-consumer (DTC) personal genetic testing has generated controversy surrounding its benefits and harms (Bloss et al. 2011; Caulfield 2011; Evans and Green 2009). Currently, few data exist documenting the experiences of people who have received such testing services. As of August 2011, at least 28 companies were offering DTC testing to U.S. consumers for at least 385 diseases and traits (GPPC 2011), and one DTC company claimed at least 60,000 customers (23andMe 2010). Social factors, including increased access to information on the Internet and a cultural shift in which individuals are assuming more responsibility for personal health care and lifestyle choices and relying less on traditional expert advice, have made DTC medical testing an attractive and viable option. Lower costs of DTC testing products, combined with a surge in genome-wide association study data will likely accelerate demand for these products.

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It has been argued that providing consumers with DTC genetic testing will increase public access to genetic and health-related information and increase understanding of lifetime risks of common diseases. DTC testing could inform health-related decisions, increase a person's attention to his or her health, and motivate changes in behavior to reduce the risk of disease. Some supporters of DTC believe that individuals have a right to obtain genetic information about themselves without the mediation of medical professionals (Helgason and Stefansson 2010; McBride et al. 2010; Foster and Sharp 2008).

On the other hand, DTC testing challenges many long-standing tenets of clinical genetics and has provoked several concerns (Evans and Green 2009; Hogarth et al. 2008; Frueh et al. 2011). Critics of DTC genetic testing argue that many of the tests currently offered lack evidence to support their clinical validity or utility, and report genetic information that cannot be interpreted with certainty, which could lead to unnecessary healthcare and emotional distress (Kuehn 2010; Murray et al. 2010; Kutz 2010; Imai et al. 2011; Ng et al. 2009). Providing consumers with complicated and potentially unreliable data outside of the medical and counseling contexts may leave them vulnerable to inappropriate health decisions (Lachance et al. 2010; Geransarel and Einsiedel 2008) and result in increased health care costs (Deyo 2002; McGuire and Burke 2011) without clear health benefits.

The DTC debate has taken place largely in the absence of data on the attitudes, expectations, and experiences of customers who have used these services. There have been few studies examining the characteristics and motivations of DTC test consumers, how consumers interpret and act on their risk reports (McBride et al. 2010), and whether health-related responses correspond appropriately to the information received (Bloss et al. 2011). Studies conducted with research participants (Gollust et al. 2011) and the general public (Leighton et al. 2011; McGuire et al. 2009) have found widespread interest in the information DTC companies are returning. More than three quarters of participants in these studies said they would go to their doctor to get help interpreting DTC test results. In a survey of 171 physicians who were offered complementary DTC testing service, 30% ordered the test (Haga et al. 2011). No data were presented on how doctors interpreted or used their results. The largest study of DTC genetic test recipients to date surveyed a convenience sample of 2,037 employees of high tech and healthcare companies, both before and three months after they received their results (Bloss et al. 2010). The study found no significant difference in anxiety levels before and after testing and no increase in the subsequent use of additional screening tests associated with the DTC results; 27% of respondents reported sharing their results with a physician. Because participants worked for companies involved in or related to this industry and were given large discounts on the testing service as part of participation in the study, it is unclear whether these findings are generalizable to the broader group of DTC genetic test customers.

Evidence from some studies of the communication of cancer risk suggests that an individual's subjective interpretation of his or her disease risk may influence health-related decisions (Croyle and Lerman 1999; Sivell et al. 2008) and may also influence the pursuit of preventative health behaviors and surgery (Katapodi et al. 2004; Meiser and Halliday 2002). To date, no data have been collected on how DTC consumers interpret the their own reported risks, whether these interpretations are related to subsequent health behaviors, and how the process of interpretation and action is influenced by other factors such as physical health, family history, and preconceptions about particular diseases. Moreover, little data exist to support or refute the idea that receiving DNA-based test results may motivate people to change their behavior (Marteau et al. 2010).

To begin to address questions about how actual DTC test customers interpret risk result data, what they do with the information, and how their interpretations influence their behavior, we surveyed random samples of customers of three DTC genetic testing companies. The cross-sectional survey measured customers' opinions at one time point, 2 to 8 months after they had received their results.

Methods

Survey Instrument

Officials at Navigenics, 23andMe, and deCODEme agreed to a survey of random samples of their customers who had received genetic risk profiles. Four main provisions were decided upon before the companies agreed to participate. The identities of a company's invited and participating customers would not be revealed to study staff or to the other test companies. The survey would not ask about specific results returned to participants. The data would be analyzed and reported in aggregate only; direct comparisons between the three companies would not be made. Finally, the number of invitees and participants from the individual companies would remain undisclosed, to avoid revealing proprietary information about an individual company's customer volume.

A 167-item online survey, qualified by the Johns Hopkins University Institutional Review Board as exempt (NA_00023396), was designed so that the majority of questions were applicable to customers of all three companies. The instruments collected information on participants' demographics, health behaviors, motivations for purchasing the personal genetic profile, and opinions and actions taken in response to their risk profile. The companies were given the opportunity to review and comment on the survey and to

approve the final version before it was fielded. Where appropriate, questions were customized for each company. For example, questions about specific diseases or conditions were matched to diseases tested by the company whose services a given participant had used. Similarly, two disease risk scenarios shown to participants were presented in the format used by the respective companies (see Supplemental Information). These risk scenarios simulated the information a hypothetical customer would see on each company's reports for risks of diabetes and colorectal cancer. All three companies' scenario displays presented both absolute lifetime risks as well as relative risks of disease. In the first example, a fictional customer was found to have a 25% lifetime risk of developing diabetes, as compared to a 30% risk in the general population. The second scenario portrayed a fictional customer with an 11% chance of developing colorectal cancer, as compared to a 5% risk in the general population. Each company approved of the graphics and information their customers were shown. The results reported here focus in part on participants' responses to two survey questions that followed the presentation of each of these scenarios. To determine whether DTC customers could correctly discern if a stated result indicated a higher or lower risk compared to the general population, we asked: "Based solely on the information above, which answer best describes how [Mary's/Mike's] risk of developing [type 2 diabetes/ colorectal cancer] compares to the average person's risk?" Second, we measured how customers would perceive their own risk if they received the two sample risks themselves by asking "If you were [Mike/Mary], knowing this information would you consider yourself to be at: high risk, moderate risk, or low risk?"

The Web-based survey instrument was programmed and administered by Knowledge Networks (KN), a research firm specializing in the conduct of online surveys. KN provided a unique hyperlink and password for each invitee. An online pilot study was conducted among 20 customers from 23andMe and deCODEme to gather feedback on the survey questions. The survey instrument was edited and checked for logic, length, vocabulary, and reading level and it was fielded in January 2010.

Selection and Recruitment of Participants

Staff at Navigenics, 23andMe, and deCODEme sent email invitations to random samples of their customers who had purchased a genetic risk profile for medical conditions and received their results 6 to 28 weeks prior to the date invitations were sent. To be eligible, customers had to reside in the United States, be over the age of 18, have logged in to view their report, and have purchased the risk report themselves or received it as a gift from a friend or family member. People who received the report from their employer, as part of research or as a gift of the DTC company or company staff were excluded.

Invitations containing a unique link to the survey and a unique KN password were sent to the randomly selected customers thought to be eligible based on data the companies had previously collected. The initial invitations were emailed in mid-January 2010 and were directed to customers who received their results between June and November of 2009. Every 4 weeks, throughout February, March and April of 2010, companies were asked to invite eligible customers who received their results in December 2009, January 2010, February 2010 and March 2010 respectively. Potential participants were asked to follow their unique hyperlink to the KN survey website and enter the password provided in their invitation. This allowed KN to track which invitees had participated without collecting identifying information. Respondents were screened to confirm their eligibility. Eligible customers were given the survey and had 4 weeks from the date of the invitation to complete it. After 2 weeks, KN emailed study staff a list of the unique links corresponding to those who had either participated or actively opted out of participation. Using this list, the companies could identify and send a reminder email to non-participants who had not opted out. Participants received a $10.00 Amazon.com gift certificate as an incentive.

Statistical Analysis

Descriptive analyses summarized the demographic characteristics of the survey population, actions taken in response to their DTC data, and their responses to the two risk scenarios.

Relationships between survey responses—including self-perceived health, family history of disease, and subjective responses to the two sample result scenarios—and DTC customers' reports of health-related behaviors undertaken in response to their own test results were analyzed. Multiple logistic regression models using SAS version 9.2, adjusting for age, gender, race and ethnic group, education, and the company from which the test was purchased, were used to assess the relationships between these variables. Because education and income were highly correlated in this dataset, we chose to use education in our analysis, and not income, under the assumption that education is more directly related to the interpretation of test results. Simple Bonferroni corrections were used to adjust p-values for multiple comparisons.

Results

Of 3,167 invited DTC customers, 1,163 (37%) responded to the invitation, and 1,046 (33%) were eligible and completed the survey. Respondents were ruled ineligible if they received the test as a promotion or were related to an employee of one

of the companies (n=50); if they resided outside the U.S. (n=21); if they purchased only ancestry testing (n=15); if they received the test from their employer (n=13), a research study or other source (n=14); or if they had not yet viewed their results (n=4). No data on the demographics of non-respondents were available. Thirteen percent of eligible participants received the test as a gift from a friend or family member, and the remainder paid for the test themselves.

Compared to the general U.S. adult population, men, white non-Hispanics, older adults, people with higher household incomes and people with postgraduate education were overrepresented in the survey sample (Table 1).

Accuracy of an Objective Interpretation of Sample Test Results

Before they were asked about their experiences with the DTC test results, respondents were shown two sample results that might be returned in a DTC company report. Customers of 23andMe saw the results displayed in the 23andMe format. Likewise, Navigenics and deCODEme customers viewed the sample results in those companies' respective formats (see Supplemental Data).

Table 1 Demographics of direct-to-consumer personal genetic test customers participating in the survey, compared to U.S. adult population

	DTC Customers (%)	U.S. 2006–08 population 18+ (%)
Age		
18–34	21	31
35–54	34	38
55–74	42	23
75+	4	8
Women	46	51
Household income		
$0–59,999	19	58
$60,000–84,999	14	16
$85,000–124,999	23	13
$125,000+	45	15
Education attained		
0–12 years	3	45
1–4 years college	46	46
Postgraduate	54	9
Race/Ethnic group		
White non-Hispanic	87	65
Asian/PI (non-Hispanic)	5	4
Black non-Hispanic	1	12
Hispanic	3	15
AI/AN (non-Hispanic)	0.5	1
Other non-Hispanic	3.5	2

The first sample result belonged to a fictional woman named Mary, who was found to carry a genotype conferring a lifetime risk of type-2 diabetes of 25%, compared to a 30% risk in the general population. The second sample result belonged to a man named Mike, whose genotype conferred a lifetime risk of colorectal cancer of 11%, compared to 5% in the general population.

To determine whether respondents were able to correctly discern if a stated result indicated a higher or lower risk compared to that of the general population, we asked the following question: "Based solely on the information above, which answer best describes how [Mary's/Mike's] risk of developing [type 2 diabetes/colorectal cancer] compares to the average person's risk?" Ninety percent of respondents answered correctly that Mary was less likely than the general population to get the disease, whereas 5% said Mary was more likely to get the disease, and 2% did not know. Another 4% believed Mary had the same risk as the average person. Similarly, 94% correctly said that Mike was more likely to get colorectal cancer, while 3% said he was less likely to get the disease, 1% did not know, and 2% believed the risks were the same. Responses that the individual risks were the same as that in the general population were excluded from the analyses.

Twelve percent of customers disagreed with the statement "I think that the [DTC] reports are easy to understand." Adjusting for age, gender, race and ethnic group, education, income, and company used, customers who disagreed that their risk report was easy to understand were significantly more likely to answer the questions about Mary (odds ratio=3.1, p<0.0001) and Mike's risks (odds ratio=2.9, p=0.0001) incorrectly or say they did not know the answer. Of the other covariates tested, older age was significantly associated with incorrect interpretations of both sample results (Fig. 1).

Subjective Interpretations of Sample Results

In addition to determining whether DTC customers objectively understood the meaning of the sample results, we wanted to measure how customers would subjectively describe their own level of disease risk if they had received these two sample results themselves. When asked "If you were [Mike/Mary], knowing this information would you consider yourself to be at: high risk, moderate risk, or low risk?", 45% said that given Mike's elevated risk of colorectal cancer they would consider themselves to be at high risk; 42%, at moderate risk; and 13%, at low risk. In response to receiving Mary's decreased 25% lifetime risk of type 2 diabetes, 34% of respondents said they would consider themselves at low risk; 63%, at moderate risk; and 3%, at high risk.

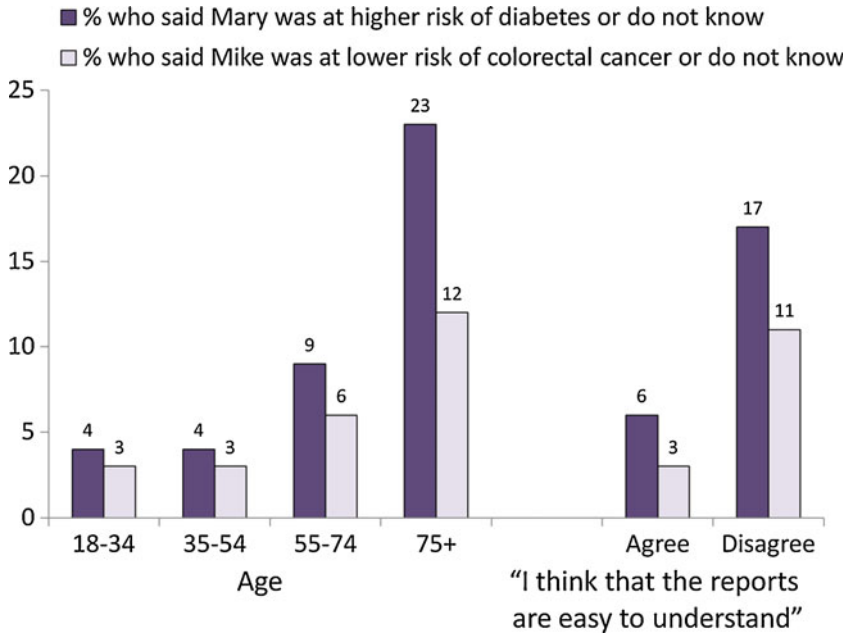

Fig. 1 Frequency of incorrect interpretations of two sample risk results presented to DTC customers

Compared to DTC customers with postgraduate education, respondents with 0–16 years of education were more likely to have lower perceived risks of both diabetes and colon cancer given the results from the two scenarios.

Health-Related Behaviors Following Test Results

A total of 43% of respondents sought additional information about at least one health condition covered in their DTC test. Twenty-eight percent discussed their results with at least one healthcare professional: 20% had shared results with a primary care physician; 1% had shared results with a genetic counselor; and 19% had told other providers (11% shared the information with more than one provider). Nearly half (49%) of the people who had discussed their results with a healthcare professional said they learned something from their DTC test results that they "did not already know and that they could use to improve their health"; the proportion who said they learned something useful was significantly lower (27%, adjusted $p<0.0001$) among those who did not discuss their results with a provider. When asked "As a result of receiving your [Company/Product] data, did you follow up with additional laboratory tests?" nearly 10% of the consumers reported following up their results with additional tests. No details were collected on the nature of these tests. Only 2% of respondents who did not share results with a healthcare provider had additional tests performed, as compared to 26% of those who had shared their results.

Sixteen percent of respondents had changed at least one medication or supplement regimen, including 10% who changed a dietary supplement, 4.4% who changed a prescription medication (0.4% did so without consulting a doctor), and 3% who changed an over-the-counter medication. Among those who changed one or more of these regimens, 54% had also shared their results with a doctor. No details were collected about the changes made or the reasons for making them.

One third of respondents said they were being more careful about their diet after viewing their results, while 14% said they were exercising more, and another 31% were "more determined to exercise." One person said s/he was being less careful about his/her diet, and nobody claimed to be exercising less. Thirty-five percent of people who had changed their diet and 32% of people who were exercising more had discussed their results with a healthcare provider.

Factors Associated with Health-Related Behaviors

Adjusting for age, DTC testing company, education, gender, race and ethnic group, several of these health-related behaviors were associated with individuals' health status and pattern of healthcare use. Participants who reported getting regular physical exams ($p=0.001$) and those who reported poorer self-perceived health ($p=0.03$) were significantly more likely to have shared their results with a healthcare provider. Participants who reported having a chronic disease ($p=0.001$) and those who had a family history of at least one condition tested for by the company they used ($p=0.05$) were more likely to have changed a prescription medication regimen. People who reported a positive family history were more likely to have gotten follow-up tests ($p=0.001$) and to have been more careful about their diet ($p=0.03$). People who reported poorer self-perceived health were more likely to have made changes in supplement regimens ($p=0.007$).

Adjusting for all of the demographic and health-related factors listed above, we also observed that people who shared their DTC results with a healthcare provider were significantly more likely to get a follow-up test ($p=0.0002$), change a prescription ($p<0.0001$) or supplement regimen ($p<0.0001$), and be more careful about their diet ($p<0.0001$)

Adjusting for all demographic factors as well as chronic disease, family history, self-perceived health, and regularity of doctor visits, participants' response to whether they would consider themselves at low, moderate or high risk upon receiving an 11% risk result for colorectal cancer, compared to 5% in the general population, was related to several health behaviors (Fig. 2). The higher a person's perceived risk of cancer, the more likely he was to have sought information about tested conditions ($p=0.03$), shared his results with a healthcare professional ($p=0.05$), paid more attention to his diet ($p=0.02$), and begun exercising more ($p=0.01$). To account for the possibility that these relationships were due to recommendations people received from healthcare professionals, we performed additional regressions adjusting for whether people had shared their results with a healthcare professional. Higher perceived risk remained significantly associated with attention to diet ($p=0.05$) and more exercise ($p=0.02$).

Finally, higher levels of education and worse self-reported current health were both significantly associated with several behaviors: seeking information about a condition ($p=0.01$; 0.004, respectively), discussing results with a healthcare professional ($p=0.0001$; $p=0.03$), getting additional laboratory tests ($p=0.02$; $p=0.04$), and changing prescription medicine regimens ($p=0.02$; $p=0.0001$).

Older participants were significantly more likely to have shared their results with a healthcare professional, changed a prescription medication, and increased their exercise (all $p<0.05$). Women were more likely than men to have changed a supplement regimen ($p=0.003$). Hispanic customers were more likely than white non-Hispanics to have changed their diet ($p=0.02$).

Discussion

Our results suggest that DTC customers' personal context and their subjective interpretation of disease risk are independently associated with certain health-related actions they take in response to receiving their test results. Specifically, contextual factors of participants' lives—including presence of a chronic disease, a family history of one or more diseases tested for by the DTC service, a poorer self-reported health, and a tendency to visit a doctor regularly—were significantly related to a number of clinically-oriented behaviors people exhibited after receiving their DTC test results. Independent of these relationships, we observed that respondents who would have considered themselves to be at higher risk than others when presented with an 11% colorectal cancer risk (as compared with 5% in the general population) were more likely to

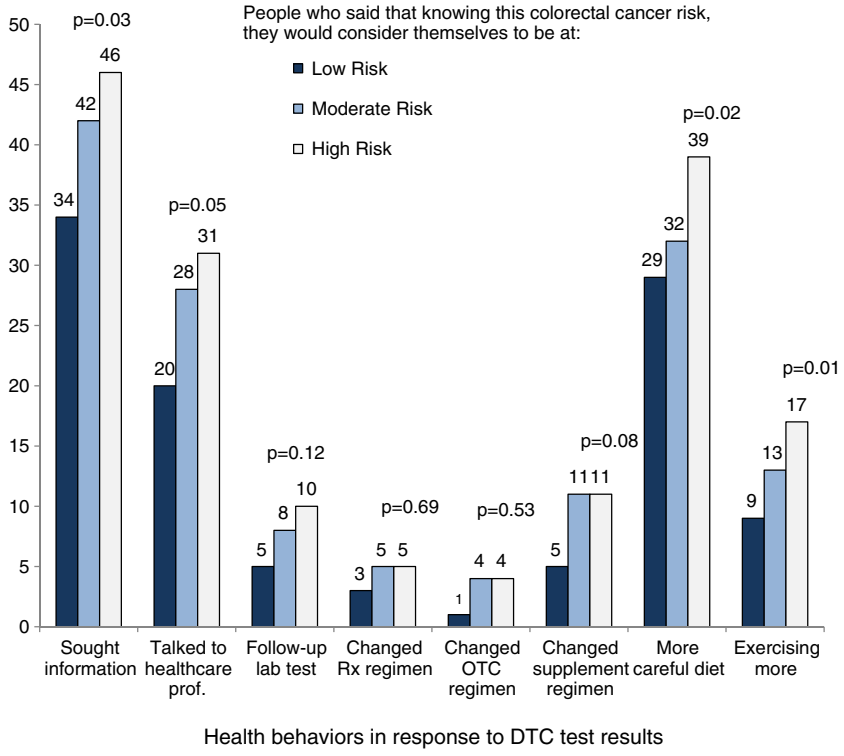

Fig. 2 The relationship between health behaviors adopted in response to results of DTC genetic testing and survey participants' perception about a sample DTC test result showing a risk of colorectal cancer of 11%, compared to a 5% risk in the general population

have taken several actions in response to their own results. The idea that an individual's personal health context and perception of a given level of disease risk relate to how s/he acts upon information is relevant to DTC companies, clinicians, and others trying to communicate complex genetic information. Discussion is necessary to explore the potential significance and applicability of these observations.

We observed independent relationships between various aspects of customers' personal health context and the health-related actions they took following testing. Personal and family history of disease, the quality of self-perceived health, and the regularity with which people visit a doctor were all predictors of various actions taken in response to test results. This finding supports previous observations that personal context can influence interpretation of cancer risk figures and subsequent decision making (Croyle and Lerman 1999; Sivell et al. 2008; Mellon et al. 2009). To address the potential importance of customers' personal situations on their interpretations of risk, DTC companies may wish to consider models of risk reporting such as the one adopted by the Coriell Personalized Medicine Collective (CPMC 2011), in which genetic risk information is augmented with risks from family history, behaviors, and co-morbidities, when such risks are known. On the other hand, it may not be realistic to think we can account for all of the relevant variables people will consider when interpreting their personal genetic data.

Independent of their personal health circumstances, we also observed a relationship between the lens through which people perceive their own risk and the actions they took in response to their results. Given the same hypothetical test result—an 11% lifetime risk of colon cancer, as compared to 5% risk in the general population—DTC customers differed in how they would characterize their own risk (low, moderate or high). The subjective interpretation of the sample colon cancer risk figure was related to several behaviors DTC consumers took in response to their own test results.

The meaning of this observed relationship depends in large part on what information is being represented in participants' responses to the question: "If you were Mike, knowing this information would you consider yourself to be at: high risk, moderate risk, or low risk?" It is tempting to assume that a person's tendency to judge the sample colorectal cancer result as more or less worrisome can serve as a good proxy for how they interpreted their actual elevated risk results (if they had received such results). This assumption is not justified a priori, however, because data were not collected on people's actual risk results, how they interpreted specific risks, or what specific steps they took in response (e.g., adopting a low cholesterol diet, changing the dose of their antidepressant, or having a full-body scan). As such, we did not make this assumption when conducting our analysis. The measurement of how people would perceive their risk if they received the sample colorectal cancer result was intended, rather, as a rudimentary proxy for the type of lens DTC customers use to interpret a test result communicating an increased risk of a serious disease. We believe the response reflects a combination of comprehension, beliefs and numeracy skills held by DTC customers at the time of the survey. These factors may include beliefs about the disease in question, one's understanding of and beliefs about absolute and relative risk including the weight one gives to these risk estimates; one's expectations of his/her own absolute or relative risk of colon cancer (in this case); or whether the risk figures presented in the example are high enough to be worrisome.

One limitation affecting our ability to interpret these results is that we do not know whether survey participants would have given the same response to this hypothetical scenario before receiving their own DTC test results (recall that surveys were completed 2 to 8 months after receiving DTC services). Participants' level of optimism or pessimism about the cancer risk figure might have been influenced by the experiences of ordering, receiving, interpreting and acting upon their DTC genetic profiles. If seeking information, discussing DTC results with a healthcare provider, or changing diet or exercise regimens increased or reinforced a tendency to view elevated risk as "high risk," this could help to explain the relationship we observed. For example, sharing data with a healthcare provider could reinforce the idea that a relatively modest increase in risk is equivalent to a "high risk," depending on the interpretation and recommendations of the provider. Of the participants who changed a prescription medication, 91% had consulted a physician. Discussing results with a provider was also significantly associated with getting follow-up laboratory tests. People receiving instructions for further testing or action from a healthcare provider might be inclined to view new risk data as more meaningful or threatening. On the other hand, studies in the breast cancer literature have found that patient interactions with genetic counselors result in decreased self-perceptions of risk (van Dijk et al. 2006; Mikkelsen et al. 2007). Moreover, in our analysis, after adjusting for whether participants shared their data with a healthcare provider, the relationships between the perception of the cancer risk and changes in exercise ($p=0.02$) and diet ($p=0.05$) remained significant. We have reason, therefore, to conclude that the act of consulting with a healthcare provider—even if influential to risk interpretations or further health-related actions—does not fully account for the relationship we observed between subjective risk interpretation and behaviors following personal DTC testing.

This suggests another possible interpretation: that people had some existing tendency to evaluate data like those shown in the colorectal cancer scenario as more or less worrisome, and that this tendency was part of the calculus people used to decide how to interpret and act upon their

own results. This hypothesis, if true, would support findings from earlier studies of cancer risk assessment that demonstrated that pre-conceived expectations of cancer risk predict behavioral reactions to genetic risk information (Hilgart et al. 2010), and individuals' subjective interpretation of disease risk figures may influence health-related decisions (Croyle and Lerman 1999; van Dijk et al. 2003; Sivell et al. 2008). People who tend to view the same piece of information as more worrisome or important than other people may be more likely to adopt one of several actions.

Whether our survey participants had long-standing opinions and numerical skills that lead them to interpret a risk figure more or less optimistically, or these interpretations were strongly influenced by the process of ordering, interpreting and acting upon their results, we believe the role that subjective interpretations of risk play in deciding future actions is important for people developing risk-communication strategies to consider. Over time, new results and interpretations will be made available to DTC customers. Individuals' health contexts may change. DTC companies will add to and adjust their methods of communicating risk information—for example, since the time of the survey, Navigenics and 23andMe have added several features to their risk reports including information on heritability and other risk factors; the effects of various tested markers on their risk calculations; and links to technical details and literature on which their calculations are based. These evolving factors will lead DTC customers to new interpretations and re-interpretation of their results, providing ample opportunity to study how people's interpretive lens and medical situations—along with a company's risk communication methods—influence consumers' understanding, risk perceptions, and their subsequent use of healthcare resources.

Because customers of all three companies were shown both relative and absolute risk estimates we cannot speak to whether one method of presenting risk data leads to more optimistic interpretations than another. One study showed that patients who viewed a risk report in the form of a figure were more likely to recommend taking action than people who were shown percentages or frequencies (Timmermans et al. 2008). It is possible that in our study, some DTC customers focused on one of the multiple representations of risk provided by the company, and that people focusing on the figures responded differently than those focusing on numerical risks.

Studies of the general public's attitudes about DTC testing found that between 78% and 92% thought they would share their DTC results with a physician if they were to get tested (McGuire et al. 2010; Leighton et al. 2011; Gollust et al. 2011). These hypothetical studies may have overestimated the proportion of consumers who would share their results with a healthcare provider. The current study supports the finding of Bloss et al. (2010) and indicates that between 25% and 30% of DTC customers share their data with a doctor or other provider in the first 3–6 months following the receipt of results. We observed that the longer customers had access to their data, the more likely they were to have shared their results with a physician (data not shown). It is thus possible that the percent of customers sharing their data might increase with time, as people have more opportunities for routine visits with their primary care physician. It is unclear to what extent the use of healthcare resources reported by our survey respondents represents an increase in care they would have sought had they not been tested. No information was collected on whether appointments with healthcare professionals were scheduled specifically to discuss the DTC results. However, those who regularly see a doctor were more likely to have shared their data with a healthcare professional, suggesting that some customers were sharing information during a regularly scheduled visit.

There is some evidence to suggest that sharing DTC risk reports with healthcare professionals may add considerable value to the test results and may facilitate appropriate health-related decisions in response to DTC results. The utility of the DTC tests, as measured in our study by "learning something new to improve one's health," was significantly higher among respondents who discussed results with a healthcare professional. Moreover, a substantial portion of reported health-related behaviors occurred among customers who consulted a physician. Nine of ten DTC customers who made a change to their prescription drug regimen did so under the guide of a physician. Additionally, 85% of those who followed up with additional laboratory tests also shared their DTC results with a healthcare professional. This is not surprising, given that prescription drugs and laboratory tests are almost always ordered by a provider; however, it provides some evidence that DTC customers making medical decisions in response to their results are consulting with healthcare professionals. Questions remain about whether DTC customers will place increased burden on the healthcare system (Giovanni et al. 2010).

Limitations

When interpreting these results, it is important to consider the survey response rate of 37%, which may be fairly typical for a web-based consumer survey using a small incentive. While the response rate is not unusual, it presents the possibility of non-response bias. Without information on the demographic makeup of non-respondents, we cannot state definitively whether this sample was subject to such a bias. It is possible that web-savvy customers are more inclined to reply to an internet survey. Nevertheless, all customers ordered and received their test results via the web, so this potential source of bias is likely to be small. Another

potential source of bias comes from the possibility that survey respondents comprise the DTC customers with the strongest opinions about these services. We cannot say whether people with strong positive opinions or strong negative opinions are more likely to be overrepresented. These opposing forces may cancel or limit the extent of such bias. It is also possible that DTC customers who are too busy or unable to respond because of health issues may be under-represented. Under these circumstances we might expect actual worry and use of health care resources to be higher than we observed, while the percentage of respondents reporting that they learned something new to improve their health may be overrepresented in our sample. It should be noted that the magnitude of non-response rate alone is a poor predictor of the presence of response bias (Groves 2006).

In comparison to the U.S. adult population, our sample of relatively early adopters of DTC genetic testing had high levels of education and household income. Given that the prices these customers paid for DTC testing ranged from $429 to $2000, this is not surprising. These demographics were also characteristic of the Coriell Personalized Medicine Collaborative, whose research participants received a small number of personal genetic risk reports (Gollust et al. 2011). As the price of genetic testing continues to decline dramatically (at the time of publication, 23andMe was selling its health test for $99 plus a monthly information fee), it would not be surprising if the demographics of DTC customers broadened considerably. Health behaviors we observed varied by the education level of the customers; respondents with more education were more likely to share and investigate their results, and had different subjective assessments of the sample risks. As the DTC genetic testing customer base expands to include larger numbers of lower-income and less well-educated individuals, the potential health benefits of DTC testing suggested by our findings may be less generalizable.

More than 90% of participants correctly interpreted whether the sample results meant that the person receiving the result had a higher or lower disease risk than the general population. Nevertheless, the fact that participants are able to understand the objective nature of the risks being communicated is distinct from whether those risks are accurate, useful, or acted upon appropriately. This survey did not collect data to address these questions.

A recent meta-analysis concluded that there is little evidence to support the hypothesis that receiving DNA-based test results motivates people to change their behavior (Marteau et al. 2010). Our study did not have the ability to determine whether DTC genetic tests led to improved health outcomes. While some changes in behavior were observed, we do not know whether any of these changes were appropriate responses to test results or individuals' health. Changes in medications, diet or exercise could be beneficial, neutral or harmful. This cross-sectional study measured customers' behaviors at one point in time. At best, the observation that measureable proportions of customers are watching their diets and exercising gives reason for cautious optimism that DTC testing could lead to improved health. Additional qualitative and quantitative research studies that follow DTC customers from the time they order their results through the time they act upon them would contribute to our understanding of how DTC customers' views and interpretations about risk change across the process, what parts of the process influence their decisions to take health-related actions, whether newly adopted habits such as diet change are sustained over time, and whether health outcomes improve as a result.

Considerable work is required to examine whether DTC customers can make use of and cope with genetic data that have little or no established clinical utility. Accepting the notion that our entire sequences will soon be available to us, we believe that studying the understanding and behavior of early recipients of these data can inform the processes and policies that will be developed as we learn to interpret our own genomic results.

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