# Value of Genetic Testing for Hereditary Colorectal Cancer in a Probability-Based US Online Sample

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***Background***. *While choices about genetic testing are increasingly common for patients and families, and public opinion surveys suggest public interest in genomics, it is not known how adults from the general population value genetic testing for heritable conditions. We sought to understand in a US sample the relative value of the characteristics of genetic tests to identify risk of hereditary colorectal cancer, among the first genomic applications with evidence to support its translation to clinical settings.* ***Methods***. *A Web-enabled choice-format conjoint survey was conducted with adults age 50 years and older from a probability-based US panel. Participants were asked to make a series of choices between 2 hypothetical blood tests that differed in risk of false-negative test, privacy, and cost. Random parameters logit models were used to estimate preferences, the dollar value of genetic information, and intent to have genetic testing.* ***Results***. *A total of 355 individuals completed choice-format questions. Cost and privacy were more highly valued than reducing the chance of a false-negative result. Most (97% [95% confidence interval (CI)], 95%–99%) would have genetic testing to reduce the risk of dying of colorectal cancer in the best scenario (no false negatives, results disclosed to primary care physician). Only 41% (95% CI, 25%–57%) would have genetic testing in the worst case (20% false negatives, results disclosed to insurance company).* ***Conclusions***. *Given the characteristics and levels included in the choice, if false-negative test results are unlikely and results are shared with a primary care physician, the majority would have genetic testing. As genomic services become widely available, primary care professionals will need to be increasingly knowledgeable about genetic testing decisions.* ***Key words:*** *genetics; oncology; internal medicine; discrete choice; utility assessment.* ***(Med Decis Making 2015;35:734–744)***

Personalized medicine—health care targeted to the characteristics of individuals, including genetics—has developed rapidly during the past decade. Patients and members of the public say they would be tested to prevent disease in themselves or family members.[1,2] Some personalized medicine applications are used to determine optimal treatments and are only relevant to individuals with specific conditions. Others are used to identify at-risk individuals who might benefit from more intensive screening or prophylactic treatment and are relevant to larger segments of the asymptomatic population.

Genetic testing used in risk assessment for hereditary colorectal cancer, specifically Lynch syndrome or hereditary nonpolyposis colorectal cancer (HNPCC), has empirical evidence for its clinical validity and utility.[3,4] Evidence-based guidelines for hereditary colorectal cancer have been widely disseminated,[5–8] and cost-effectiveness evaluations have been conducted.[9–11] The purpose of genetic testing in this case is to identify those who would benefit from more frequent and intensive screening or from prophylactic surgery, such as colectomy.[12] While the mutations associated with hereditary colorectal cancer are not common, many people will have questions about colorectal cancer, a highly prevalent disease, and whether they or related family members would benefit from genetic testing.

Many view genetics as ‘‘a very good or good thing,’’[13] but patients and the public also have significant concerns about genetic testing, including cost, accuracy, and potential for discrimination based on genetics.[14–17] Individuals facing these decisions make complex tradeoffs among these factors. Beyond studies

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of attitudes toward testing, there is little quantitative information on how individuals at risk for health conditions or members of the general public weigh costs with benefits.[18] This information is critical for health professionals who will need to be prepared for discussions about genetic testing with their patients. Without knowledge of tradeoffs, it is not possible to fully understand the value of genetic testing or the factors associated with its adoption and utilization.[19] In this study, we sought to examine the relative value of specific characteristics of genetic testing for hereditary colorectal cancer in a probability-based sample of adults 50 years and older from the general US population, a group for whom routine colorectal cancer screening is relevant.

## METHODS

We used a choice-format conjoint survey to measure the value of genetic testing in a probability-based sample of adults from the US population. Choice-format conjoint, also known as choice-based conjoint (CBC), is a form of conjoint analysis. Over the past decade, this type of survey has been used increasingly to quantify preferences for characteristics of health care and policy.[20–23] These surveys simulate clinical and policy decision making and provide a systematic method of eliciting tradeoffs to quantify the relative importance that individuals place on treatment characteristics or outcomes. This approach is based on the premise that medical interventions are composed of sets of characteristics (e.g., efficacy, safety) and that the relative value of a particular intervention is a function of these characteristics.[24]

### Survey

The construction of choice alternatives used in the survey was based on 7 focus groups of clinical experts, average-risk community members, and patients at risk for hereditary cancer (total $n$ = 42). There were 2 groups of average-risk community members ($n$ = 19), 2 groups of high-risk and cancer patients ($n$ = 8), 1 group of genetic counselors ($n$ = 3), and 2 groups of physicians ($n$ = 12). The focus groups were moderated by 2 experienced qualitative researchers using a structured guide and were recorded using a digital audio recorder. Also, each group was observed by several of the investigators who took handwritten notes. A whiteboard was used to record the specific characteristics and levels of these characteristics identified by focus group members as important in decisions about genetic testing. After each group, a digital photograph was taken to record the material from the whiteboard.

Two genetic testing scenarios were presented to the groups for discussion. The questions included in the structured guide were the same for each group, but the perspective was different. For example, while patients were asked about their personal likelihood of being tested, physicians were asked how likely they thought their patients were to be tested, given a particular scenario. Each focus group included 2 formats: 1) an open-ended discussion of genetic testing and 2) a highly structured discussion of the specific characteristics that focus group members indicated had influenced or would influence their decisions about genetic testing.

To understand how the characteristics might influence decisions about genetic testing, the focus groups included a highly structured discussion of potential levels or categories for each characteristic (e.g., accuracy: 0%, 10%, or 20% chance of a false negative; privacy: primary care doctor, genetics health professionals, or insurance companies will receive genetic test results). During the structured discussion, we asked focus group members to identify a range of relevant levels or categories for each characteristic. For

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characteristics such as sensitivity, specificity, false positive, and false negative, we provided definitions in plain language and with verbal and graphical illustration appropriate for those of lower numeracy. Definitions and supporting graphics were displayed on a whiteboard that was visible to all in the room. We then moderated a discussion of the levels and categories leading to refinement of the relevant and important levels and categories. Finally, we asked the members vote on the highest, lowest, and intermediate levels or categories that were most meaningful to decisions about genetic testing.

Three experienced qualitative investigators coded the verified transcripts of the recordings and the notes using a content analysis approach.[25] First, the 3 coders reviewed the transcripts and defined a total of 10 unique coding categories. Second, after the categories were clearly defined and a coding manual developed, the investigators then conducted an initial round of coding each transcript. Discrepancies in coding were resolved through discussion, and the coding manual was refined. A final round of coding the transcripts was then conducted again with discrepancies being resolved through discussion.

Analysis of the transcripts generated a range of test characteristics important to decisions about genetic testing for cancer risk. While the discussions of the patient and community member groups weighed heavily in the considerations, input from all groups was considered in selecting the characteristics and levels to be used in the choice task. Among these, we selected specific test features based on 2 considerations: 1) frequency of mentions and 2) conceptual distinctness. This allowed us to construct choice alternatives that included as many relevant characteristics as possible while constraining the total number to reduce the cognitive burden of the choice task. In addition, we considered the potential of selected characteristics to provide information on the relative importance of one test characteristic compared with another. For example, while concerns about privacy are well known, no quantitative information is available on the importance of privacy compared with other characteristics, such as accuracy.

The final selection of levels and categories was based on both the focus group discussions and the current literature. For example, cost levels were based on what focus group members said they would pay for genetic testing and what was published in the literature on typical copayments for genetic tests to identify Lynch syndrome. In addition, categories or levels were selected to provide contrasts that had not been examined in previous studies and that would reveal new information about the characteristics. For example, privacy categories were based on focus group mentions of primary care doctors, genetic counselors and other genetic specialists, and insurance companies as potential test result recipients in addition to the person being tested. While previous surveys have suggested that the public values primary care physicians as sources of information about genetic testing, the relative value of involving primary care compared with other health professionals, such as genetic specialists, also valued for their knowledge, is not known.

The final choice alternatives included 3 genetic test characteristics: accuracy (false-negative results), privacy (who other than the person being tested has access to the results), and cost (personal cost not covered by insurance).[25,26] While test accuracy can be described in several ways, we selected one dimension of accuracy to include among the choice alternatives to examine comparisons between distinct characteristics. Among the dimensions of accuracy, we selected chance of a false-negative test result because our focus groups and prior studies have indicated that both patients and community members see as important having information about cancer or a predisposition for cancer so that something can be done and see as concerning having a cancer risk or a genetic mutation even when test results are normal.[25,27,28]

Each characteristic was associated with 3 of 4 levels or categories, as shown in Table 1. One level or category was used in each choice task alternative. The fractional factorial design used to create the survey versions was generated using SAS version 9.2 (SAS Institute, Cary, NC). To create test profiles for the choice questions, we employed a D-optimal algorithm to construct a fractional factorial main-effects experimental design in SAS version 9.2, resulting in 36 choice pairs.[29–31] The final experimental design consisted of 4 survey versions, each containing 9 choice questions. Each respondent was randomly assigned to 1 of the 4 versions, and the 9 choice questions were randomized in each survey version.

Each choice task included 2 hypothetical genetic test alternatives (blood test A, blood test B) and a no-test option. The no-test option was included so we could estimate predicted test uptake. Including the no-test option was also important in making the choice scenario more realistic to participants, as in the real world, ''no test'' is an option they can choose.

Each set of alternatives (blood test A, blood test B) was presented with a hypothetical level of colorectal cancer risk (i.e., 10%, 25%, 50%) (Figure 1) to

**Table 1** Domains, Characteristics, and Levels for the Choice Tasks[a]

| Domain | Characteristic | Levels |
|---|---|---|
| Risk[b] | Chance that you will get colorectal cancer | 10 out of 100 (10%)<br>25 out of 100 (25%)<br>50 out of 100 (50%) |
| Accuracy | Chance of a false-negative test result (the test result says people DO NOT HAVE the gene when people actually DO HAVE it) | 0 out of 10 times (0%)<br>1 out of 10 times (10%)<br>2 out of 10 times (20%) |
| Privacy | In addition to you, who else sees the test results | Your primary care doctor<br>Your genetics health professionals<br>Your life insurance and health insurance companies |
| Cost | Personal cost to you not covered by insurance | \$250<br>\$500<br>\$1000 or \$1500[c] |

a. The domains, characteristics, and levels were developed using focus groups of high-risk clinic patients, average risk community members, and clinicians and pretested using structured interviews of community members.
b. Risk of colorectal cancer given the presence of the genetic mutation.
c. Half the participants saw \$1000, and half the participants saw \$1500.

provide a baseline context for each choice task. The level of risk was varied throughout the choice tasks in the questionnaire. In the full experimental design (36 choice questions), each of the 3 baseline risk levels occurred exactly 33% of the time (24/72). In addition, in each of the 4 versions, each baseline risk occurred 33% of the time (6/18).

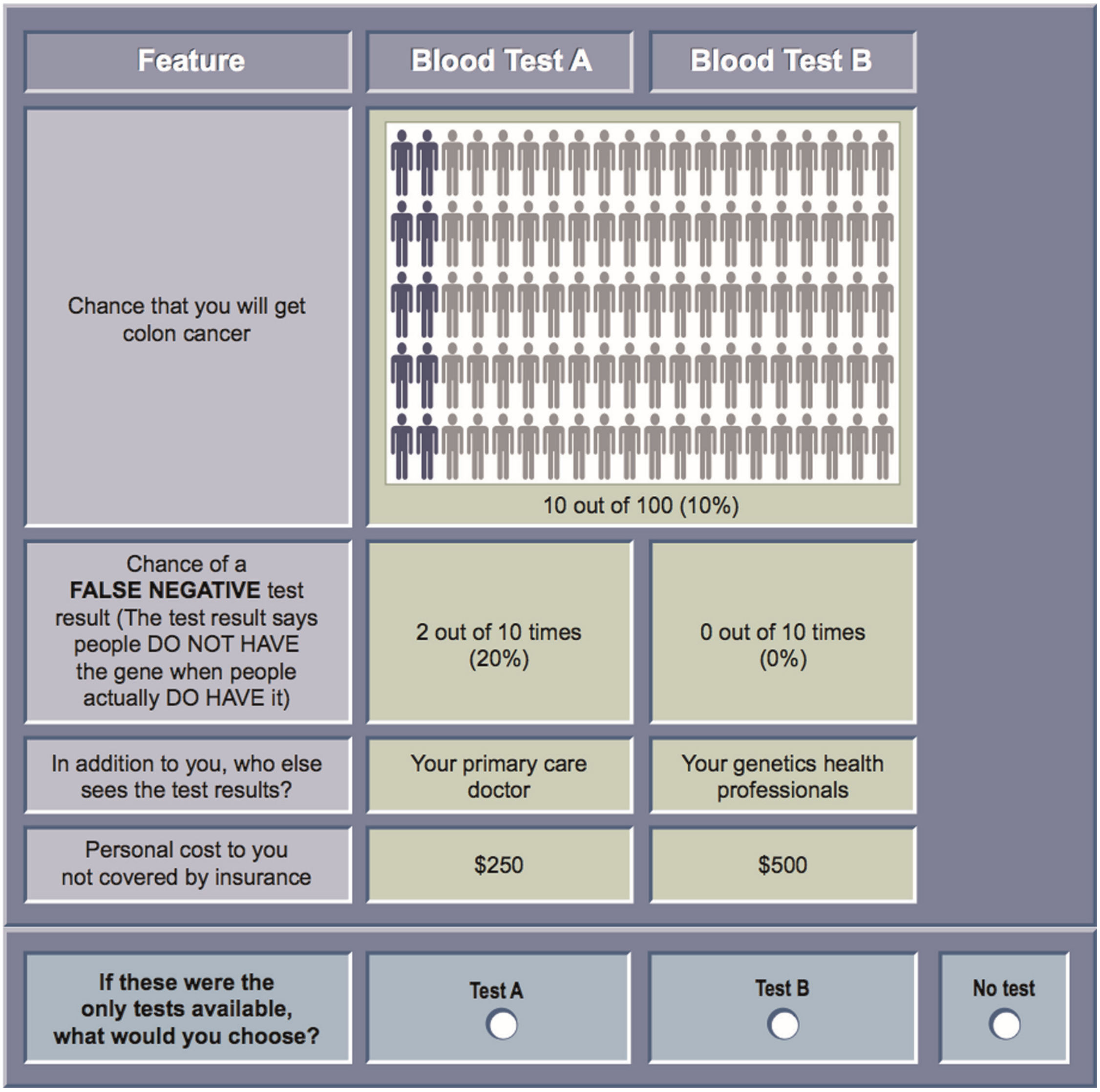
The survey included several questions in addition to the choice tasks. Because genetic test results for colorectal cancer have consequent risk reduction recommendations, we included a separate question on choice of risk reduction strategy given hereditary colorectal cancer in the family. In this question, we asked participants to choose either colectomy to eliminate the risk or colonoscopy to reduce the risk of dying of colorectal cancer. If colectomy was selected, the participant was asked about the maximum that he or she would pay for a colectomy. If colonoscopy was selected, the participant was asked to identify the maximum that he or she would pay for colonoscopies over a lifetime. Several independent questions were included in the survey to measure relevant person characteristics (e.g., cancer history) that were mentioned in the focus groups, but that cannot be manipulated experimentally.

Because understanding numerical probabilities is often cognitively challenging, we used pictographs to provide graphical risk information.[32] Plain-language definitions and pictures were used to define concepts used in the choice tasks, such as false-negative test result, genetics health professionals, colectomy, and colonoscopy.

We tested and refined the initial version of the survey using structured interviews with 10 community-dwelling adults age 50 years and older. We asked participants to ''think aloud'' as they completed the survey and then asked debriefing questions to determine whether they understood definitions and instructions and accepted the hypothetical context of the survey. Survey questions and the choice task were refined after initial feedback from the first 5 participants. Using the final version of the survey, we confirmed that participants saw the characteristics and levels as relevant and of concern and were willing to accept tradeoffs among the levels of characteristics.

On the basis of the International Society for Pharmacoeconomics and Outcomes Research recommendations for good research practice, we used a standard algorithm to construct a fractional-factorial experimental design. This type of design is advantageous because it maximizes the statistical information obtainable from choice tasks using fewer questions than required in a full factorial design (i.e., all combinations of characteristics and levels or categories).[33,34] The final survey consisted of 4 versions, each including 9 choice tasks. Participants were randomly assigned to 1 of the 4 versions.

Each survey was designed to take approximately 20 minutes to complete. In addition to choice tasks, the survey included a written introduction to the



| Feature | Blood Test A | Blood Test B |
|---|---|---|
| Chance that you will get colon cancer | 10 out of 100 (10%) | |
| Chance of a **FALSE NEGATIVE** test result (The test result says people DO NOT HAVE the gene when people actually DO HAVE it) | 2 out of 10 times (20%) | 0 out of 10 times (0%) |
| In addition to you, who else sees the test results? | Your primary care doctor | Your genetics health professionals |
| Personal cost to you not covered by insurance | $250 | $500 |

| | | | |
|---|---|---|---|
| **If these were the only tests available, what would you choose?** | **Test A** ○ | **Test B** ○ | **No test** ○ |

*Figure 1 Choice question example.*

topic of genetic testing for colorectal cancer and questions about demographics, personal and family history of cancer, experience with genetic testing, and comprehension of risk information.

The final survey was approved by the University of California at San Francisco Committee on Human Research Participation and the RTI International Office of Research Protection and Ethics (Research Triangle Park, NC). Participants were required to provide online informed consent by clicking on the statement ''Yes, I agree to participate.''

## Procedures

Knowledge Networks administered the Web-enabled survey to members of their online survey panel in April 2010. Using a combined random-digit dial sample from the US landline population and an address-based sample from the US Postal Service National Address File, the panel is a representative, probability-based sample of the US population. Probability-based samples are preferred to non-probability based samples because of their greater representativeness of the US population.[35] Knowledge Networks panel members who do not already have Internet service are provided with Web access.

Participants were eligible for the survey if they were 50 years or older and a US resident. Of the 650 members of the Knowledge Networks panel asked to participate in the survey in April 2010, 451 were determined to be eligible and completed the survey (70% participation rate). The conjoint analysis included the 355 surveys that met data quality standards needed for analytic model assumptions. Those with incomplete responses to the question on value of risk reduction with colectomy or colonoscopy ($n = 5$) and those who selected the same genetic test alternative indiscriminately (test A, test B, no test) for all nine choices ($n = 91$) were excluded from the analysis as is standard.[36] These response patterns may indicate that the survey respondents were not paying attention to the choice questions. Survey pretesting demonstrated that participants similar to those in the online panel understood the instructions for the choice questions, making poor comprehension of the choice task an unlikely explanation for these responses. However, because these responses inflate error in the model estimates, it is common practice to omit such participants in the analysis.

### Analysis

We estimated random-parameters logit models using NLOGIT 4.0 (Econometric Software, Inc., Plainview, NY) to obtain preference parameters from the choice-format questions.[37,38] This analysis yields relative preference weights for all levels of each characteristic included in the survey and estimates of the relative importance of each characteristic over the range of levels included in the survey. The observed pattern of answers to the choice questions in a conjoint survey reveals the relative importance of each characteristic to participants, the rate at which participants are willing to accept tradeoffs among characteristics and levels, and the relative value of different combinations of levels.

The final model included all of the characteristics shown in Table 1—colorectal cancer risk, accuracy, privacy, and the personal cost of the test not covered by insurance. We used effects coding in the model to estimate the reference levels as the sum of the rest of the preference weights for each attribute. To estimate the standard errors, we used the variance-covariance matrix. Once we obtained the standard error, we estimated the $z$ score as the beta/standard error and calculated the $P$ value.

We obtained the participant-specific value of avoiding colorectal cancer risk by combining the specific colorectal cancer risk information shown to that participant (i.e., 10%, 25%, 50%) and that participant's response to the questions about willingness to undergo colectomy or colonoscopy to eliminate or reduce the risk of having colorectal cancer. In the analysis, the baseline risk variable was interacted with the risk reduction level chosen in the questions about willingness to undergo colectomy or colonoscopy. This value was then used to adjust the estimate for risk of a false-negative test result. This was done to provide individual-level information on the value of test accuracy or avoiding the consequences of a false-negative test.

To provide a common metric for the value of genetic test alternatives, we calculated the monetary value of genetic testing across participants as the difference in the value of a particular genetic testing alternative and the value of the no-test alternative divided by the value per dollar of cost. The overall value of genetic testing compared with no testing was calculated by setting the test characteristics (e.g., cost, privacy, accuracy) to the mean values in the experimental design. This calculation provides the mean maximum dollar amounts that participants would pay for privacy (who other than the person being tested has access to genetic test results) and for test accuracy (risk of a false-negative test result). The value of test accuracy was adjusted by the value of risk reduction as measured by the questions on the risk reduction strategy. Thus, the value of accuracy depends partly on the perceived value of avoiding a mistake (i.e., an undetected genetic mutation).

## RESULTS

### Sample Characteristics

Participant age was on average 63 years (range, 50–96 years) (Table 2). More than half were white (77%), were married (62%), and had at least some college education (54%). A majority had health insurance (89%), and more than half had previous experience with colonoscopy (62%). Relatively few had experience with genetic testing (8%), had a personal history of cancer (14%), or had a family history of colon cancer (14%).

### Value of Genetic Test Characteristics

All 3 genetic test characteristics (i.e., cost, privacy, accuracy) were included in estimating

**Table 2** Demographic and Clinical Characteristics ($n$ = 355)

| Characteristic | Value[a] |
|---|---|
| Age, mean (range), y | 63 (50–96)[b] |
| Sex | |
| Male | 176 (50) |
| Female | 179 (50) |
| Children | |
| Yes | 289 (82) |
| No | 65 (18) |
| Missing | 1 |
| Race/ethnicity | |
| White, non-Hispanic | 273 (77) |
| Black, non-Hispanic | 37 (10) |
| Hispanic | 19 (5) |
| Other, non-Hispanic | 14 (4) |
| Two or more races, non-Hispanic | 12 (3) |
| Educational attainment | |
| Less than high school | 40 (11) |
| High school | 122 (34) |
| Some college | 107 (30) |
| Bachelor's degree or higher | 86 (24) |
| Marital status | |
| Married | 218 (61) |
| Widowed | 38 (11) |
| Divorced | 48 (14) |
| Separated | 6 (2) |
| Never married | 33 (9) |
| Living with partner | 12 (3) |
| Household income level | |
| Less than $25,000 | 78 (22) |
| $25,000 to $49,999 | 98 (28) |
| $50,000 to $74,999 | 81 (23) |
| $75,000 to $99,999 | 47 (13) |
| $100,000 or more | 51 (14) |
| Employment status | |
| Working, as a paid employee | 109 (31) |
| Working, self-employed | 31 (9) |
| Not working, retired or disabled | 174 (49) |
| Not working, other[c] | 37 (11) |
| Health insurance (yes) | 316 (89) |
| Colonoscopy ever (yes) | 219 (62) |
| Genetic testing ever (yes) | 29 (8) |
| Blood relative with colon cancer (yes) | 49 (14) |
| Personal history of cancer ever (yes) | 51 (14) |

a. $n$ = 354 for colonoscopy, genetic testing, and cancer; $n$ = 352 for relative with colon cancer. Values are presented as number (%) unless otherwise indicated. Percent may not total to 100 due to rounding.

b. Range of age.

c. Includes "not working, but looking for work" and "not working, other."

a random-parameters logit model. Table 3 shows the logit model estimates. For each level of each characteristic, log odds (LO), also called preferences weights, are shown with their standard errors, standardized estimates ($z$ scores), and $P$ values. Table 3 also shows the estimates of the relative importance of each characteristic relative to the other characteristics included in the model and conditional on the ranges of the levels used for the characteristics. The 95% confidence interval is shown for the relative importance weights.

Higher log odds indicate higher preferences or values for a characteristic or levels of a characteristic. The level with the greatest value is assigned a preference weight of 10 (i.e., genetic testing results shared with a primary care doctor), and the level with the lowest value ($1500 on personal cost not covered by insurance) is assigned a preference weight of 0. Usually, the highest and lowest coefficients coincide with the same attribute, and we can scale between 0 and 10 for the most important attribute. In this model, the highest and lowest coefficients occurred for different attributes, and we used the same process as we would if they occurred for the same attribute. All other characteristic levels were scaled relative to these 2 levels. Characteristic levels that are more highly valued have higher preference weights than those that are less valued.

Estimated preference weights for personal cost not covered by insurance and risk of false-negative results were consistent with the natural ordering of these categories. That is, lower cost was valued more than higher cost, and better clinical outcomes were valued more than worse clinical outcomes. For privacy, genetic testing results shared with primary care doctors and genetics health professionals (LO = 0.978 and 0.684, respectively) were more highly valued than results shared with life and health insurance companies (LO = −1.661). Also, participants preferred the average test shown to no test (LO = −1.643 v. –3.286). The no-test option was preferred in only 13.6% of the choices.

The relative importance of each characteristic can be interpreted as indicating the relative value for each characteristic level compared with the mean effect of that characteristic and considering the other characteristics and levels and categories included in the model. For effect-coded characteristics (e.g., privacy), the mean effect is zero. For cost, which was modeled as linear, the mean effect is the mean level shown for cost in the survey. For the interaction of risk reduction and baseline risk on the false-negative levels, the mean effect is the effect of the combined variable. Personal cost of genetic testing not covered by insurance (LO, 3.302; 95% CI, 2.759–3.845) was considered most important relative to the other characteristics. Among the clinical characteristics,

**Table 3** Preference Weights and Relative Importance of Characteristics

| Characteristics | Levels | Coefficient[a] | SE | $z$ Score[b] | $P$ Value | Relative Importance[c] | 95% CI |
|---|---|---|---|---|---|---|---|
| Chance of a false-negative test result (the test result says people DO NOT HAVE the gene when people actually DO HAVE it)[d] | 0 out of 10 times (0%) | 0.665 | 0.084 | 7.872 | 0.000 | 1.339 | 1.021–1.657 |
| | 1 out of 10 times (10%) | 0.009 | 0.059 | 0.149 | 0.882 | | |
| | 2 out of 10 times (20%) | −0.674 | 0.088 | −7.647 | 0.000 | | |
| In addition to you, who else sees the test results? | Your primary care doctor | 0.978 | 0.092 | 10.576 | 0.000 | 2.639 | 2.240–3.038 |
| | Your genetics health professionals | 0.684 | 0.083 | 8.190 | 0.000 | | |
| | Your life insurance and health insurance companies | −1.661 | 0.124 | −13.393 | 0.000 | | |
| Personal cost to you not covered by insurance | $250 | 0.496 | 0.040 | 12.251 | 0.000 | 3.302 | 2.759–3.845 |
| | $500 | −0.165 | 0.013 | −12.251 | 0.000 | | |
| | $1000 | −1.486 | 0.121 | −12.251 | 0.000 | | |
| | $1500 | −2.806 | 0.229 | −12.251 | 0.000 | | |
| Genetic testing preference[e] | Test | −1.643 | 0.145 | −11.336 | 0.000 | 1.643 | 1.357–1.929 |
| | No test | −3.286 | 0.290 | −11.336 | 0.000 | | |

CI, confidence interval; SE, standard error.

[a] Log odds (also termed *preference weights*) relative to the mean effect of the characteristic, which are normalized at zero using $z$ scores to clearly distinguish where the differences occur between the log odds. The marginal log odds from the random-parameters logit model can be interpreted as weights indicating the relative strength of preference for each characteristic level. With this model, the relative changes between characteristic levels are the main focus. For example, the largest noncost improvement in genetic testing features occurs between life insurance and health insurance companies and the primary care doctor as test result recipients.

[b] The $z$ score is the coefficient divided by the standard error; $z$ scores are used to identify statistically significant differences between attribute levels. A $z$ score $\geq 1.96$ corresponds to a $P \leq 0.05$.

[c] Relative importance represents the weight of each characteristic (over the levels of each characteristic included in the survey), which is estimated by taking the difference in the parameter estimates between the best and worst levels for each attribute.

[d] Estimate for chance of a false negative was adjusted for the value of eliminating or reducing the risk of colorectal cancer shown in each choice question (i.e., 10%, 25%, or 50%).

[e] Comparison of no testing to the alternative of the average test.

privacy (LO, 2.639; 95% CI, 2.240–3.038) was relatively more important than accuracy (LO, 1.339; 95% CI, 1.021–1.657).

The overall monetary value of testing relative to no testing was $622 (95% CI, $476–$778) (Table 4). We also calculated the marginal monetary value for any change in preference weight from a less valued to a more valued level of accuracy and privacy. The largest marginal value was $999 (95% CI, $815–$1193) for sharing results with primary care doctors compared with insurance companies. The value of sharing test results with genetics health professionals compared with insurance companies was somewhat less at $888 (95% CI, $719–$1063). The value of reducing the chance of a false-negative test result from 20% to 0 and from 10% to 0 was $507 (95% CI, $379–$640) and $248 (95% CI, $161–$338), respectively.

### Genetic Testing Intentions

When cost was set at $500, almost all participants (97%; 95% CI, 95%–99%) would be willing to have genetic testing done given the best scenario—results shared with the primary care doctor and no chance of a false-negative test. In contrast, less than half (41%; 95% CI, 25%–57%) would be tested with results disclosed to life and health insurance companies and a 20% chance of a false-negative test.

## DISCUSSION

In this study of adults in a probability-based online panel drawn from the US general population, we found strong interest in genetic testing to identify hereditary colorectal cancer risk when personal cost not covered by insurance was set at $500 and given the best test features included in the choice task—results shared with a primary care physician and no chance of a false-negative test result. In this situation, almost all (97%) would have testing, similar to the interest expressed in members of families newly identified with hereditary colorectal cancer.[16] In contrast, when genetic information would be released to

**Table 4** Monetary Value of Test Characteristics

| Characteristic, Comparison[a] | Mean[b] (95% CI) |
|---|---|
| Chance of a false-negative test result | |
| 1 out of 10 times (10%) to 0 out of 10 times (0%) | \$248 (\$161–\$338) |
| 2 out of 10 times (20%) to 0 out of 10 times (0%) | \$507 (\$379–\$640) |
| 2 out of 10 times (20%) to 1 out of 10 times (10%) | \$258 (\$164–\$356) |
| Who else sees the test results | |
| Genetics health professionals to primary care doctor | \$111 (\$18–\$208) |
| Life insurance and health insurance companies to primary care doctor | \$999 (\$815–\$1193) |
| Life insurance and health insurance companies to genetics health professionals | \$888 (\$719–\$1063) |
| Genetic testing preference | |
| Test v. no test | \$622 (\$476–\$778) |

CI, confidence interval.

[a] A comparison between levels of a characteristic (e.g., 20% to 10% false-negative test result).

[b] Mean value in dollars of an improvement from one level to another of a characteristic. For example, the mean value in dollars of disclosing genetic test results to a primary care doctor rather than a genetics health professional is \$111.

life and health insurance companies and the chance of a false-negative result was 20%, interest in genetic testing fell to less than half of the sample. In analyses that included the personal cost of testing not covered by insurance, both cost and privacy were the most important factors in test decisions.

Acceptability of genetic testing is an important question studied primarily in families at risk for hereditary cancers[15,39] but is not well understood in the general population. In our study, we found that US adults would pay on average \$622 in out-of-pocket costs for genetic testing. A population-based study in Canada found that fewer (27%) would pay more than \$500 in out-of-pocket costs for genetic testing.[40] In contrast to samples of adults from the general population in the United States and in Canada, patients at low, medium, and high risk for breast cancer in Great Britain would pay up to £3000 (approximately \$4850) for genetic testing, a value above the cost of testing to the National Health Plan.[41]

Despite public enthusiasm for the use of genomics to individualize health care,[13,42] there are concerns about the privacy of genetic tests.[16,43,44] The Genetics Information and Nondiscrimination Act (GINA) was enacted in 2008 to supplement existing state law on genetic discrimination, providing a minimum level of protection against health insurance and employment discrimination based on genetics.[45] The findings of our population-based choice-format survey are consistent with recent public opinion studies suggesting ongoing concerns about the privacy of genetic information not fully answered by recent policy.[46]

One of the central findings of our work is that adults from the general population value the participation of primary care in the genetic testing process as much as they value genetics specialists (e.g., genetic counselors) compared with disclosure of genetic test results to insurance companies. Our results quantify the relative value of primary care professionals compared with genetic specialists, information not captured in opinion surveys indicating general positive attitudes toward primary care involvement in genetic consultation.[47] Because the workforce of genetics professionals is limited,[48] and patient and public knowledge of hereditary cancer and appropriate surveillance is incomplete,[49,50] primary care has been identified as a potential resource in the delivery of genomic health services.[51] Evidence that primary care professionals are not yet fully prepared to discuss the costs and benefits of genetic testing with their patients suggests an opportunity for professional development.[52–55]

Our results should be interpreted in light of several considerations. First, our study provides information on stated choices and not real-life decisions about genetic testing. However, our findings can be seen as indicating the perceived value of genetic testing to members of the general public, and this information is necessary for decision analyses and policy development. Second, while the sample size is small, according to published guidelines for choice-format conjoint analyses, the sample is more than sufficient to conduct the tests based on the experimental design.[34] Furthermore, the small confidence intervals associated with our estimates suggest robust results. Third, our results are not generalizable to the general US population. While our sample was recruited from a probability-based online panel of US adults, it is not representative of the entire US population possibly considering genetic testing.

Fourth, as in other choice-format studies, our findings on the importance of each characteristic should be interpreted as relative to the other characteristics and levels included in the survey. While additional research will be needed to clarify the relative value of characteristics not included in this study, the rigorous methods used to develop and pretest the survey should increase confidence in the external validity of our findings and should provide a strong basis for future studies of test characteristics and other factors.

This study of adults from the general population identified out-of-pocket costs of genetic testing and the privacy of the results as the most important characteristics associated with genetic testing. However, controlling for cost, most appear willing to be tested to reduce the risk of morbidity and mortality from a heritable condition if the test results are shared with the person's primary care physician and the chance of a false-negative test result is low. As genetic testing becomes more widely available for many conditions, primary care professionals will need to be increasingly prepared for discussing decisions about genetic testing with patients and guiding them through appropriate subspecialty consultations.

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