Impact of Delivery Models on Understanding Genomic Risk for Type 2 Diabetes

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\section*{Key Words}
Genomic risk · Risk communication · Risk comprehension · Risk perception

\section*{Abstract}
\textbf{Background:} Genetic information, typically communicated in-person by genetic counselors, can be challenging to comprehend; delivery of this information online – as is becoming more common – has the potential of increasing these challenges. \textbf{Methods:} To address the impact of the mode of delivery of genomic risk information, 300 individuals were recruited from the general public and randomized to receive genomic risk information for type 2 diabetes mellitus in-person from a board-certified genetic counselor or online through the testing company’s website. \textbf{Results:} Participants were asked to indicate their genomic risk and overall lifetime risk as reported on their test report as well as to interpret their genomic risk (increased, decreased, or same as population). For each question, 59\% of participants correctly indicated their risk. Participants who received their results in-person were more likely than those who reviewed their results online to correctly interpret their genomic risk (72 vs. 47\%, \textit{p} = 0.0002) and report their actual genomic risk (69 vs. 49\%, \textit{p} = 0.002). \textbf{Conclusions:} The delivery of personal genomic risk through a trained health professional resulted in significantly higher comprehension. Therefore, if the online delivery of genomic test results is to become more widespread, further evaluation of this method of communication may be needed to ensure the effective presentation of results to promote comprehension.

\section*{Introduction}
The increase in the understanding of the impact of genetic variation for many different conditions and traits has resulted not only in the development of more genetic and genome-based tests, but also new routes of delivery. Specifically, direct-to-consumer testing enables consumers to order a range of services from single gene to genome-wide genotyping without the involvement of a health professional. The expanded use of testing both within medical specialties highly familiar with genetic and genomic testing (pediatrics and obstetrics/gynecology) and other less experienced medical specialties has also created a need to develop new approaches to counseling. Traditionally, genetic test results are communicated in-person, often by a genetic counselor. However, results
may be returned online through the testing company or perhaps accessed through an electronic medical record, phone counseling and telemedicine.

The delivery of genetic test results through informal methods such as mail and phone has been demonstrated to be generally very reassuring with respect to anxiety, patient understanding and satisfaction [1–4], with some reported confusion and uncertainty about preventive interventions and familial risks [5]. With more than 70% of American adults estimated to have Internet access [6], the Internet presents a convenient mode for communicating test results, enabling individual control over the pace and amount of information consumed [7]. Interactive computer programs have been found to be more effective than standard genetic counseling for increasing knowledge of genetic testing [8, 9]. Comfort with online communication about genetics has been associated with previous online health information – seeking a healthy life-style and positive attitudes towards genetics [10]. However, few studies have assessed the effectiveness of online communication of genetic or genomic risk with respect to understanding one’s personal risk and decision making [11, 12].

Several factors may influence an individual’s ability to understand and emotionally process genomic risk information, including health literacy and genetic knowledge. Some evidence suggests that individuals with low health literacy may struggle to comprehend the information presented at a face-to-face genetic counseling session [13, 14]. Specifically, lack of familiarity with genetic concepts and terms as well as preconceived perceptions of personal and familial risk may pose barriers to understanding genetic or genomic test results [5]. While strong factual knowledge of genetics seems likely to result in higher levels of comprehension of genomic risk, it is unclear whether this is an essential component for understanding risk and/or adopting healthy behavior to mitigate genomic risk [15]. Some studies have reported that women with lower health literacy recalled less information about a genetic test to predict breast cancer recurrence [16, 17]; however, participants in these studies did not actually undergo genetic testing. The concept of ‘being at-risk’ for healthy individuals may also prove challenging [18]. The ‘meaning’ or implication of a test result is further framed by ethnicity and culture [19–23].

To address the effectiveness of in-person versus online delivery models for understanding genomic risk and adoption of healthy behaviors, we conducted a randomized clinical trial comparing delivery of genomic risk results for type 2 diabetes mellitus (T2DM). We also assessed the relationship between genetic knowledge and risk comprehension. We hypothesized that the delivery of genomic risk information in-person would result in greater comprehension. Furthermore, we hypothesized that the comprehension of genomic risk information would be higher in individuals with higher genetic knowledge, a positive family history, and/or college-level or higher education status. However, we also hypothesized that any differences between the 2 communication groups would be smaller in individuals with higher genetic literacy and education status. The analyses described here provide the foundation for future analysis and communication of genomic risk information to optimize public/patient understanding.

**Materials and Methods**

**Overall Study Design**

As reported elsewhere [24], we conducted a randomized study of 300 non-diabetic participants to explore the impact of (1) health literacy, genetic knowledge and attitudes about genomic testing, and (2) delivery approach of T2DM genomic risk on comprehension and perceptions of risk as well as changes in health. Participants were randomized to receive their results in-person from a certified genetic counselor or access them online through the testing laboratory’s secure website. Participants were recruited from Duke University (Durham, N.C., USA) and surrounding areas. Participants completed a pre-testing survey to assess baseline knowledge and attitudes, family history and 3 follow-up surveys on risk comprehension and behavior change post-test, respectively. This study was approved by the Institutional Review Board of Duke University Health System.

**Testing**

deCODE, Inc. conducted the T2DM genomic risk test for the study. Participants’ gender and race were submitted with a buccal swab specimen for testing. As is deCODE’s protocol, participants who self-identified as White were tested for SNPs in 4 genes: TCF7L2, PPARG, CDKAL1, and CDKN2A/B. Participants who self-identified as Asian were tested for SNPs in 3 genes: TCF7L2, CDKAL1, CDKN2A/B, and those who self-identified as African-American were tested for SNPs in the TCF7L2 gene. Participants who did not self-identify as one of these 3 categories were notified that results may not be as accurate or informative for them.

**Delivery of Test Results**

Test reports (online and print) included 2 types of risk information in varying formats: (1) relative genomic risk [numerically (e.g. 1.20)], text (e.g. 20% greater than someone of similar race and gender), and graphically [shaded bar: green (<1.0), yellow (= 1.0) and red (1.0+)], and (2) overall lifetime risk estimate [numerically (percentage)], text (e.g. 20 out of 100), and pictorially [10 people figures with affected proportion shaded]). Participants randomized to receive their test results online were provided a log-in name and password directly to the company’s site; no changes were made to deCODE’s site for the purposes of this study. Participants in the
online arm could review their results as presented on the deCODE website (online suppl. fig. 1; for all online suppl. material, see www.karger.com/doi10.1159/000358413) or download a copy of their test report from the site (online suppl. fig. 2). Participants randomized to meet with the genetic counselor to review their test results were provided a print-out of the same test report accessible to participants in the online arm. Using the test report as a script, the genetic counselor reviewed each section of the report, highlighting each result, pointing to figures or section of the report to ensure that the participant could follow along. The in-person review of results lasted approximately 10–15 min; additional time was necessary for some participants who had questions about the results or testing. Standard responses were developed to anticipate commonly asked questions. No additional information was provided without prompts from the participant to ensure that participants in both study arms received the same information as much as possible.

**Surveys**

Participants were asked to complete a battery of surveys at baseline (pre-testing) to gather information on participant demographics and to assess health literacy, genetic knowledge and attitudes about genomic testing, and disease risk perception for T2DM, as described previously [24]. After receiving their test results, participants were surveyed at 1-week, 3-months and 6-months post-testing. The 1-week post-testing assessed their understanding of the test result, risk perception, psychosocial impact, test satisfaction, intention to change health behaviors, and information-seeking behaviors. To assess risk comprehension, we asked participants to interpret their genomic risk (increased risk, decreased risk or same as population risk – also referred to as categorical genomic risk in the paper). In addition, participants were asked to indicate (open-answer questions) their actual genomic risk score (reported as a relative risk on the test report) and overall lifetime risk (reported as a percentage). Since participants completed the online survey on their own, it is possible that some used their report to answer the questions about risk scores instead of relying on recall.

To assess perceptions and emotional response to health risks, we used the validated revised Illness Perception Questionnaire (IPQ-R) [25–27]. We assessed the psychosocial impact of testing using 3 subscales from the Multidimensional Impact of Cancer Risk Assessment (MICRA) questionnaire (the validated distress, uncertainty and relief from distress) [28]. Participants were also asked with whom they shared their test results and if they sought additional health information and from what sources (e.g. health professional, internet) [29]. To assess health literacy, we used the validated Short Test of Functional Health Literacy in Adults (S-TOFHLA) [30, 31]. We used a validated 16-item survey to measure actual knowledge about genetics (such as the association between genes, chromosomes, and cells and diseases) [32] and an 11-item survey to assess perceived knowledge of medical possibilities and social consequences of genetic testing [33].

| Table 1. Characteristics of enrolled participants |
|-----------------------------------|-----------|-----------|-----------|
|                                  | Baseline (n = 300), n (%) | 1-week follow-up (n = 257)*, n (%) | 3-month follow-up (n = 239), n (%) |
| Sex                               |                       |                       |                       |
| Female                            | 210 (70)              | 188 (73)              | 175 (73)              |
| Male                              | 90 (30)               | 69 (27)               | 64 (27)               |
| Race                              |                       |                       |                       |
| Black/African-American            | 86 (29)               | 64 (25)               | 52 (22)               |
| White                             | 179 (60)              | 168 (65)              | 163 (68)              |
| Other                             | 29 (9.7)              | 20 (8)                | 19 (8)                |
| Prefer not to answer              | 4 (1.3)               | 4 (1.5)               | 4 (1.7)               |
| Unsure                            | 2 (0.7)               | 1 (0.4)               | 1 (0.4)               |
| Age                               |                       |                       |                       |
| 18–29 years                       | 131 (44)              | 108 (42)              | 104 (44)              |
| 30–39 years                       | 58 (19)               | 48 (19)               | 42 (18)               |
| 40–49 years                       | 48 (16)               | 42 (16)               | 37 (15)               |
| 50–59 years                       | 34 (11)               | 33 (13)               | 30 (13)               |
| 60–69 years                       | 28 (9)                | 25 (10)               | 25 (10)               |
| ≥70 years                         | 1 (0.3)               | 1 (0.4)               | 1 (0.4)               |
| Education                         |                       |                       |                       |
| Some high school or high school graduate | 29 (10)             | 24 (9)                | 21 (9)                |
| Some college (no degree) or Associate’s degree | 74 (25)             | 55 (21)               | 45 (19)               |
| Bachelor’s degree or higher       | 196 (65)              | 177 (69)              | 172 (72)              |
| Missing response                  | 1 (0.3)               | 1 (0.4)               | 1 (0.4)               |
| Family history of T2DM            | 210 (70)              | 180 (70)              | 167 (70)              |

*Significant differences in dropout between baseline and 1 week were observed for males (23% as compared to 10%, p = 0.006) and African-Americans and other (26 and 31%) as compared to Whites (6%).
Statistical Analysis

The impact of delivery methods on knowledge and interpretation of their genomic risk and associations to categorical participant characteristics was assessed using χ² tests. These tests were used to analyze associations between categorical variables. Logistic regression was used to model the association between comprehension and the covariates of interest. Since only one participant was found to be of below adequate health literacy, this variable was not included in any further analysis. Change in agreement between patient responses to questions of risk was assessed using Kappa statistics and McNemar’s test. Change in perceived risk after testing (1-week follow-up) was evaluated using Wilcoxon signed rank test and Spearman correlation coefficient. Two-sided p values are reported for all tests using a Type I error level of 0.05.

Results

Participant Characteristics

Three hundred individuals were enrolled in the study; 257 (86%) and 239 (80%) participants completed the 1-week and 3-month follow-up survey, respectively (table 1). Enrolled participants were primarily female (70%), White (60%), between 18 and 29 years of age (44%), college-educated (65%) and reported a family history of T2DM (70%) [24]. Participants were randomized to receive their results in-person from a board-certified genetic counselor (n = 151) or to receive their results through the testing laboratory’s secure website (n = 149).

Of the 257 participants who completed the 1-week follow-up survey, 115 (45%) participants were found to be at increased genomic risk, 118 (46%) at decreased, and 24 (9%) same as general population risk. The percentage of participants with increased genomic risk did not differ among those who did or did not report a family history of T2DM (p = 0.99).

Table 2. Participant responses to questions about comprehension of their genomic and overall lifetime risk at the 1-week follow-up assessment (n=257)

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct response, n (%)</th>
<th>Incorrect response, n (%)</th>
<th>Unsure response, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Please indicate the genetic risk number you received for developing diabetes (answer choices: open response/unsure)</td>
<td>151 (59)</td>
<td>30 (12)</td>
<td>76 (30)</td>
</tr>
<tr>
<td>Please indicate your percent chance to develop diabetes in your lifetime (answer choices: open response/unsure)</td>
<td>152 (59)</td>
<td>55 (21)</td>
<td>50 (19)</td>
</tr>
<tr>
<td>Please indicate your genetic risk for diabetes (answer choices: increased/decreased/same as population/don’t know or unsure)</td>
<td>152 (59)</td>
<td>88 (34)</td>
<td>17 (7)</td>
</tr>
</tbody>
</table>

Impact of Delivery Method on Comprehension

One week after receipt of their test results, we asked participants to indicate their genomic risk and overall lifetime risk as reported on their test report. In addition, participants were asked to interpret their genomic risk (increased, decreased or same as population). For each question, 59% of participants correctly indicated their risk (table 2). Thirty-eight percent of participants correctly indicated their genomic risk score and overall lifetime risk and correctly interpreted their genomic risk.

At 1-week follow-up, we observed that a significantly greater proportion of participants who received the results in-person correctly interpreted their genomic risk (72%) compared to online (47%, p = 0.0002; table 3). However, the difference in correct reporting of overall lifetime risk between the study arms did not reach statistical significance (p = 0.154).

Participants who received their results in-person from the genetic counselor were again more likely to correctly interpret or report their genomic risk compared to those receiving their results online at 3 months, but, like the 1-week follow-up, no significant difference was observed between study arms with respect to correctly reporting overall lifetime risk score (table 3). We also analyzed retention-of-risk information between 1 week and 3 months and found that participants’ likelihood to correctly report their genomic risk score and actual overall lifetime risk score decreased significantly (p < 0.001); no difference was observed with respect to retention of categorical genomic risk between 1 week and 3 months (p = 0.17) (data not shown). No significant difference was observed in the proportion of participants who accurately recalled either risk score or genomic risk interpretation between 1 week and 3 months between the study arms (in-person vs. online).
Impact of Genetic Knowledge and Other Factors on Comprehension of Test Results

As described previously, participants’ knowledge of genetics ranged from 50 to 100% (mean = 84%; median = 88%) [24]. At 1-week follow-up, a one SD increase in genetic knowledge resulted in an estimated OR of comprehension (overall lifetime risk) of 1.3. (table 4).

Participants with a higher overall lifetime risk were observed to have a lower overall odds of correctly indicating their risk; using a logistic regression model, a 10-unit increase in overall lifetime risk results in a 0.73 decreased odds of correctly indicating overall lifetime risk (p = 0.02). Comprehension was significantly associated with the level of education for all risk comprehension questions (p < 0.001 for genomic risk score and categorical risk; p = 0.009 for overall lifetime risk). Risk comprehension (genomic or overall) and risk interpretation were not associated with age.

When comparing delivery methods to risk comprehension, the level of genetic knowledge did not impact comprehension. However, for participants with higher levels of educational status, their interpretation of risk and reporting of actual scores decreases in those who reviewed the results in-person with the genetic counselor (interaction p = 0.02 and 0.04, respectively).

Risk Perceptions

We asked participants to indicate their perceived risk of T2DM before and after receipt of their test results. At baseline, an equal proportion of participants (37% each) believed they were at either low or intermediate risk for developing T2DM in their lifetime, and 20% felt they were at high risk. Among those who perceived themselves at high risk, 27% reported a family history of T2DM in contrast to 4% reporting no family history (p < 0.001). No significant differences in perceived risk were observed at baseline with respect to race (White vs. non-White) or age. Women were more likely to perceive themselves at higher risk than men (p = 0.01) as well as participants with lower education level (p = 0.03).

At 1-week follow-up, a significant number of participants’ perceived risk increased (p = 0.019). The change in risk assessment was correlated to the degree of actual risk (p = 0.0068), indicating improved concordance between

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**Table 3.** Comparison of delivery models and level of correct interpretation and reporting of risk scores at the 1-week and 3-month follow-up

<table>
<thead>
<tr>
<th>Study arm</th>
<th>Correctly interpreting categorical genomic risk</th>
<th>Correctly reporting genomic risk</th>
<th>Correctly reporting overall lifetime risk</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1 week</td>
<td>3 months</td>
<td>1 week</td>
</tr>
<tr>
<td>In-person</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Online</td>
<td>72%*</td>
<td>71%*</td>
<td>69%**</td>
</tr>
<tr>
<td>Online</td>
<td>47%</td>
<td>45%</td>
<td>49%</td>
</tr>
</tbody>
</table>

* p < 0.001, ** p < 0.01. a Nonsignificant.

**Table 4.** Logistic regression analysis of select participant factors and risk comprehension

<table>
<thead>
<tr>
<th></th>
<th>Correct response genomic risk score</th>
<th>Correct response overall lifetime risk</th>
<th>Correct interpretation of categorical riska</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>OR</td>
<td>p value</td>
<td>OR</td>
</tr>
<tr>
<td>Genetics knowledge</td>
<td>1.2</td>
<td>0.18</td>
<td>1.3</td>
</tr>
<tr>
<td>Level of overall lifetime risk</td>
<td>0.80</td>
<td>0.08</td>
<td>0.73</td>
</tr>
<tr>
<td>Level of education</td>
<td>2.73</td>
<td>0.001</td>
<td>2.15</td>
</tr>
<tr>
<td>Family history</td>
<td>1.3</td>
<td>0.37</td>
<td>1.3</td>
</tr>
</tbody>
</table>

a Increased/decreased/same as population.
perceived and actual genomic risk. No significant differences in perceived risk were observed at 1-week follow-up with respect to gender, education status or age. A significant association was observed between race and perceived risk (p = 0.015); further analysis revealed that this association is driven by differences in those who perceived themselves to be at intermediate or high risk. The direction of association differed between race groups; participants who self-identified as White were more likely to indicate their risk as intermediate, and all other participants were more likely to perceive their T2DM risk as high. At 1-week follow-up, those with a family history of T2DM were no longer significantly more likely to indicate that they were at high risk (p > 0.05).

**Perceived Causes and Illness Representations of T2DM**

With respect to perceived causes of T2DM, participants were asked to consider 18 items from the IPQ-R causal beliefs subscale. At baseline, the largest proportion of respondents (57%) indicated that they strongly agreed that ‘diet or eating habits’ was a factor affecting the risk of developing T2DM, followed by their ‘own behavior’ (46%) and ‘heredity’ (38%). When asked what they considered to be the primary risk factor, 47.1% indicated genes/family history, followed by diet (28%) and lifestyle (14%). More participants with family history of T2DM selected genes/family history as a risk factor (43%), than without (29%, p = 0.0432).

At 1-week follow-up, 44% of participants indicated that the primary risk factor for them was genes/family history, followed by diet (26%) and lifestyle (19%). There was not a significant difference in the proportion of participants at increased genomic risk who indicated genes/family history as the primary cause (p = 0.5144). In addition, no statistically significant difference in IPQ-R subscale scores and risk perception between those at increased and nonincreased genomic risk for T2DM or between those with and without a family history for other factors related to illness perception was observed.

**Psychological Impact**

At 1-week follow-up, the majority of participants expressed no regret after receiving their test results (92%). Based on the distress and uncertainty subscale from the MICRA, participants exhibited very low levels of distress (range: 0–30; median: 0; mean: 2.27). Distress subscale scores were not significantly different between participants with and without increased genomic risk for T2DM (p = 0.123), but were increased in participants with higher overall lifetime risk (Spearman rho = 0.33, p < 0.0001).

Similarly, participants with and without increased genomic risk for T2DM did not show significant differences in uncertainty scores about their test results (range: 0–40; median: 3; p values = 0.22). The emotional representations subscale of the IPQ-R similarly demonstrated low levels of anxiety and depression based on T2DM results (range: 6–30; mean = 11.9). Of the 43% who had children, 54% indicated that they sometimes/often worried about the possibility of their children developing T2DM (regardless of their test result), and 17% indicated that they sometimes/often felt guilty about possibly passing on the risk of T2DM to their children.

**Discussion**

Risk communication and comprehension are challenging issues for clinicians and patients alike, affected by several physician- and patient-related factors as well as patient risk perceptions [34]. For genetic and genomic testing, information about testing and test results are often delivered in-person by trained health professionals such as genetic counselors. Over the past decade, due to an increased number of genetic and genomic tests, limited access to counselors, and the launch of direct-to-consumer testing, genetic counseling services and the delivery of genetic and genomic risk information have evolved from in-person to phone-based counseling to self-access online. While evidence shows that discussing genetic testing with a counselor in-person [35–37] or over the phone [38, 39] is effective in improving accuracy of perceived risk, the impact of online self-access to results on comprehension has not been ascertained. To our knowledge, we are the first group to assess the impact of delivery methods (in-person vs. online) of a genomic risk assessment on comprehension. Although our population was of high literacy (including numeracy), we found significant differences in comprehension associated with the mode of communication.

It may be reasonably anticipated that accessing one’s genomic risk assessment without the opportunity to discuss it with a health professional can result in inaccurate comprehension. However, even with traditional modes of in-person genetic counseling, difficulties with genetic risk comprehension have been demonstrated for several conditions [11, 36, 40–44]. We found that participants receiving their T2DM genomic risk results in-person from a genetic counselor were significantly more likely to correctly indicate their risk compared to those receiving results online. About 69% of participants who received...
Comprehension of Genomic Risk for Diabetes

Irrespective of mode of communication or type of risk, about 59% of participants correctly comprehended their risk. Although there is limited data on comprehension of genomic risks delivered online, the level of risk comprehension of all participants in our study appears to fall below other published findings. In a population-based personalized medicine study with highly educated participants, 85% correctly reported their genomic risk (received online) for at least 1 of 7 diseases [50]. Similarly, another study with a comparable study population with respect to education reported that more than 80% of the participants correctly recalled their results for 8 common conditions within 10 days of reviewing their results (mailed to them) [51]. About half of participants acknowledged reviewing the whole report, and another 38% indicated they had read most of it [51]. Linnenbringer et al. [52] reported that 64% of participants with a family history of Alzheimer disease correctly indicated their genetic risk 6-weeks after receiving their test results in-person from a genetic counselor or physician. As we did not assess or inquire how much of the online report was reviewed, our low levels of comprehension may be partly due to the fact that they did not review the entire report (we could only confirm that they had logged on to access the report). Kaphingst et al. [53] tracked participants' access and time on web pages about genomic risk assessments and showed that participants' review of the websites varied by subject matter (tendency to review more introductory material than information about diseases or genes tested), averaging less than a third of the total number of pages available. Typically, participants reviewed the first few pages of a site; however, the first page of the site viewed by our participants included genomic risk and lifetime risk.

Comparison of our study with other reports may be difficult due to differences including participant characteristics, motivation to enrollment, types of disease risks reported, and familiarity with disease. In addition, several studies have shown differences in risk comprehension depending on the type of risk and how it is presented [54–57]. For example, risks can be presented as a possibility (categorical, descriptive, non-numeric statements) and/or probability (e.g. absolute or relative risk) [58] and in various other formats (numerically, descriptive, pictorially, graphically). In our study, the test report available to all participants included 2 types of risk information presented in multiple formats. Although the strategy of using multiple formats to present the same or related results may improve comprehension [54, 55] (though not always [59]), we speculate that some participants (particularly those in the online arm) may have been overwhelmed with the amount of information presented in the test report, perhaps not recognizing that the figures and text conveyed the same information. As many genetic and genomic test results are reported as a probability, numeracy (the ability to use and understand numbers [55]) may be challenging [60–65]. If actual numerical risks are not needed or perceived to be of less value, perhaps the ‘less can be more’ concept should be adopted to minimize confusion about the results [66, 67]. Alternatively, test reports may be reorganized to present a simple one-page summary, perhaps containing possibility statements, followed by actual probabilities on subsequent pages to emphasize the take-home message.

The processing and comprehension of risk information can also be influenced by the manner in which it is received and the degree of participant involvement. Active involvement in disease risk communication portends greater likelihood for recall and behavior change as it involves ‘central processing’ of the information [11, 68]. In contrast, the receipt of test results online is a more passive form of communication and involves ‘peripheral processing,’ which is more susceptible to counter persuasion [11]. Greater involvement in the process of risk communication is linked to the greater perceived relevance of the information to the individual.

We did not observe a higher likelihood of comprehension in participants at increased risk for T2DM, suggesting that the risk did not make a substantial impression or was not considered to be a cause for concern. This finding conflicts with Gordon et al. [50], who reported that more than half of participants who correctly reported their results specifically remembered diseases for which they were found to be at elevated risk. However, other studies have reported that normal results were more likely to be accurately recalled than mutation carriers [69].

Retention of risk scores decreased significantly by 3 months, but we found no significant change in correct interpretation of genomic risk at 3 months, comporting with other study findings of higher recall of categorical
risk [50, 70, 71]. In contrast, Aktan-Collan et al. [72] reported near perfect recall one month and one year later for mutation status for hereditary non-polyposis colorectal cancer; however, individuals were uncertain what their actual risk was post-testing, particularly those who tested positive. The impact of learning of their genomic or overall lifetime risk in this population, already highly familiar with T2DM due to the high prevalence of family history, may account for the low retention rate. One study reported that T2DM was not perceived by some as a severe enough disease to warrant DNA-based testing (compared to the option of family history-based risk assessment) [73]. In contrast, of 8 conditions for which genomic risk (including 3 types of cancer) was provided to participants in another study, T2DM had the highest rate of correct risk interpretation [51].

Some evidence suggests that personalizing risk factors may increase individuals’ perceived disease risk [74–76]. At baseline, our study population was divided between low and intermediate perceived risk, perhaps reflecting some optimistic bias given the high prevalence of reported near perfect recall one month and one year later and intermediate perceived risk, perhaps reflecting some optimistic bias given the high prevalence of report-low and intermediate perceived risk, perhaps reflecting some optimistic bias given the high prevalence of report-

In conclusion, while the opportunity to access test results directly without assistance from a trained health professional may offer benefits such as convenience and privacy, it may also adversely impact comprehension as patients attempt to understand their test results on their own. Thus, discussion of results with a health professional knowledgeable or familiar with genomic testing appears to be an important part of the testing process to increase the likelihood of risk comprehension and improved health outcomes. To accommodate the anticipated increase in genomic testing, however, changes may be needed to the delivery of genetic counseling services to enable greater access since in-person sessions may not be feasible, given the limited number and location of counselors. Alternative methods for patient education, counseling and risk communication will be needed to promote individuals’ ability to understand genomic test results reviewed on their own.

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