ORIGINAL RESEARCH ARTICLE | Genetics inMedicine

Return of incidental findings in genomic medicine: measuring what patients value—development of an instrument to measure preferences for information from next-generation testing (IMPRINT)

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Purpose: Little is known about the factors that influence patients' preferences for the return of incidental findings from genome sequencing. This study identified attributes of incidental findings that were important to patients and developed a discrete-choice experiment instrument to quantify patient preferences.

Methods: An initial set of key attributes and attribute levels was developed from a literature review and in consultation with experts. The attributes' salience and communication were refined using focus group methodology ($n = 12$) and cognitive interviews ($n = 6$) with patients who had received conventional genetic testing for familial colorectal cancer or polyposis syndromes. The attributes and levels used in the hypothetical choices presented to participants were identified using validated experimental design techniques.

Results: The final discrete-choice experiment instrument incorporates the following attributes and levels: lifetime risk of disease (5, 40, 70%); disease treatability (medical, lifestyle, none); disease severity (mild, moderate, severe); carrier status (yes, no); drug response likelihood (high, moderate, none); and test cost ($250, $425, $1,000, $1,900).

Conclusion: Patient preferences for incidental genomic findings are likely influenced by a complex set of diverse attributes. Quantification of patient preferences can inform patient–provider communication by highlighting the attributes of incidental findings that matter most to patients and warrant further discussion.

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Key Words: discrete-choice experiment; incidental findings; patient preferences; whole-genome sequencing

INTRODUCTION

Next-generation genomic sequencing offers significant clinical promise, and the rapidly decreasing cost of these technologies implies that whole-exome and whole-genome sequencing may soon replace conventional genetic testing. In contrast to current methods, these comprehensive sequencing approaches are likely to generate large numbers of incidental findings (IFs).[1] These findings will be of varying clinical significance: some may be well studied and medically actionable, whereas others may not be validated or clinically relevant.[2]

Previous research on the benefit to the "end user" of genetic information—the patient—focused on the clinical utility of IFs,[3,4] with clinical utility defined as the potential for a given finding to improve health outcomes. Researchers in the social sciences, however, have long acknowledged that patients also value information that does not inform clinical management.[5–7] In genetics, the "value of knowing" is an important aspect regarding the utility of genomic technology. For example, Facio et al.[8] reported that a third of participants receiving genomic sequencing in the ClinSeq study preferred to receive genomic results on the grounds that all knowledge is positive. Another study found that lay participants felt that they—not experts—were best able to judge the utility of potential genetic information.[9]

Although several studies have measured preferences for genetic testing,[10–16] to our knowledge, no studies have been designed or conducted to quantify patient preferences for IFs from genomic sequencing. Such studies are needed to gain a comprehensive understanding of the personal utility of genome sequencing and to inform practice guidelines and other policies related to the return of results from genomic testing. Furthermore, individuals' preferences surrounding IFs can inform the development of educational materials and decision-support tools to guide patients and providers through the process of returning these findings.

Discrete-choice experiments (DCEs) are tools for quantifying patient preferences for a good or service.[17–20] DCEs are a type of conjoint analysis that includes a range of methods (ranking,

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rating, or choice-based approaches) that quantify preferences for attributes of a health-care good. DCEs are popular in health economics because of their underlying behavioral framework for modeling choice behavior. In a DCE, participants are asked to choose from a set of two or more options that describe a particular good or service using a limited set of characteristics (called attributes). Participants repeat this hypothetical choice multiple times, and for each choice, the specific values (called levels) of the characteristics are varied in a predetermined manner. For each choice, participants must make trade-offs as to the relative importance of each attribute and its level. These choices are modeled using limited dependent regression to determine which attributes drive patient preferences and quantify the marginal and aggregate values of the good or service. DCEs are therefore particularly well suited to measuring the value of multidimensional technologies such as genomic sequencing.

Construction of a DCE instrument begins by determining the attributes that are jointly most relevant to the research question and salient to the patient population. Next, plausible and relevant levels are chosen for each attribute. DCEs typically include three to seven attributes and three to four levels for each attribute.[21] Hypothetical scenarios to describe the technology are constructed using all possible combinations of the attributes and levels. Validated experimental design techniques that ensure unbiased and statistically efficient parameter estimates are used to reduce the number of hypothetical choices presented to respondents to a manageable number, typically 8–16.[18]

The aim of this study was to identify, in the context of genetic testing for colon cancer susceptibility, the attributes and levels of IFs that are most important to, and cognitively understood by, patients, and to develop a DCE instrument that will enable the quantification of patients' personal utility for IFs from next-generation sequencing technologies.

MATERIALS AND METHODS

Development of the DCE instrument (Instrument to Measure PReferences for Information from Next-generation Testing (IMPRINT)) was conducted as part of a randomized controlled trial of whole-exome sequencing as compared with usual care for patients evaluated for a possible inherited colorectal cancer or polyposis syndrome (the NEXT (New EXome Technology in) Medicine Study). The trial has several aims, including developing a framework for returning IFs in a clinical setting and measuring the clinical, patient, and economic outcomes of using whole-exome sequencing in lieu of usual-care genetic testing. The trial began enrollment in September 2012 and will accrue ~220 patients over 3 years. The institutional review boards of the University of Washington and British Columbia Cancer Agency approved all activities described in this article.

Because the DCE instrument was developed for a clinical trial setting, the development was completed within a short time line and followed a multifaceted and iterative process (**Figure 1**). The initial attributes and attribute levels were chosen based on a literature review and the aim of the study, and in consultation with experts. In the second iteration, we supplemented and refined the attributes and levels with input from two focus group sessions and two cognitive interviews. Finally,

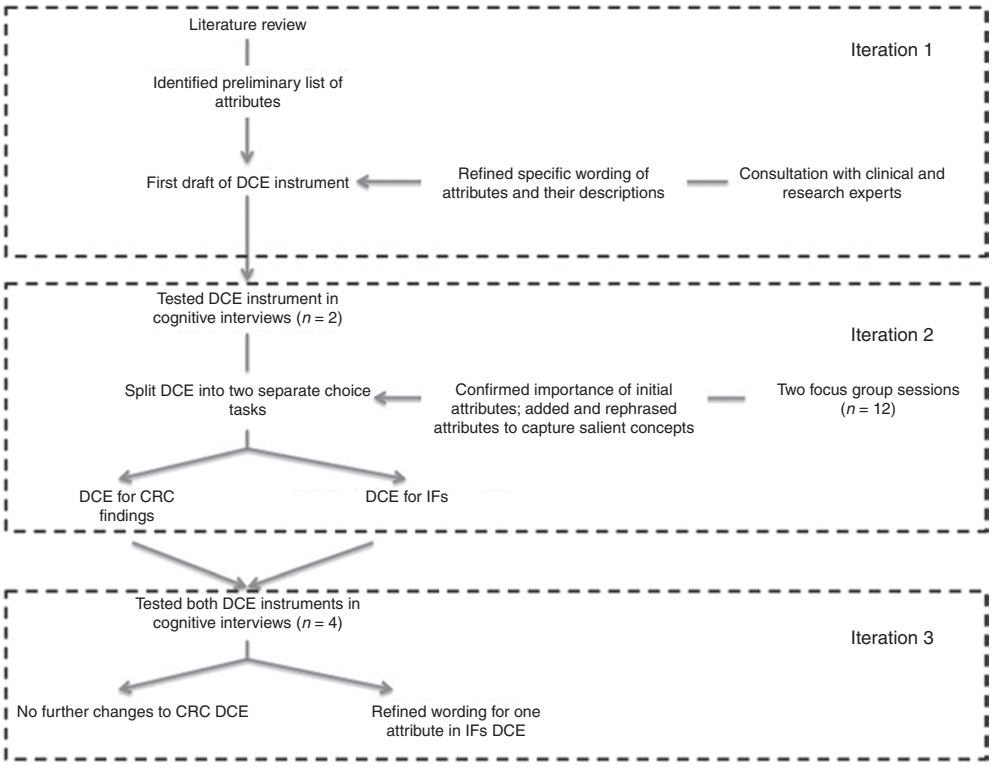


Figure 1 Flowchart of the development process. CRC, colorectal cancer; DCE, discrete-choice experiment; IF, incidental finding.

we modified the instrument and refined attribute wording and descriptions based on four additional cognitive interviews.

Literature review and consultation with experts

A literature review for English-language studies was conducted to identify previous DCEs examining the use of genomic technologies in a clinical setting.[10–16] We searched Medline for the following terms: ("genetic testing" or "genomic") and ("conjoint analysis" or "discrete choice experiment") and evaluated articles for potential factors that affected patients' preferences for genomic or genetic testing. The initial list of attributes was drafted by several members of our team (D.L.V., D.A.R., and C.S.B.) and then discussed with experts in medical and research genetics (F.M.H., G.P.J., and W.B.) regarding salience and how best to communicate the complicated concepts as meaningful and comprehensive attributes.

Focus groups

We conducted two focus groups in May 2012 with patients who underwent a clinical workup for familial colorectal cancer/polyposis syndromes at the University of Washington within the previous 24 months (see **Table 1** for demographic information); these included patients with cancer and some individuals with a strong family history of but no clinical diagnosis of cancer. Focus groups are a useful technique for gathering information from laypeople about complex topics, and observing the dynamics among participants can offer insights into the kinds of information that influence individuals' attitudes about the topic under study.[22,23] To capture a range of genetic testing experiences, we followed a purposeful sampling strategy:[24] one discussion ($n = 8$) was with patients whose genetic testing was noninformative and one ($n = 4$) included patients who had received a definitive genetic result. Recruitment response rates were 19 and 14%, respectively.

Two experienced qualitative researchers (S.B.T. and S.M.F.) led the focus group discussions using a semistructured guide to explore participants' experiences, beliefs, and attitudes about genetic testing and whole-exome sequencing. Each session lasted 2 h. The discussions were audio-recorded and transcribed. Following review of the field notes and transcripts, we determined that additional sessions were not required, as our aim was not to conduct a summative study but rather to gather formative data that could be combined with the literature review, expert input, and cognitive interviews to inform development of the DCE. We performed a close reading of the transcripts to identify features of the testing experience that mattered to patients, seeking any attributes that were salient to patients but not identified in the literature, and gaining insight into the language that the patients used.

Table 1 Participant characteristics

	Focus group 1 (noninformative result) ($n = 8$)	Focus group 2 (definitive finding) ($n = 4$)	Cognitive interviews ($n = 6$)
Age, mean (range)	54 (30–64)	61 (50–67)	55 (25–71)
Sex (*n*)			
Male	3	0	2
Female	5	4	4
Race (*n*)			
Black/African American	1	1	0
White	7	3	6
Educational attainment			
High school	2	1	3
College degree	3	1	2
Graduate degree	3	2	1
Annual income			
0–$25,000	2	1	3
$25,000–$50,000	0	1	0
$50,000–$75,000	1	0	2
>$75,000	3	1	1
Declined	2	1	0

Cognitive interviews

We conducted cognitive interviews ($n = 6$) between March and May 2012 with participants who fulfilled the eligibility criteria (**Table 1**). Cognitive interviews offer the opportunity to identify items that respondents do not understand or cannot answer, where they "get stuck," and how long it takes members of the target audience to complete the questionnaire.[25] A single member of the research team (C.S.B.) conducted the interviews, and another member (D.L.V.) observed and took detailed notes. Sessions were audio-recorded but not transcribed. In all interviews, we asked respondents to complete the choice tasks and explain their understanding of each attribute, as well as the particular trade-offs they considered when making their choice. The interview guide included both "think-aloud" and "probing" techniques; we also asked respondents to describe any additional attributes they judged important when considering genetic testing.

Construction of the DCE

We developed the DCE questionnaire from an initial set of attributes and levels identified from the literature review and expert opinion (first iteration) as well as focus groups and cognitive interviews (second and third iterations). All iterations included 16 choice tasks; each task asked participants to choose between two genetic tests described by the chosen attributes or select an "opt-out" option to accommodate those who may not wish to receive any information from genetic testing.[26,27]

RESULTS

Iteration 1: creating the initial list of attributes

We identified seven DCEs that examined individual preferences for different aspects of genetic testing in our literature review.[10–16] The factors broadly related to the domains of testing effectiveness, risk of disease, type of results returned to the patient or family member, potential consequences to the patient's family, convenience of the testing procedure, recommendation of the

doctor to undergo genetic testing, time waiting for results of the test, and cost of the test.

These factors informed the initial list of attributes for the DCE. We included attributes for the likelihood that patients received a genetic diagnosis for their colon cancer as well as the time waiting for genetic results. These attributes were chosen because a hypothesis of the NEXT Medicine Study is that whole-exome sequencing will yield a greater number of genetic diagnoses in a shorter time frame than usual care, and we wanted to evaluate how patients valued these attributes. After consultation with several genetics health-care providers and researchers, we stratified the type of "extra" information received from genomic testing into two attributes: one that addressed treatability and severity of the newly identified disease and another that described the specific health consequences for family members. Finally, we included an attribute for the total cost of the test so that we could calculate a willingness-to-pay metric for each of the attributes, which can be used to inform cost–benefit analysis.[28] **Table 2** shows the list of attributes and levels included in our initial DCE; an example choice task is shown in **Figure 2a** (see the **Supplementary Data** online for the patient education section of the DCE instrument).

Iteration 2: findings from the focus groups and cognitive interviews

We solicited information about novel themes primarily from the focus groups and information about readability and usability primarily from the cognitive interviews; however, due to

Table 2 List of all attributes and attribute levels of the initial and final iterations of the DCE

Attribute	Levels
Initial iteration	
Number of individuals who receive a genetic diagnosis related to their current disease	• 40 Individuals of 100 who are tested receive a genetic diagnosis • 60 Individuals of every 100 who are tested receive a genetic diagnosis • 80 Individuals of 100 who are tested receive a genetic diagnosis • 95 Individuals of every 100 who are tested receive a genetic diagnosis
Type of "incidental findings"	• Your risk for diseases that are readily treatable • Your risk for treatable diseases AND your risk for diseases with moderate health consequences but that are not treatable • Your risk for treatable diseases AND your risk for diseases with severe health consequences but that are not treatable
Family impact of "incidental findings"	• Risk of passing on to children genetically linked diseases that can be treated or prevented, allowing for normal child development • Risk of passing on to children diseases that have effective treatments, allowing normal child development • Risk of passing on to children all diseases, including nontreatable conditions, and child will be limited by the disease
Time waiting for results of the test	• 3 Weeks • 1 Month • 3 Months • 4 Months
Total cost to you	• $1,500 • $2,500 • $3,000 • $4,000
Final iteration (DCE for incidental findings)	
You receive information on diseases that have the following lifetime risk or higher	• Diseases with a 5% lifetime risk or higher • Diseases with a 40% lifetime risk or higher • Diseases with a 70% lifetime risk or higher
Treatability of the newly identified disease(s)	• No effective medical treatment or lifestyle change recommended • Recommended effective medical treatment only • Recommended effective lifestyle change only
Health consequences of the newly identified disease(s)	• Moderate health consequences • Severe health consequences
Carrier status for a gene not affecting you, but that may affect family members' health	• Information on if your family members could be affected • Does not provide information on carrier status
Information on your likely response to medications you may or may not be currently taking	• No information on drug response • List of medications that are moderately likely to be more effective or cause side effects • List of medications that are highly likely to be more effective or cause side effects
Cost to you not covered by insurance	• $250 • $425 • $1,000 • $1,900

DCE, discrete-choice experiment.

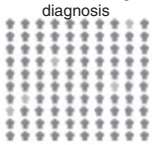
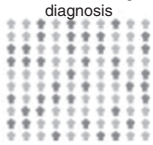
the tight time line under which the DCE was developed, these efforts overlapped, and the qualitative findings are therefore presented thematically rather than chronologically.

The cognitive interview and focus group participants exhibited a range of qualitative preferences for information from genomic testing. Some participants expressed a desire to know all genomic information ("I'd want to know everything. I'd want no sugar coating at all"), whereas others preferred to receive findings dependent on certain factors ("I think if I could be treated I would want to know, but if it's something that they may not be able to treat or if it's something that they can't guarantee that I'm going to get or the percentage is like 50/50, then I have to just live wondering about this"), and still others expressed a general apprehension about receiving genomic information ("I just think you could go nuts treating all these little possibilities. I mean it just seems like there would be no end to, I don't know, trying to research what you should be eating or not eating for this condition. I would go crazy"). Despite the wide range of preferences for IFs, several specific attributes of the test or its results emerged as important for patients considering genomic testing, specifically treatability and severity, family impact, and lifetime risk of the incidentally identified disease.

The participants confirmed that both treatability and severity of the IFs were important but indicated that they regarded

a

Choice question 1: Which genetic test do you prefer?

Test characteristic	Genetic test A	Genetic test B	No genetic testing
Number of Individuals that receive a genetic diagnosis related to their current disease	95 Individuals out of every 100 who are tested receive a genetic diagnosis 	40 Individuals out of every 100 who are tested receive a genetic diagnosis 	You will neither receive a genetic test, nor a conclusive genetic diagnosis
Type of "incidental findings"	Your risk for all diseases that are treatable	Your risk for treatable diseases and your risk for diseases with moderate health consequences but are not treatable	Risk of genetically linked diseases is based only on your family history
Family impact of "incidental findings"	Risk of passing on to children treatable diseases but child will be limited by the disease	Risk of passing on to children all diseases, including nontreatable conditions, and child will be limited by the disease	No information
Time waiting for results of the test	3 Weeks	1 Month	Not relevant
Total cost to you	$1,500	$2,500	$0
Which genetic test do you prefer? (Check one box only)	Genetic test A ☐	Genetic test B ☐	Neither test ☐

b

Choice question 1: Which genetic test do you prefer?

Test characteristic	Genetic test A	Genetic test B	No genetic testing
Number of individuals tested who receive a definitive genetic diagnosis causing CRC	60 Individuals out of every 100 who are tested receive a genetic diagnosis 	40 Individuals out of every 100 who are tested receive a genetic diagnosis 	You will neither receive a genetic test, nor a conclusive genetic diagnosis
Number of genetic tests you will undergo. *Each genetic test will require a clinic visit and a blood draw*	1 Genetic test	5 Genetic tests	Not relevant
Total time waiting for results of all genetic tests	6 Weeks	3 Months	Not relevant
Total cost to you of all testing	$2,500	$1,900	$0
Which genetic test do you prefer? (Check one box only)	Genetic test A ☐	Genetic test B ☐	Neither test ☐

How certain are you of your choice?

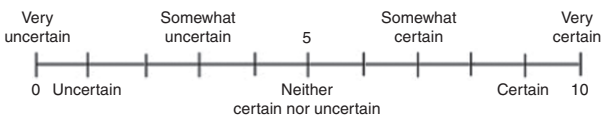


Figure 2 Example of choice task from (**a**) the initial draft of the discrete-choice experiment and (**b**) the final draft of the discrete-choice experiment for colorectal cancer (CRC) results.

these as distinct concepts. Whereas some participants wanted to know about IFs that were treatable ("Because particularly in those areas where if lifestyle changes or treatment are available, then even with a small risk, I'd want to know because I'd want to implement those lifestyle changes and consider the treatments available"), others wanted information that would have important health consequences ("Essentially how it's going to affect my independence and how much pain I'm going to have").

Participants also confirmed that family impact was important but that they considered it to be an inherent and inevitable aspect of genetic testing. For some participants, the possible implications for family members were a strong reason to initially pursue testing ("I felt like I needed to know [my genetic results] as a parent, but I also felt like I needed to know for my children"). Carrier status testing emerged as an important and distinct type of IF, although again participants exhibited a range of qualitative preferences for such information. One focus group participant thought he would use the information to make reproductive decisions ("If we knew that there was a likelihood that there would be a bad [reproductive] outcome, then that would maybe prompt me to push more towards like "Okay, we should research other options for having children""), whereas another worried that such information would burden and complicate such decisions ("I think the really hard part would be getting it before you conceive and feeling an obligation to take it into account when deciding whether or not to have children").

The lifetime risk of developing the IF disease emerged as a salient concept in both focus groups and cognitive interviews. Some participants stated that they simply wanted to know the information ("I want to know if it's 10% or 90%. I think that's important information"), whereas others stated that this risk would be an important factor in determining what IFs they would want to receive ("I think I'd probably only want to know if [the risk] was over 50%").

The two cognitive interviews using the first draft of the DCE questionnaire indicated that participants found the initial DCE instrument difficult to understand. Specifically, the respondents struggled to distinguish the attributes related to familial colorectal cancer/polyposis syndromes and IFs when presented together in one choice task. For example, one respondent expressed confusion at the initial combination of attributes: "I guess I am a little confused then. Because this test is much more inclusive [for IFs], why is it going to have much less positive results [for colorectal cancer]?" The second participant stated that she preferred to evaluate attributes relevant to her cancer diagnosis independently of the IFs: "I think it would actually be good to have a [colorectal cancer–]specific one, and okay, here's all the other stuff. I think that would be helpful."

Conclusions from iteration 2 and changes to the DCE

We made several changes to the DCE instrument based on feedback from the focus groups and initial cognitive interviews (**Figure 2a**). The most significant change was to split the DCE into separate instruments for colorectal cancer genetic results and IFs. The development of the DCE for colorectal cancer results is similar to previous DCEs that have been developed for genetic testing and was complete by the end of iteration 2.[10] It included attributes for (i) chance the test will find a gene that caused your cancer, (ii) number of genetic tests, (iii) time waiting for the results, and (iv) total cost (**Figure 2b**). We did not include an attribute to reflect the doctor's recommendation regarding genetic testing because all participants in the NEXT Medicine Trial are referred for genetic testing. The development of a DCE for the return of IFs was more complex and required several additional changes to incorporate the findings from our qualitative work.

First, we reiterated the family impact of genetic testing in the background section (see **Supplementary Data** online) and rephrased the attribute to be specific to carrier testing. Second, we split treatability and severity of disease into separate attributes. Third, we added lifetime risk of developing the IF as an attribute. Finally, we added an attribute to describe sequencing results that could provide information on participants' likely response to medications, in part because this attribute emerged in our literature reviews and qualitative work, and also because a decision was made to offer such information to participants in the NEXT Medicine Study (based on expert recommendation).

Iteration 3: confirming relevance and understanding by the target population

With the exception of lifetime risk of disease, participants in the four additional cognitive interviews interpreted the attributes similarly and in the manner intended by our research team. Several respondents understood the "lifetime risk of disease" attribute as a measure of test accuracy, taking higher values (i.e., tests that returned only IFs with a relatively high probability of developing the disease) as representing a "better" test. For example, one respondent interpreted an 80% lifetime risk as: "80% of the time they're going to show me other possibilities with this test." To clarify the construct, we rephrased the attribute from "Your identified risk of developing a disease you currently do not have" to the text shown in the example choice task (**Figure 3**). The revised instrument presents lifetime risk as a threshold, below which one would not receive results of incidentally identified diseases, and emphasizes that lower thresholds would provide more results.

Our final DCE therefore included the following attributes: disease risk, disease treatability, disease severity, carrier status, drug response, and total cost (see **Table 2** for attribute levels and the **Supplementary Data** online for descriptions of each attribute and level). In addition, respondents' certainty of choice was included after each question to examine how the complexity of each choice task affects the statistical efficiency of the parameter estimates. An example choice task from the final DCE is shown in **Figure 3**. Individuals completed the instrument within ~10–30 min, and the Flesch–Kincaid grade level of the final instrument was 7.0, indicating that an average seventh grader should understand the text.

Question 1

	Option A	Option B	No information
Disease risk You receive information on diseases that have the following lifetime risk or higher. *Note that more disease will be identified if the lifetime risk is lower.*	Diseases with a 40% lifetime risk or higher	Diseases with a 70% lifetime risk or higher	No information
Disease treatability Treatability of the newly identified disease(s)	Recommended effective medical treatment only	Recommended effective lifestyle change only	Not relevant
Disease severity Health consequences of the newly identified disease(s)	Mild health consequences	Moderate health consequences	Not relevant
Carrier status Carrier status for a gene not affecting you, but that may affect family members health	Information on if your family members could be affected	Does not provide information on carrier status	No information
Drug response Information on your likely response to medications you may or may not be currently taking	No information on drug response	List of medications that are highly likely to be more effective or cause side effects	No information
Cost Personal cost to you not covered by insurance	$1,000	$425	$0
Which test would you prefer? (Check one box)	**Genetic test A** ☐	**Genetic test B** ☐	**Neither test** ☐

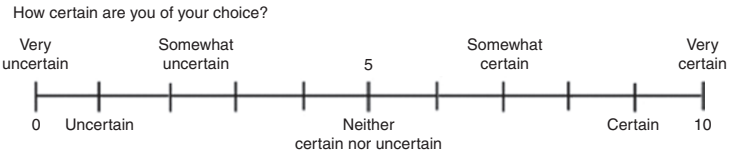


Figure 3 Example of choice task from the final draft of the discrete-choice experiment for incidental findings (IFs).

DISCUSSION

Despite a wide range of qualitative preferences for IFs, we identified six key attributes that encapsulate the most salient patient preferences relevant to our study aims: lifetime risk, treatability, severity, carrier status, drug response, and total cost. We also found that patients who stated a preference for receiving genomic information often wanted to know the results even in the absence of clinical utility.

Implications

The existence of heterogeneous preferences,[10,29] particularly across multiple dimensions, highlights the need for practical approaches to elicit these preferences in clinical practice to facilitate shared decision making. Some researchers have suggested binning IFs into categories based on their clinical utility and validity.[30] A recent study evaluated attitudes toward genomic information based on treatability and found that intentions to receive information were only mildly influenced by the treatability of the findings.[8] In our focus groups, some participants wished to know any type of genomic information if the risk of developing the disease was sufficiently high, whereas others wished to know if the incidentally identified disease was life threatening and treatable, regardless of the lifetime risk. These findings suggest that approaches to classifying IFs based on a single dimension, such as treatability or clinical utility, may not adequately reflect the totality and nuance of patient preferences for IFs.

Comparison to similar studies

Previous studies have assessed patient attitudes and qualitative preferences for return of genomic findings in both research and clinical settings.[8,9,31–35] Although there is a wide body of literature concerning the ethical issues surrounding return of IFs from research studies and the preferences of individuals to know this information,[31–35] the preferences of patients for the return of genomic results in the context of clinical care are less well studied.

Two recent studies assessed preferences for the return of results in a clinical setting.[8,9] Townsend et al.[9] solicited attitudes toward receiving IFs from health-care providers, the general public, and parents whose children received genetic testing. Lay participants felt strongly about their right to choose the types of IFs they received. Participants with a preference for knowing some, but not all, IFs favored the idea of categories based on clinical relevance but felt that relevance was a subjective concept that should not be determined by their physician. Our work elaborates on these findings by identifying the categories patients might use to classify which IFs they wish to receive.

Facio et al.[8] assessed attitudes toward learning results from whole-genome sequencing in a sample of participants from ClinSeq, a research study focused on individuals at familial risk of cardiovascular disease. Nearly all participants expressed a desire to know genomic information. By contrast, several participants in our study expressed apprehension about receiving IFs. These differences are likely explained by the different patient samples. The ClinSeq study had the explicit goal of sequencing

genomes and returning information from that analysis;[36] thus, patients were likely to have a stronger than average desire to know genomic information.

Similarly to Facio et al.,[8] we found that among those who did express an interest in knowing genomic information, a primary reason was a confidence in their ability to use the information to prevent future disease, even in the absence of clear medical utility. A challenge going forward therefore will be for healthcare providers to manage these expectations and help patients interpret and understand the inevitable findings of uncertain significance.

Limitations

Our study has several important limitations. First, we conducted a limited number of interviews and focus groups, all of which were with patients who received genetic testing for colorectal cancer or polyposis. We acknowledge that not all possible attributes were likely captured by our multifaceted approach; however, our intended goal was not complete saturation of relevant concepts but rather a preliminary (and pragmatic) assessment of the most important attributes that drive patient preferences in order to build a DCE instrument. We hope that future work builds on ours and, in particular, evaluates how the relevance of these and/or other attributes varies in other populations. Because all participants in our interviews and focus groups had received genetic counseling and testing, they may be more interested in the type of information that could be obtained from genomic sequencing and have better knowledge than the general public on issues surrounding genetic testing. However, in the study by Townsend et al.,[9] the participants from the public and parents of children who received genetic testing exhibited very similar qualitative preferences toward genomic sequencing and IFs. Moreover, we did observe a wide range of preferences and attitudes toward IFs, with some participants wishing to know "everything" and others exhibiting strong reservations about learning IFs.

Second, we acknowledge that the brief descriptions of attributes and levels will likely not fully capture the breadth and complexity of attitudes underlying individual preferences. Again, our findings suggest that a more comprehensive exploration of attitudes is warranted. However, our qualitative work was sufficient for the purposes of reducing the key concepts relevant to genomic testing into a set of attributes that could be used in a stated preference DCE that was not cognitively burdensome to respondents.

Conclusion

In conclusion, this study describes the development of a DCE instrument to measure patient preferences for information from next-generation genomic sequencing and to quantify patients' personal utility. We found that patients exhibited a range of attitudes toward genomic testing and the return of IFs. Despite these heterogeneous attitudes, we were able to identify several key attributes that were consistently associated with patient preferences and views on genomic testing, allowing us to develop a DCE instrument for future studies. The results of the DCE could be used to inform patient–provider dialogue, perhaps by prompting discussions about attributes that would otherwise have been ignored. The results could also be used to design educational materials to improve patient–provider communication and shared decision making. Implementation of a DCE in clinical practice may be challenging because of time constraints and complexity, but such approaches warrant further study. Finally, our findings highlight the need for further research on patient preferences in genomic sequencing and for effective ways of helping patients and providers understand and address these preferences in the return of IFs.

SUPPLEMENTARY MATERIAL

Supplementary material is linked to the online version of the paper at http://www.nature.com/gim

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DISCLOSURE

The authors declare no conflict of interest.

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