ARTICLE | Genetics inMedicine

Research participants' preferences for receiving genetic risk information: a discrete choice experiment

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Purpose: This study aims to determine research participants' preferences for receiving genetic risk information when participating in a scientific study that uses genome sequencing.

Methods: A discrete choice experiment questionnaire was sent to 650 research participants (response rate 60.5%). Four attributes were selected for the questionnaire: type of disease, disease penetrance probability, preventive opportunity, and effectiveness of the preventive measure. Panel mixed logit models were used to determine attribute level estimates and the heterogeneity in preferences. Relative importance of the attribute and the predicted uptake for different information scenarios were calculated from the estimates. In addition, this study estimates predicted uptake for receiving genetic risk information in different scenarios.

Results: All characteristics influenced research participants' willingness to receive genetic risk information. The most important characteristic was the effectiveness of the preventive opportunity. Predicted uptake ranged between 28% and 98% depending on what preventive opportunities and levels of effectiveness were presented.

Conclusion: Information about an effective preventive measure was most important for participants. They valued that attribute twice as much as the other attributes. Therefore, when there is an effective preventive measure, risk communication can be less concerned with the magnitude of the probability of developing disease.

Genetics in Medicine (2019) 21:2381–2389; https://doi.org/10.1038/s41436-019-0511-4

Keywords: secondary findings; incidental findings; genetic risk information; research participants; preferences for genetic risk information

INTRODUCTION

New genomics technologies, such as next-generation sequencing (NGS) and other high-throughput investigations, generate large quantities of data in research. This includes potentially unexpected genetic risk information that goes beyond the scope of the original research aims, also known as secondary, incidental, or unsolicited findings. A relevant argument for disclosure of such risk information is that it promotes participants' health. For several diseases, receiving genetic risk information in time can motivate preventive measures that delay, mitigate, or avoid future disease. This is also in line with participants' preferences.[1–5] However, participants express mixed feelings toward receiving such information. Some individuals participate in research studies for the sake of research and do not want to be bothered or worried with additional personal risk information. Receiving such information might lead to feelings of fear, anxiety, and depression.[6–9]

Several studies have investigated individuals' preferences for receiving findings from genetic testing by asking people to rate different aspects of genetic risk information one at a time.[1–4,10,11] Allen et al.[4] showed that preferences for disclosure varied depending on availability of treatment, level of disease risk, and seriousness of the disease. For example, the percentage of the participants who wanted to know their genetic test information decreased from 90% to 64% for diseases where no treatment is available. However, we lack knowledge of the relative importance of these aspects. Asking respondents to weigh different aspects of genetic risk information against each other would be more informative because risk information is complex and contains different features such as the probability of getting the disease, disease severity and expressivity, disease occurrence, and reproductive impact.

A better understanding of research participants' preferences for genetic risk information can guide principal investigators toward more sophisticated approaches regarding disclosure.

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Submitted 8 August 2018; accepted: 25 March 2019
Published online: 17 April 2019

It is also of clinical interest to understand research participants' preferences because they may end up as patients seeking counseling on the basis of the information they receive. In summary, there are a number of aspects or characteristics to consider when deciding about receiving genetic risk information. Because we wanted to identify how individuals balance the characteristics of various aspects of genetic risk information, we used a discrete choice experiment (DCE). DCE allows us to investigate research participants' trade-offs between different characteristics of genetic risk information and estimate predicted uptake for receiving genetic risk information in different scenarios. We hypothesized that more than one of the attributes contribute to peoples' willingness to receive genetic risk information.

MATERIALS AND METHODS

Discrete choice experiment (DCE)

DCEs are increasingly being used to determine individuals' preferences regarding different characteristics of interventions or medical treatments. Hypothetical alternatives regarding characteristics of genetic risk information can be described by attributes and their levels.[12] These levels can be characterized as nominal, ordinal, interval, or ratio. Hypothetical choice sets of two alternatives are designed by varying the levels of the attributes in the different choice sets in a systematic way. Respondents are provided with a series of such choice tasks and asked to choose the alternative they prefer the most within each choice task and the preferences for the characteristics are deduced from the choices.

DCE development

To construct the DCE used for this study, attributes of genetic risk information were identified in a three-step procedure. First, a literature search was performed to find possible characteristics that influence respondents' willingness to receive genetic risk information.[1–5,10,11,13–17] Second, after mapping possible attributes from the literature, a focus group with four experts (i.e., specialists in clinical genetics) was formed to preselect plausible and relevant attributes. The focus group met twice. During the first meeting, they shared their perception of important aspects in disclosing genetic risk information. During the second meeting, a hypothetical DCE was presented, and the experts commented on relevant attributes and levels. The experts agreed that the following aspects are important in disclosure of genetic risk information: likelihood of getting the disease, accuracy of the test, association between gene variant and disease, seriousness of the disease, opportunity for genetic counseling during the disclosure situation, and availability of actionable/preventive measure or follow-up program. Third, four focus group interviews ($n = 16$) with members of the target population were performed. Research participants' own understanding of genetic risk was investigated.[18] After that, nominal group technique (NGT)[19] was used to let participants rank the different aspects that they stated as important in receiving risk information. During this second part, participants were asked to rank the potential attributes from most to least important, and then discuss them in the group. Based on the total mean score of all ten attributes, four attributes were finally selected for the DCE (Table 1): type of disease (4 levels), disease penetrance probability (3 levels), preventive opportunities (4 levels), and effectiveness of the preventive measure (5 levels).

The attributes and levels were discussed thoroughly between authors. When the participants in the qualitative work talked about what a serious disease was, both quantity and quality of life were discussed; i.e., serious diseases hindering life. Therefore, we chose levels that described different types of diseases. Experts' experiences and empirical findings show that individuals' perception of risk presented in words (Low and High) vary.[20,21] We chose therefore to present the level of risk in a quantitative way with different disease penetrance probabilities. The levels were based on possible probabilities of different genetic disorders that are of clinical relevance for this particular study sample. The level Preventive opportunities was based on what participants mentioned in the qualitative work and what genetic and clinical experts considered relevant. Effectiveness of the preventive measure was important to add from a clinical perspective because preventive measures are not always efficient on an individual level. Based on that discussion, any form of out-of-pocket costs was deemed unusable (participation in any form of research study as well as general medical services are free in Sweden). Recommendations from Harrison et al.[22] were adhered to when we framed risk as an attribute, designed the opt-out option, and formed supplementary questions. To keep the number of choice sets manageable for participants, a Bayesian D-efficient design[12] was developed with the software NGene 1.0 (ChoiceMetrics, 2011). This is the most used design strategy and in line with the guidelines from the International Society for Pharmacoeconomics and Outcomes Research (ISPOR) on good research practice.[23] In the design, we introduced, for example, the restriction that the level No prevented opportunities can only occur together with the level "0% effective prevention measure". The DCE consisted of 15 unique choice tasks, each containing two alternatives. Following each choice task, participants were asked whether they would really like to know the outcomes of the genetic test in the chosen situation or not (i.e., opt-out). Before participants were asked to complete the choice tasks, they received detailed information on the meaning of all attributes and levels as well as an explanation and an example of how to complete a choice task (Table 1).

We pilot tested the draft questionnaire among a subgroup ($n = 22$) of our target population. Eight of these pilot tests were Think aloud tests.[24,25] Additionally, we tested whether correct wording was used and if the target population understood the attributes, levels, the educational information, and choice tasks. The attribute level estimates that were retrieved from the pilot study served as input for the design of the final DCE questionnaire.

Table 1 List of all attributes and levels included in the final DCE and an example of choice task

Attributes (bold text) and their levels (underlined text)		
Type of disease: The type of disease specifies the impact the disease may have on you. Genetic tests can be used to identify different types of diseases and conditions:		
Life-threatening disease (ref.): These diseases are life-threatening and you have a high probability of dying prematurely from such a disease. Treatment of the disease will therefore be crucial. Some of these diseases can be cured, but not all.		
Physical disability: This means that you become hindered or "disabled" in your everyday life (e.g., work, cleaning, family, or leisure activities) and you may experience pain and discomfort. You may need help in your everyday life. You may experience some problems with walking, washing, and dressing yourself. You might have problems with your ability to hear and/or see. Treatment for this disease mainly aims to relieve pain; otherwise, there is no cure.		
Mental disease: This means that your mental health will be affected. You may have trouble remembering, planning, and thinking in a structured way. In addition, such diseases can lead to psychiatric illness or a change in your personality. You may need help in everyday life activities. Treatments that ease these diseases are possible, but there are no treatments that will totally cure these diseases.		
Physical disease: These diseases are not life threatening and rarely lead to disability. However, these diseases deteriorate your general health condition and may affect your quality of life and need treatment. Examples of these diseases are diabetes, high blood pressure, and allergies.		
Disease penetrance probability: If the results from the genetic test show that you are at risk of a disease, it is not certain that you will get the disease. The probability of getting the disease can be calculated in several ways. One way to explain the probability is to say how many people out of every 100 will be affected:		
5 out of every 100 (ref.)		
30 out of every 100		
80 out of every 100		
Preventive opportunities: Some diseases are preventable. By taking action, you can decrease the risk of getting the disease. It does not mean that the risk disappears, but it can be decreased.		
Nothing (ref.): There is no action to reduce your risk of disease.		
Operation: You will be hospitalized for at least one day to perform the preventive surgery. It will take some time for your body to heal. The operation poses a risk for complications.		
Medication: You do not have to go to the hospital regularly. You can get the medicine at the nearest pharmacy. You may need to take the medicine for the rest of your life. It may take time to get the correct dose set. All medications carry the risk of side effects.		
Lifestyle changes: You need to change your living habits. This may mean changing your diet (change what you eat or decrease calorie intake) or stop smoking or consuming alcohol. It may also mean that you have to change your physical activity or sleeping pattern.		
Effectiveness of the preventive measure: Different preventive measures have different abilities to reduce the risk. If you take an action, it is possible to reduce the risk. The number of individuals who do not get the disease is calculated based on how many got the disease in the first place. For example, if 30 out of 100 people are likely to get the disease and all of them take the preventive measure, with an effectiveness of 50%, 15 of 30 will not get the disease:		
0% (ref.)		
25%		
50%		
75%		
90%		
Imagine that you are getting genetic risk information from participating in the SCAPIS research program. In which situation would you prefer to receive information, situation 1 or situation 2?		
	Situation 1	**Situation 2**
Type of disease	Life-threatening disease	Physical disability
Disease penetrance probability	30 out of every 100	80 out of every 100
Preventive opportunities	Operation	Medication
Effectiveness of preventive measure	25%	90%
Tick the box of the situation that you prefer:	☐	☐
If you had the possibility to receive the information you preferred above, would you want to be told?		
☐ Yes		
☐ No		

DCE Discrete Choice Experiment, *SCAPIS* Swedish CArdioPulmonary bioImage Study.

Questionnaire

The final questionnaire consisted of 15 choice tasks. Additional demographic questions were asked regarding age, gender, education level, and health literacy. Health literacy was assessed by the Swedish Communicative and Critical Health Literacy Scale (S-CCHL scale).[26] Finally, questions were added regarding

estimated health, earlier experience with genetic tests, experience with severe disease in the family, worries of being affected by a severe disease, and attitudes toward genetic risk information. The survey was web-based and constructed in Sawtooth Software SSI Web 8.4.8.

Study population

The respondents are research participants from a large research program named SCAPIS (Swedish CArdioPulmonary bioImage Study). The SCAPIS research program is a population study involving extensive measurements of 30,000 Swedes aged between 50 and 64 years. One of the aims of the project is to find risk markers that can predict who is at risk of cardiopulmonary disease and therefore can use the predictor to take prevention measures.[27] The participants get an extensive check-up regarding their health. The SCAPIS study is a collaboration between six university hospitals in Sweden, and our study only recruited participants from one of the university hospitals, Sahlgenska in Gothenburg, where 6265 participants were recruited from a random sample from the general public aged between 50 and 64 years. For the current study, 650 participants consecutively recruited in SCAPIS during 2015–2016 were asked for their preferences toward genetic risk information. Informed consent was obtained from all subjects. The Regional Ethical Review Board of Gothenburg approved the study (Dnr: 610–16).

Statistical analysis

Descriptive statistics

Descriptive statistics were used to summarize all variables of interest. Age was reported as mean (± standard deviation). Health literacy was measured using the S-CCHL scale, which consists of five items assessing different aspects of health literacy (HL), each with five response categories: never, seldom, sometimes, often, and always. An overall level of HL was calculated for each respondent.[28] Individuals responding with Strongly disagree or Disagree to one of the items were categorized as having Inadequate HL. Individuals responding with Neither agree nor disagree to one of the items were categorized as having Problematic HL. Finally, individuals responding Agree or Strongly agree to all the items were categorized as having Sufficient HL.

Being worried about having a severe disease was measured on a five-item Likert scale describing worry as Every day, More than once a week, but less then every day, More than once a month, but less than once a week, Less than once a month, and Never. Participants who expressed worry of being affected by a severe disease Every day or More than once a week, but less than every day were categorized as Very worried. The result from the questions about participants' attitudes toward genetic risk information was categorized into two groups from the five-item Likert scale (Strongly disagree, Disagree, Neither agree nor disagree, Agree, and Strongly agree) and summarized using frequencies (percentages). Persons responding Agree or Strongly agree were categorized into Agreed (Table 2).

Table 2 Demographic characteristics of the sample and percentage of participants that agreed with different genetic risk information–related statements

		No. (%) of the study cohort ($n = 351$)
Mean age (SD)		58.8 (4.3)
Gender	Male	42.5%
	Female	57.3%
	Other	0.3%
Highest educational level	Primary school	10.8%
	High school	30.2%
	University or higher education	59.0%
Health literacy	Inadequate	17.1%
	Problematic	47.3%
	Sufficient	34.8%
General health status	Poor	2.0%
	Average	14.5%
	Good	83.5%
Earlier experience of genetic tests	Yes	2.3%
Prior experience of a severe disease requiring extensive treatment	Yes	42.7%
Worry of being affected by a severe disease		3.2%
Every day	Very worried	3.2%
More than once a week, but less than every day		13.3%
More than once a month, but less than once a week	Not worried	21.4%
Less than once a month		44.5%
Never		17.6%
Statements		
If I know my genetic risk, I can prevent serious disease		61.3%
I am not sure it would help me to know my genetic risk		17.9%
I think my family would be helped if I know my genetic risk		49.3%
Knowing my genetic risk is meaningless		4.3%
Knowing my genetic risk would make me live my life differently		52.4%
Knowing my genetic risk will worry me		27.1%
I will learn more about myself if I know my genetic risk		66.1%
Knowing my genetic risk would scare me		18.5%
If it was possible to get genetic risk information, I would like to get it		66.1%

Preferences for receiving genetic test results

All results were considered statistically significant when $p < 0.05$. Data were analyzed using panel mixed logit (p-MIXL) models to account for preference heterogeneity and to adjust for the multilevel structure of the data with the econometric software Nlogit 5.0. Respondents who had missing answers to more than 10% of the 15 choice tasks were excluded from the analysis. All attributes were tested for linearity. Nonlinear attributes were recoded using effect codes.[29] The reference

Table 3 Research participants' preferences for receiving findings based on a panel mixed logit model

Basic model	Estimate	SE	SD	SE
Constant ASC	−2.77[a]	0.30	4.13[a]	0.25
Type of disease				
Life threatening (ref.)	0.83[a]	0.10	0.96[a]	0.19
Physical disease	−0.25[a]	0.06	0.39[a]	0.10
Mental disease	−0.43[a]	0.06	0.57[a]	0.11
Physical disability	−0.14[b]	0.06	0.68[a]	0.09
Disease penetrance probability				
5 out of 100 (ref.)	−0.86[a]	0.08	1.07	1.84
30 out of 100	0.07[c]	0.04	0.05	0.08
80 out of 100	0.79[a]	0.07	1.07[a]	0.07
Preventive opportunities				
None (ref.)	−1.47[a]	0.14	0.68	0.12
Operation	0.21[a]	0.07	0.31[b]	0.14
Medication	0.34[a]	0.06	0.02	0.29
Lifestyle changes	0.92[a]	0.06	0.60[a]	0.06
Effectiveness of the preventive measure	0.04[a]	0.00	0.02[a]	0.00

ASC Alternative Specific Constants, [a]$P < 0.01$, [b]$P < 0.05$, [c]$P < 0.10$.

category was coded as -1 and the sum of the Effect coded attribute levels is zero. This resulted in Equation 1:

$$\begin{aligned} U_{a,b} &= V + \varepsilon = \beta_1 \times type\ of\ disease_{disability_i} + \beta_2 \times type\ of\ disease_{mental\ disease_i} \\ &+ \beta_3 \times type\ of\ disease_{physical\ disease_i} + \beta_4 \times likelihood\ of\ disease_{30\ in\ 100_i} \\ &+ \beta_5 \times likelihood\ of\ disease_{80\ in\ 100_i} + \beta_6 \times preventive\ measure_{operation_i} \\ &+ \beta_7 \times preventive\ measure_{medication_i} + \beta_8 \times preventive\ measure_{lifestyle\ changes_i} \\ &+ \beta_9 \times effectiveness_{25\%_i} + \beta_{10} \times effectiveness_{50\%_i} + \beta_{11} \times effectiveness_{75\%_i} \\ &+ \beta_{12} \times effectiveness_{90_i} + \varepsilon \\ U_{opt-out} &= \beta_0 \times ASCC_i + \varepsilon \end{aligned} \quad (1)$$

U describes the utility of receiving genetic risk information for the respondents. V can be calculated as the observed utility that is the sum of $\beta_1 - \beta_{12}$, which are the attribute estimates that indicate the relative importance of each attribute. β_0 (Alternative Specific Constants (ASC)) represents respondents' preference for the opt-out over receiving genetic test results. The ε-term describes the unmeasured variation in respondents' preferences.[30–32] Random parameters were identified based on model fit tests (Akaike Information Criterion (AIC) and Chi-squared) and the significance of the standard deviations. By allowing attributes to be treated as random parameters, the model accounts for any heterogeneity in the preferences of the respondents concerning those attributes. For example, respondents probably differ in their preferences for which type of disease they would request genetic test results.

Relative importance of the attributes

For each attribute, a difference value was calculated by subtracting the lowest estimate from the highest estimate within one attribute. All attributes were divided with the highest difference value. This resulted in a relative distance between all other attributes and the most important attribute.

Predicted uptake

For different hypothetical genetic risk information scenarios, the predicted uptake was calculated as $1/(1+\exp^{-V})$. The standard deviation of the predicted uptake is determined by taking 10,000 draws from normal distributions with a mean and standard deviation (SD) for the particular random parameters in question (i.e., the mean and SD values were retrieved from the p-MIXL model). For every draw of the random parameters value, the predicted uptake was calculated. The average of the 10,000 calculated predicted uptakes were reported for every given genetic risk information scenario.

Influence of respondents' characteristics

We used panel mixed logit (p-MIXL) models to analyze how participants' prior experiences and worry of being at risk for a severe disease influence their willingness to know genetic risk information and how these characteristics influence the importance of the attribute Effectiveness of the preventive measure.

RESULTS

Respondents' characteristics

Of the 650 invited respondents, 393 started to complete the questionnaire. After removing the respondents with >10% missing answers on their choice tasks, 351 questionnaires (89.3%) were used in the analysis. The mean age of the respondents was 58.8 years, 57.3% were female, 59.0% reported higher education, 34.8% were observed to have sufficient health literacy, and 84.5% reported that they were in good health. Table 2 also displays that 42.7% expressed that they or a close relative had prior experience of severe disease that required extensive treatment, and 16.5% stated that they were very worried about being affected by a severe disease.

Preferences for receiving genetic risk information

All attributes showed a significant estimate. Participants preferred being informed about risk of having a life-threatening disease over other types of diseases (physical disability, mental disease, and physical disease). Their willingness to receive genetic risk information increased with increasing disease penetrance probability. Finally, respondents preferred to know about diseases for which lifestyle changes were available as preventive opportunity over other types of prevention (such as operation or medication), and their likelihood of wanting to receive genetic risk information increased with increasing effectiveness of the preventive measure (Table 3).

The negative constant shows that participants prefer to receive genetic risk information to the opt-out (constant ASC = −2.77) (Table 3). This means that participants prefer knowing their genetic risk information over not knowing it. However, the range of the standard deviation shows a large heterogeneity in respondents' desire to know genetic risk information (constant ASC, SD = ±4.13) (Table 3).

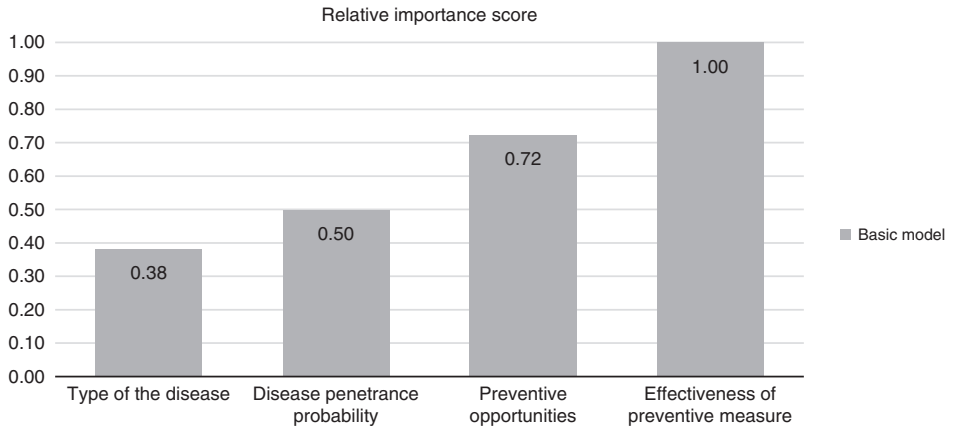

Fig. 1 Relative importance score for participants' preferences. Effectiveness of preventive measure is the most important attribute.

Table 4 Predicted uptake for different risk information scenarios, i.e., different combinations of the different characteristics of genetic risk information

	None	Operation	Medication	Lifestyle changes
Effectiveness of the preventive measure: 25%				
Life-threatening disease 5 out of 100	24%	69%	70%	76%
Life-threatening disease 30 out of 100	40%	84%	86%	91%
Life-threatening disease 80 out of 100	53%	88%	89%	93%
Effectiveness of the preventive measure: 90%				
Life-threatening disease 5 out of 100	24%	89%	90%	92%
Life-threatening disease 30 out of 100	40%	95%	96%	97%
Life-threatening disease 80 out of 100	53%	96%	97%	98%

Relative importance of different aspects of receiving genetic risk information

With respect to the relative importance of these attributes, Effectiveness of preventive measure was most important for research participants in their decision to know genetic risk information. This was followed by Preventive opportunities, Disease penetrance probability, and Type of disease. Figure 1 shows the relative importance scores of all attributes on a scale of 0–1.

Predicted uptake

The uptake probabilities for different risk information scenarios ranged from 24% to 98%, depending on what preventive opportunities are presented (Table 4). The combination that gives the highest rates (98%) is information about a life-threatening disease with penetrance probability 80 out of 100, lifestyle changes as preventive opportunity, and 90% effectiveness of the preventive measure.

Influence of respondents' characteristics

The willingness to receive genetic risk information among participants who were very worried about being affected by a severe disease or participants with prior experience of having a severe disease was not affected by the effectiveness of potential preventive measures. However, worry about a severe disease positively influenced the overall willingness to know genetic risk information (-1.59 ($p < 0.01$)). Prior experience of severe disease negatively influenced the overall willingness to know genetic risk information. Participants with prior experience were more positive about the opt-out alternative (1.37 ($p < 0.01$)).

DISCUSSION

Previous studies indicate that research participants want genomic findings to be disclosed.[1–4] Even though participants are confronted with the complexity of genetic risk information (using the DCE method), they still prefer to be informed about genetic test results to a large extent (Table 4). The majority of our participants even want to know regardless of whether there are preventive opportunities. For the combination of characteristics Life-threatening disease, 80 out of 100, and No preventive opportunity, the likelihood that our participants would like to know the information is 53%. This result is similar to earlier studies where many participants expressed a preference for findings related to high-risk disorders, regardless of possible treatment.[16,33] According to other studies, two other reasons are important to participants. They want the information because it is about them and their family, or because they want to be prepared for what is coming.[5,18,34]

This study shows that research participants' willingness to receive genetic risk information increased when the information concerned risk of a life-threatening disease, high disease penetrance probability, lifestyle changes as preventive opportunity, and high effectiveness of the preventive measure. As expected, information about risk of a life-threatening disease is preferred, a finding confirmed in other studies as well.[16,35] However, our finding that respondents prefer Lifestyle changes over Operation and Medication as a preventive measure is noteworthy because the participants might perceive information that requires lifestyle changes as more empowering than information that requires operation or medication. Information that suggests the need for an operation or medication can be associated with side effects and might therefore cause more concerns and require more consideration by the participants, whereas lifestyle changes can be perceived as having no side effects. This perception might explain why information that requires lifestyle changes is preferred. Another possible explanation is that the sample for this study is healthy research participants who participate in a research program where the aim is to find risk markers for cardiopulmonary disease. They might be more positive toward risk information in general and especially if it requires lifestyle changes, because they presumably already were thinking in such terms when they signed up for the research program.

Our DCE reveals more dynamic aspects of participants' perceptions on risk information than a questionnaire asking separate questions one at a time would be able to do. These insights into the dynamic aspects of people's decision-making processes can help researchers and policy makers to make more realistic decisions about what findings to disclose and what dimensions of the information to highlight in risk communication. We learned how participants make use of different aspects of decision-making (see Table 4). When prevention strategies are available, for example, penetrance probabilities are not so crucial for people's decision-making. The probability that participants would like to know information about life-threatening diseases that can be prevented with lifestyle changes decreased very little (from 98% to 92%) when moving from the option of information with penetrance probability of 80 out of 100 to information with a penetrance probability of 5 out of 100. However, when there is no preventive opportunity available, penetrance probabilities seem to be more important. The probability that participants would like to know information decreased from 53% to 24% when moving from the option of information with the penetrance probability of 80 out of 100 to 5 out of 100. Research participants tend to avoid the uncertainty of genetic risk information as much as possible and instead focus on what seems more certain, namely, whether preventive opportunities are available. Therefore, when there is an effective preventive measure, risk communication can be less concerned with whether the probability is high or low.

When implementing a genetic test in a clinical setting, clinical validity (the ability to predict a disease) and clinical utility (the likelihood to improve health) are considered,[36] and these concern mostly monogenic disorders.[37] This study provides a basis to believe that the clinical utility of genetic information differs partly from research participants' personal utility. Research participants in this study prefer information about risks that they can reduce through lifestyle changes (compared with no preventive opportunity, operation, and medication). Most monogenic disorders are rare and the possibilities to prevent these diseases through lifestyle modifications are often limited or unknown. However, participants may find risk information about multifactorial diseases particularly useful because it motivates them to change their lifestyle (e.g., diet or smoking behavior).[5,34] Whether the association (which is contiguous to disease penetrance probability in this study) between genetic makeup and disease is strong or not might not concern research participants as much as it concerns researchers and clinicians.

This study also investigated the interaction between willingness to receive genetic risk information and (1) prior experience of serious disease as well as (2) worries about having a severe disease. Our study indicates that previous experience of having a serious disease significantly reduces participants' willingness to receive genetic risk information. This might be because participants with prior experience of severe disease perceive that knowing their genetic risk would imply additional mental strain, which was evident in the qualitative study[18] and in the individual interviews used to prepare the DCE. Moreover, worry about having a severe disease also positively influences the overall willingness to know genetic risk information. This might be because people who worry about having a severe disease want to prevent or postpone developing the disease as much as possible.

Conclusion

Discrete choice experiments can help researchers and policy makers to make more realistic decisions about what findings to disclose and what dimensions of the information to highlight. In this study, effectiveness of the preventive measure was most important for participants. They valued that attribute twice as much as the other attributes. Thus, when there is an effective preventive measure, risk communication can be less concerned with the magnitude of the probability of developing disease. Worry about having a severe disease had a positive influence on the willingness to receive genetic risk data, whereas previous experience of severe disease had a negative influence. Further studies are warranted to understand in more detail how these aspects affect the way people reason about genetic risk information.

Limitations

These results were in line with the expected direction of the estimates (e.g., participants prefer level 80 out of 100 over 5 out of 100) and therefore provide support for the theoretical validity of the model. However, there might be a hypothetical bias as in all discrete choice experiments. Because participants are not bound by their hypothetical choices, there may be differences between what they say they will choose and what they actually choose.

This is the first DCE study on genetic risk information in a Nordic country and it indicates the preferences of this particular group (healthy, middle-aged research participants that undergo an extensive health check-up) for genetic risk information. The results of this study may not be generalizable to people in their reproductive phase of life. Concerns about healthy offspring probably is of such importance to this group that they can be expected to reason differently about genetic risk information. It would be interesting to include this group in future DCE studies, including recessive genetic diseases as an attribute.

The response rate (60.5%) is good compared with previous studies on this topic.[4,16,17] It is likely that the people we approached are interested in the subject of our survey because they were already participating in the SCAPIS study, where feedback from research is a relevant issue.

While it is interesting to note that respondents preferred to know about diseases for which lifestyle changes were available as preventive opportunity, we acknowledge that lifestyle changes are not known to have any major impact on the majority of genetic diseases. However, recommendations on lifestyle changes are important for a few severe and potentially life-threatening genetic diseases, e.g., smoking cessation in α-1-antitrypsin deficiency, exercise restrictions in inherited cardiomyopathies, and dietary modifications in inherited metabolic diseases.

ACKNOWLEDGEMENTS

This work has been supported by a grant to the project Mind the Risk from the Swedish Foundation for Humanities and Social Sciences (grant number PR2013–0123); the European Union Seventh Framework Programme (FP7/2007–2013) under grant agreement number 305444 (RD-Connect); the Innovative Medicines Initiative project BTCure (grant agreement number 115142–1); the BioBanking and Molecular Resource Infrastructure of Sweden project, BBMRI.se (financed by the Swedish Research Council); Euro-TEAM; BiobankCloud; and Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) LPC (313010). We acknowledge support from SCAPIS funded by the Swedish Heart–Lung Foundation for providing access to their cohort.

DISCLOSURE

The authors declare no conflicts of interest.

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