HHS Public Access

Author manuscript

*J Genet Couns*. Author manuscript; available in PMC 2021 February 01.

Published in final edited form as:

*J Genet Couns*. 2020 February ; 29(1): 131–134. doi:10.1002/jgc4.1186.

# Comparing preferences for return of genome sequencing results assessed with rating and ranking items

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**Keywords**

breast cancer; genetic counseling; genome sequencing; patient; preferences; return of results; secondary findings

## 1 | INTRODUCTION

Genome sequencing is being used in research and clinical settings for identification of genetic variations (Biesecker & Green, 2014). Because sequencing technologies may yield secondary findings, which are genetic variations unrelated to the indication for sequencing, the American College of Medical Genetics and Genomics recommended the evaluation and reporting of secondary findings based on its minimum gene list (Blackburn et al., 2015; Kalia et al., 2017). This list currently contains 59 medically actionable genes and will be updated based on new research findings (Kalia et al., 2017). To inform policy regarding return of secondary findings further, physicians and researchers have explored the preferences of patients for the return of different types of sequencing results. Existing evidence suggests that patients most often prefer to receive results for medically actionable genes, although they may also want to receive other types of results (Kaphingst et al., 2016).

Among surveys designed to assess preference, rating is usually preferred over ranking due to its convenient implementation (Harzing et al., 2009). Rating scale questions use Likert response options to assess each individual item, this is an absolute assessment; while a

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AUTHOR CONTRIBUTION

Substantial contributions to the conception and design of the work; acquisition, analysis, and interpretation of data for the work: SG, MG; KK. Drafting the work or revising it critically for important intellectual content: SG, MG, KK. Final approval of the version to be published: SG, MG, KK. Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved: SG, MG, KK.

Conflict of interest

The authors have no conflict of interest to declare.

Human studies and informed consent

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

Animal studies

No non-human animal studies were carried out by the authors of this article.

ranking scale asks participants to rate their preference for each item compared to the others, this is a relative measure (Hein, Jaeger, Tom Carr, & Delahunty, 2008). Absolute scales provide information about preferences for each item, however, when several of the ratings are similar, relative scales provide information about the preferred importance of each item (Mccarty & Shrum, 2000). Here, the two methods are compared with respect to determining preferences for return of results from genome sequencing.

In a previous study using a dataset generated by a survey of 1,080 women diagnosed with breast cancer at a young age, researchers explored the association between psychological and clinical predictors and preference for return of seven different types of genome sequencing results as assessed with Likert Scales as a rating method for level of interest in return (Kaphingst et al., 2018). The results suggested that psychological factors were related to the preferences; participants with more knowledge about sequencing benefits, higher worry about genetic risks, and stronger health information orientation were more likely to be very interested in learning each type of result while clinical factors largely were not related to these preferences (Kaphingst et al., 2018). Here, we examine the association between psychological and clinical predictors and preferences for return of results using the ranking of the seven types of sequencing results and compare similarities and differences to the previous analysis using the rating scale (Hong, Biesecker, Ivanovich, Goodman, & Kaphingst, 2019; Kaphingst et al., 2018).

The study population was a nationwide cohort (Young Women's Breast Cancer Program [YWBCP]) of women diagnosed with breast cancer at age 40 or younger. YWBCP participants were surveyed between June and December 2014. Among contacted individuals ($n$ = 1,778), 1,080 (61%) women completed the survey. Only participants with non-missing responses for the ranking question were included ($n$ = 1,045; 97%). All participants reviewed a consent information sheet and gave consent to participate. Primary data collection was approved by the Human Research Protection Office at Washington University in St. Louis. This secondary analysis study was approved by the New York University Institutional Review Board, University Committee on Activities Involving Human Subjects.

*Ranked Preference* was the outcome variable, indicating participants' interest in learning different types of results that might be generated using genome sequencing, which was assessed by asking them to rank their interest in order from the most (marked as 1) to the least interested (marked as 7). There were seven types of possible results: 1) increase individual's risk for preventable or treatable diseases, 2) increase individual's risk for unpreventable or untreatable diseases, 3) affect individual's response to medication, 4) affect children's health, 5) affect health of other relatives, 6) have uncertain meaning (VUS), and 7) are unrelated to health (e.g., ancestry, physical traits). We examined the top choice (rank 1) for each participant and classified it into three groups: Actionable (types 1 or 3), Carrier (types 4 or 5), Non-actionable (types 2, 6, or 7).

We included psychological, clinical and demographic predictors in our model. For psychological predictors, we had *Genetic Causal Beliefs* (The degree to which respondents believed that breast cancer was caused by genes), assessed using one item answered on a five-point scale (Weinman, Petrie, Moss-Morris, & Horne, 1996). Answers were

dichotomized into ‘Low causal belief’ (‘1-Not at all’; ‘2-A little’; ‘3-Somewhat’) and ‘High causal belief’ (‘4-Mostly’; ‘5-Completely’) for analysis. We assessed two sub-scales: *Health Consciousness* (five items) and *Health Information Orientation* (eight items) (Dutta-Bergman, 2003). The average scores for each sub-scale were modeled as continuous variables. *Worry about genetic risk*, we assessed genetic worry with the average score from three items (Biesecker et al., 2009). *Worry about cancer*, we assessed worry related to cancer with three items (Gotay & Pagano, 2007). The average score was included as a continuous variable. *Genome Sequencing Knowledge*, we used two sub-scales: knowledge about sequencing benefits and knowledge about sequencing limitations (Kaphingst et al., 2012). In the analysis, average sub-scale scores were included as two continuous variables. *Decision-making preference*, we utilized two items adapted from the Control Preferences Scale (Lillie et al., 2007). We categorized the responses into passive decision-making (‘1-I prefer to leave the decision to my doctor’; ‘2-I prefer my doctor makes the decision, but seriously considers my opinion’); shared decision-making (‘3-I prefer my doctor and I share responsibility for deciding’); and active decision-making (‘4-I prefer to make my own decision after seriously considering my doctor’s opinion’; ‘5-I prefer to make my own decision regardless of my doctor’s opinion’). *Family communication* responses were assessed using two items inquiring having shared genetic test results with family members, which were categorized as ‘0-did not share/not sure’; ‘1-shared with relatives’; ‘2- spouse/partner/other/not specified’ (Elrick et al., 2017).

For clinical predictors, we had *Prior Genetic testing*. We combined the responses to ‘Have you ever received genetic testing as part of your clinical care?’ and ‘Which of these types of clinical tests have you received? Check all that apply.’ into ‘Did not have testing/not sure/ other genetic test’ and ‘Had *BRCA1/2* genetic testing.’

Finally, for demographic characteristics, we included *Having biological children* (yes/no), and *education* (Graduate degree or more/College or less), which were included as dichotomous variables; *Employment status* (Full-time/Part-time/Other) was a categorical variable.

Multinomial logistic regression analysis was used to examine relationships between predictors and the top choice category (actionable/carrier/non-actionable). Psychological variables were tested for entry into the multivariable model if they were statistically significant in bivariate analyses. The final model contained psychological variables that remained significant in the multivariable model, controlling for clinical (prior genetic testing) and sociodemographic variables (age, biological children, education and employment). We conducted the analyses using StataSE version 15 (StataCorp.). Statistical significance was assessed as $p < .05$.

Participant characteristics are shown in Table 1. In Table 2, we examined associations between predictors and top choice in the ranking. Controlling for education attainment, employment status, and age, compared to women without biological children, women with biological children were significantly more likely to rank learning about carrier status results as their top choice (RRR = 9.084 [3.56–23.18]).

Compared to people with lower genetic causal beliefs, people with higher genetic causal beliefs were more likely to rank learning non-actionable results as their top choice over actionable or carrier status results (RRR = 0.415 [0.21–0.81]; 0.388 [0.18–0.85]).

We found differences in the results based on rating scale (absolute) with those of the ranking scale (relative). Using the Likert scale rating system, participants were able to select all types of genetic results as 'very interested', whereas the ranking system forced participants to state a preference order for genetic results. In the rating system analysis, knowledge about sequencing benefits, worry about genetic risks, and health information orientation were found to be significant predictors of interest in learning all types of genome sequencing results. In this analysis, these variables were not significantly associated with the ranked outcome, suggesting that variables affecting being very interested in return of a particular result are different from those affecting prioritization of results.

The results presented here suggest that, among these women diagnosed with breast cancer at a young age, patients with biological children most valued carrier status results as secondary findings, even more than secondary findings of actionable results related to their own health. This is somewhat consistent with previous findings using the interest rating scale, which showed that having biological children was significantly associated with strong interest in learning at least one type of sequencing result (Kaphingst et al., 2018).

In another analysis using rating scale data from this survey, high genetic causal belief was found to be related to strong interest in learning genetic risk information for results associated with preventable and treatable diseases (Hong et al., 2019). In this analysis with the ranking scale, we found that participants with higher genetic causal beliefs were more likely to rank non-actionable results as their top choice for return than either actionable or carrier status results. Taken together, these results indicate that patients with strong genetic causal beliefs may be interested in return of a broad range of possible secondary findings.

These findings should be considered in light of several study limitations. The study population included breast cancer patients who were diagnosed at age 40 or younger, and thus, findings are not generalizable to other patient populations. Moreover, the majority of respondents were Caucasian (92%) with high educational attainment (46% graduate degree), which resulted in limited variability of some covariates (Yu, Crouch, Jamal, Bamshad, & Tabor, 2014). Lastly, though the mandatory order of ranking provided us information on preference for options other than the most preferred, we assessed the preference using the top choice.

This study showed that ranking and rating scales give different and complementary information about patients' preferences for return of secondary findings generated using genome sequencing. In designing methods of determining patient preferences, genetic counselors may want to consider using both types of items to give a complete picture of preferences. We also found that having biological children was related to having a preference for actionable information, and having higher genetic causal beliefs for breast cancer was related to prioritizing information other than actionable results. Future studies

should use both ranking and rating measurements to examine factors associated with preferences for genetic information in other contexts and populations.

## ACKNOWLEDGMENTS

The authors thank the women who agreed to participate in the study and the research staff. This work was supported by the National Cancer Institute, National Institutes of Health (R01CA168608).

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## TABLE 1

Characteristics of participants in analytic sample ($n$ = 1,025)

| **Continuous variables** | **Mean(*SD*)** | **Range** |
|---|---|---|
| Age | 45.8(9.0) | 26–78 |
| **Categorical variables** | ***N*** | **Percent(%)** |
| [a]Top choice (Dependent variable) | | |
| Actionable | 822 | 80.2 |
| Carrier | 104 | 10.1 |
| Non-actionable | 39 | 3.8 |
| Prior genetic testing | | |
| Did not have testing/not sure/had other genetic testing | 149 | 14.5 |
| Had BRCA1/BRCA2 testing | 876 | 85.5 |
| Educational attainment | | |
| College degree or less | 559 | 54.5 |
| Graduate level | 466 | 45.5 |
| Employment status | | |
| Full-time | 601 | 58.6 |
| Part-time | 199 | 19.4 |
| Other | 225 | 22.0 |
| Having biological children | | |
| No biological children | 323 | 31.5 |
| Have biological children | 702 | 68.5 |
| Race/ethnicity | | |
| Black/multiracial | 13 | 1.2 |
| White, non-Hispanic | 945 | 92.2 |
| Hispanic | 31 | 3.1 |
| Other | 36 | 3.5 |

Abbreviation: *SD*, standard deviation.

[a]Types of genetic variants: associated with (1) preventable/treatable diseases; (2) non-preventable/non-treatable diseases; (3) response to medication; (4) affect children; (5) affect relatives; (6) had uncertain meaning; (7) not related to health.

## TABLE 2

Multinomial logistic regression model to predict choosing actionable, carrier, non-actionable genetic results as top choice (Reference: Non-Actionable; $n$ = 1,025)

| | Actionable | | Carrier | |
|---|---|---|---|---|
| **Reference: non-actionable** | **RRR** | **95% CI** | **RRR** | **95% CI** |
| [a]Genetic causal belief | **0.415** | 0.21–0.81 | **0.388** | 0.18–0.85 |
| [a]Prior genetic testing | | | | |
| Had BRCA1/BRCA2 testing | 1.012 | 0.36–2.86 | 0.586 | 0.17–2.09 |
| [a]Having biological children | | | | |
| Have biological children | 0.816 | 0.26–2.52 | **9.084** | 3.56–23.18 |
| [a]Education attainment | | | | |
| Graduate degree or higher | 0.689 | 0.90–3.63 | 0.696 | 0.32–1.49 |
| [a]Employment status | | | | |
| Part-time | 0.816 | 0.32–1.95 | 0.757 | 0.27–2.09 |
| Other | 0.640 | 0.23–1.45 | 0.453 | 0.17–1.19 |
| Age | 1.026 | 0.98–1.07 | 0.987 | 0.94–1.04 |

Abbreviations: CI, confidence interval; RRR, relative risk ratio; Bold coefficients indicate significance ($p < .05$).

Reference: Actionable: ‘preventable/treatable diseases’ or ‘response to medication’ as top choice; Carrier: ‘affect children’ or ‘affect relatives’ as top choice; Non-actionable: ‘non-preventable/non-treatable diseases’ or ‘had uncertain meaning’ or ‘not related to health’ as top choice.

[a]Causal belief: Low genetic causal belief (REF); Prior genetic testing: Did not have testing/not sure/had other genetic testing (REF); Having biological children: No biological children (REF); Education attainment: College or less (REF); Employment status: Full-time (REF).