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Decision Role Preferences for Return of Results from Genome Sequencing amongst Young Breast Cancer Patients

Cindy B. Matsen^{a,b}, Sarah Lyons^c, Melody S. Goodman^d, Barbara B. Biesecker^e, and Kimberly A. Kaphingst^{a,f}

^aHuntsman Cancer Institute at the University of Utah, Salt Lake City, UT, USA 84112

^bDepartment of Surgery, University of Utah, Salt Lake City, UT, USA, 84132

^cDivision of Public Health Sciences, Washington University in St. Louis, St. Louis. MO, USA 63110

^dDepartment of Biostatistics, College of Global Public Health, New York University, New York, NY, USA, 10003

^eResearch Triangle Institute, International, Washington DC, USA, 20005

^fDepartment of Communication, University of Utah, Salt Lake City, UT, 84112

Abstract

Objective: To better understand decision role preferences in women diagnosed with breast cancer at a young age for return of results of genome sequencing in research and clinical settings.

Methods: Participants were surveyed about communication and decision-making preferences related to genome sequencing results and factors that may affect these preferences. The primary outcome was decision role preference (Control Preference Scale) for selecting what results to receive within medical care or within a research study.

Results: For results returned as part of medical care, most patients preferred a collaborative (N=481, 45%) or active (N=488, 45%) role with only 107 (10%) choosing a passive role. When making the decision as part of a research study, most patients preferred an active role (N=617, 57%), 350 (33%) choosing a collaborative role, and 110 (10%) choosing a passive role.

Conclusion: Most women in this study preferred to share in decision making. Participants had somewhat different role preferences for clinical and research contexts, with greater preference for active roles in the research context.

Corresponding author at: Cindy B. Matsen, MD, MSCI, FACS, Huntsman Cancer Institute at the University of Utah, Department of Surgery, 1950 Circle of Hope, Suite N6405, Salt Lake City, UT, USA 84112, cindy.matsen@hsc.utah.edu, Tel: 801-581-2304, Fax: 801-585-3749.

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CONFLICTS OF INTEREST

The authors have no conflicts of interest to declare.

Practice Implications: We advocate for practice guidelines that incorporate discussion of decision role as an integral part of patient centered care and shared decision-making and recognize that more work is needed to inform guidelines.

Keywords

breast cancer; BRCA; genetic knowledge; advanced genetic testing; decision making; decision role preferences

1. INTRODUCTION

Genome sequencing is on the rise in both the clinical and research settings [1, 2], since rapidly improving technology has decreased the costs and time needed. This has been an important step towards personalized medicine. This test provides vast amounts of information, but there are scientific limitations to interpreting gene variants for clinical use. As such, it is uncertain how to best use the data and how to communicate results to patients, even in the research setting [3–5]. There are many types of information that can result from the use of sequencing technologies including information about: risk of preventable disease, risk of non-preventable disease, risk of future disease for relatives, and efficacy of medications to name a few [1, 5–7]. Clinical sequencing holds the promise to provide much needed, clinically relevant information. However, what is currently clinically relevant is dwarfed by information with potential personal utility for patients, but marginal or no clinical utility. The resultant ethical and legal implications of these tests can be difficult for patients and providers to consider since the results may affect not just the individual, but the entire family [8, 9]. Because this is a difficult situation to navigate, it is important to understand the role(s) patients want to play in decisions about results to be returned.

In making this type of decision, patients may prefer different levels of participation, with some preferring to make the decision themselves, some preferring to have the provider make the decision, and some wanting to share the responsibility for the decision with the provider [10–15]. Shared decision-making is defined as a collaborative process in which the patient and provider each contribute to the final decision. It is advocated as the most patient-centered decision making process and decision role preference is a key part of the process [16–19]. When it comes to decision preferences for returning test results generated by sequencing technologies, there is very little known about how patients want to engage in the process and what factors might be associated with these role preferences. Studies in other clinical decision-making contexts have shown that the role a patient wants to play in the decision is associated with multiple demographic factors with younger patients and more educated patients preferring to take on more active roles where they take primary responsibility for the decision. Role preference, however, is likely dynamic, and depends on contextual variables such as type of disease, age, clinic setting, gender, and dominant cultural values [10, 13, 20–32]. With genome sequencing becoming more common, there are many opportunities for understanding how patients want to make decisions about testing and how they want the information communicated back to them (i.e. return of results). Specific disease contexts, such as breast cancer, where single gene testing has long been available, but more comprehensive genetic testing such as genome sequencing is new, provide rich

resources for understanding these processes. Because sequencing results may generate personalized data related to multiple health conditions, patient decisional preferences for receiving these results may differ from their preferences for receiving results from disease specific genetic tests.

Breast cancer is the most commonly diagnosed malignancy in women of all ages [33]. Women diagnosed at a young age (40 or younger) are recommended to undergo genetic testing [34]. These women are more likely than women over 40 to carry genetic mutations that put them at increased risk for developing breast cancer. While the best known and highest risk of these mutations are in the BRCA genes, the list of gene mutations for which to test is ever evolving and increasing. Genetic testing for high risk mutations at the time of a breast cancer diagnosis can have clear impacts on treatment choice whereas the clinical usefulness of other types of information generated by genome sequencing is much less clear [1, 2, 6, 7].

Decisions about what results patients would like to receive and how to communicate the results can be even murkier in the research setting where the usefulness of the results to an individual is less immediate [35, 36]. Breast cancer patients may have greater comfort with genetic testing and its implications, but it is not clear that this familiarity with genetic testing leads to a desire for more active participation in decisions to receive results from less familiar testing technologies such as genome sequencing. While prior opinion surveys of the general public have indicated that most individuals want as much information as possible, this may not apply to actual cancer patients [36]. To better understand how a population for whom sequencing is increasingly relevant might approach this issue, we surveyed women who had been diagnosed with breast cancer at or younger than age 40 about their preferred role in return of results decisions about genome sequencing and evaluated what factors might be associated with their preferred role. Since the context of the decision may matter, we looked at role preference in decisions for return of results from sequencing in a research or a clinical setting.

2. METHODS

2.1 Study Participants

Description of the study participants has been provided in a prior publication [37]. Study participants were recruited from an existing nationwide cohort (Young Women's Breast Cancer Program; YWBCP; <https://siteman.wustl.edu/ywbcp>) comprised of women diagnosed with breast cancer at the age of 40 or younger who had agreed to be contacted about other studies. Before any contact, we searched the Social Security Death Index and removed those individuals who were deceased. We mailed letters to 1,778 YWBCP members inviting them to participate in a survey online, by telephone, or by mail, followed by an email invitation that contained a link to the online survey. We sent two follow-up emails with links to the survey to those who did not opt-out of further contact, followed by a mailed paper version of the survey. Sixteen participants without an email address were mailed a paper survey at baseline and follow up. Each participant completed a single survey. All participants reviewed a consent information sheet or were read the information by phone and gave consent to participate. Participants received a \$10 gift card in recognition of their

time spent. The study was approved by the Human Research Protections Office at Washington University in St. Louis.

2.2 Survey Procedures

Participants were surveyed on their communication and decision-making preferences related to genome sequencing results and factors that might affect these preferences (e.g., genetics-related knowledge, self-efficacy, worry, and beliefs; cancer-related worry; health information seeking and orientation; health consciousness; numeracy; clinical and sociodemographic characteristics). These factors were selected based on prior research linking them to clinical treatment decisions and decision role preference, and expert opinion.

2.3 Measures

The primary outcome of this analysis was decision role preference for selecting what results to receive from genome sequencing as part of medical care or as part of a research study. Decision role preference was measured using the Control Preferences Scale which asks the participant to choose from one of five roles: two passive roles (provider takes primary responsibility for the decision with varying degrees of involvement from the patient), one collaborative role (the decision is shared between the two parties), and two active roles (patient takes primary responsibility for the decision with varying degrees of involvement from the provider).

We tested sociodemographic variables, clinical and disease variables, genetic testing variables, genetic self-efficacy, genetic-related beliefs, subjective numeracy, and personal health perception as possible factors affecting decision-making preferences. Sociodemographic variables included current age, biological children, brothers and sisters, family history, race and ethnicity, education level, marital status, and income. Clinical variables included age at diagnosis, having more than one cancer and other types of cancer. Variables related to genetic testing were also analyzed including having met with a genetic counselor, having had cancer genetic testing, type of testing done, and *BRCA* mutation status. The survey did not ask about the actionability of genetic changes in the context of decision role preferences. Genetic self-efficacy was measured using three Likert-type items: assessing the role of genes in health, assessing genetic risk of disease, and comfort explaining genetic issues to others [38]. Genetic-related beliefs were measured with an adaptation of the Illness Perception Questionnaire with 5 items measuring beliefs around possible genetic and environmental causes of breast cancer with higher scores indicating stronger beliefs in genetic causality (i.e., belief that genetic makeup determines whether a woman will get breast cancer) [39–41]. Subjective numeracy was measured with the eight-item Subjective Numeracy Scale [42]. Personal health perception was measured using a single item asking participants to rate their overall health as poor, fair, good, very good, or excellent.

2.4 Analysis

Descriptive statistics of characteristics for the study participants are shown in Table 1. Bivariate analyses between each of the factors and the categorical outcomes was carried out using multinomial logistic regression. Using factors that were associated in bivariate

analyses at $p < 0.1$, multivariable models were built for each of the outcomes using stepwise multinomial logistic regression, with a passive role as the reference for each outcome (preferred role for decisions related to return of results from whole genome sequencing as part of medical care, and preferred role for return of results as part of research). Statistical significance was assessed at $p < 0.05$. All analyses were conducted using SAS/STAT 9.4 (SAS Institute Inc., Cary, North Carolina).

3. RESULTS

Of those contacted, 1,080 (61%) women completed the survey. Comparing the respondents to the YWCBP cohort, we found no significant difference in age at diagnosis. The sample had a lower proportion of African American women (1.5%) than the cohort (3.8%; $p < .01$). There were no significant differences in the other racial and ethnic groups. As shown in Table 1, the majority of participants were non-Hispanic white (92%), married (78%), and had biological children (69%). Half had an annual household income of greater than \$100,000 per year and 79% had a college degree or higher. Over half considered themselves to be in very good or excellent health (58%).

The mean age at the time of their breast cancer diagnosis was 35 years old (standard deviation (SD)=4 years), with a mean of 10 years (SD=8 years) since their diagnosis. Most of the women (84%) had not had a recurrence of their cancer or a new breast cancer. The majority (77%) had received cancer genetic testing, 11% had a known deleterious mutation in the BRCA genes, and 18% had no mutation, but a strong family history of breast cancer. Most (72%) had lower genetic causal beliefs, responding “somewhat,” “a little,” or “not at all” to a causal role for genes in breast cancer. Slightly fewer women had lower causal beliefs related to family history (63%). Most scored in the middle for genetic self-efficacy (mean=2.89, SD=1.08) and had fairly high subjective numeracy (mean=4.92, SD=0.84).

When asked to identify their preferred role in decisions to have genome sequencing results returned to them as part of medical care, most patients preferred either a collaborative (N=481, 45%) or active (N=488, 45%) role with only 107 (10%) choosing a passive role. When making the decision for return of genome sequencing results as part of a research study, most patients preferred an active role (N=617, 57%), with 110 (10%) choosing a passive role, and 350 (33%) choosing one of the collaborative roles (Figure 1).

3.1 Role Preference for Return of Results as Part of Medical Care

In bivariate analysis of factors related to decision making preferences for return of genome sequencing results as part of medical care (Table 2), women who perceived their health to be very good or excellent were more likely to prefer an active (OR: 1.91, 95% CI: 1.25–2.91, $p < 0.01$) or collaborative role (OR: 1.52, 95% CI: 0.99–2.31, $p = 0.05$) when compared to a passive role. Those who had higher genetic self-efficacy (OR: 1.38, 95% CI: 1.13–1.67, $p < 0.01$), or those with increased time since diagnosis (OR: 1.04, 95% CI: 1.01–1.07, $p = 0.02$) were significantly more likely to prefer an active role compared to a passive role. Women with higher genetic causal beliefs (OR: 0.63 95% CI: 0.40–0.99, $p = 0.04$), and those who had biological children (OR: 0.57, 95% CI: 0.35–0.91, $p = 0.02$) were significantly *less* likely to prefer an active role compared with a passive role. Those with more than one

primary cancer (OR: 0.67, 95% CI: 0.47–0.97, $p=0.03$) were *less* likely to prefer a collaborative role compared with a passive role.

In multivariable analysis, factors associated with preferring an active role compared with a passive role were perceiving one's health to be very good or excellent (OR: 1.73, 95% CI: 1.11–2.71, $p=0.02$) and longer time since diagnosis (OR: 1.04, 95% CI: 1.01–1.07, $p=0.02$). Those who had biological children (OR: 0.57, 95% CI: 0.35–0.94, $p=0.03$) were *less* likely to prefer an active role than a passive one. Perceiving one's health as very good or excellent was also significantly related to preferring a collaborative role compared with a passive role (OR: 1.56, 95% CI: 1.00–2.44, $p<0.05$).

3.2 Role Preference for Return of Results as Part of a Research Study

For decision making preferences related to return of results for genome sequencing as part of a research study (see Table 3), bivariate analysis showed that women who perceived their health to be very good or excellent (OR: 2.14, 95% CI: 1.42–3.23, $p<0.01$), had higher genetic self-efficacy (OR: 1.29, 95% CI: 1.07–1.55, $p<0.01$), had an older current age (OR: 1.06, 95% CI: 1.03–1.08, $p<0.01$), were older at diagnosis (OR: 1.06, 95% CI: 1.01–1.11, $p=0.01$), had more time since diagnosis (OR: 1.07, 95% CI: 1.03–1.11, $p<0.01$), or had higher subjective numeracy (OR: 1.32, 95% CI: 1.04–1.66, $p<0.01$) were significantly more likely to prefer an active role compared with a passive role. Those who perceived their health to be very good or excellent (OR: 1.88, 95% CI: 1.22–2.90, $p<0.01$), had an older current age (OR: 1.06, 95% CI: 1.03–1.09, $p<0.01$), were older at diagnosis (OR: 1.09, 95% CI: 1.04–1.15, $p=0.01$), or had more time since diagnosis (OR: 1.07, 95% CI: 1.03–1.12, $p<0.01$) were also significantly more likely to prefer a collaborative role compared with a passive role.

In multivariable analysis, women who perceived their health to be very good or excellent (OR: 1.86, 95% CI: 1.20–2.88, $p<0.01$), were older at diagnosis (OR: 1.06, 95% CI: 1.01–1.12, $p=0.02$), had more time since diagnosis (OR: 1.06, 95% CI: 1.01–1.12, $p<0.01$), or had higher subjective numeracy (OR: 1.31, 95% CI: 1.04–1.65, $p=0.02$) were more likely to prefer an active role than a passive role. Those who perceived their health to be very good or excellent (OR: 1.67, 95% CI: 1.05–2.67, $p=0.03$), were older at diagnosis (OR: 1.09, 95% CI: 1.03–1.15, $p<0.01$), or had more time since diagnosis (OR: 1.07, 95% CI: 1.03–1.12, $p<0.01$) were also more likely to prefer a collaborative than a passive role.

4. DISCUSSION AND CONCLUSION

4.1 Discussion

This survey of a large group of women diagnosed with breast cancer at a young age provides valuable insights into the decision-making preferences of these patients for return of genome sequencing results in the clinical and research contexts. Consistent with past studies on role preferences in decision-making, most women prefer to share in the process with their providers to some degree. The results from this study show that a number of different variables were associated with these preferences [28, 43, 44].

We found that participants had somewhat different decision-making preferences for clinical and research contexts, with a greater preference for active roles in the research context. This may be due to differences in the immediate applicability of the results or differences in levels of trust and longer relationships for doctors versus researchers. Results in a research context are generally not used to make decisions about current health issues unless clinically verified, but may be viewed as potentially relevant in the future. This potential future application requires patients to project what may be important to them in the future which may impact the amount of control they want to have over their results in making future decisions. Results from research may also have more implications for the individual's relatives rather than the individual and this may also lead to differences in the preferred level of control in these decisions. Participants may want more control over return of results when they are less immediate and less useful for the individual.

In addition, different factors were associated with preferred role depending on whether the sequencing would be done as part of medical care or as part of research. The participating women's perceptions of themselves as healthy, with a good understanding of how to use and communicate about genetic information (higher genetic self-efficacy), and a good understanding of how to use numbers (greater subjective numeracy) illustrates this group of women as well-educated. This is coupled with a longer time to have processed their disease experience as shown by being diagnosed at younger ages and having more time since their diagnosis. All of these factors were associated with their decision making preference, though in different ways depending on the medical care or research context. Based on prior studies, these factors are what one might predict to be associated with a desire to be more involved in decisions to participate in research studies and the return of results that may come from their participation. Younger age and higher educational level have been associated with preference for more active roles in past studies [45–47], but this study is the first to look at perceptions of health and genetic-related variables. In most past studies of breast cancer patient role preferences, about half of patients preferred the collaborative role with the other half split between the more active and passive roles [46, 47]. In our study, preferences were skewed towards the more active roles, but this was much more striking in the research study context.

On the other hand, it is somewhat surprising that women with biological children were less likely to prefer active or collaborative roles compared with passive roles when making the decision to have results returned from genomic sequencing as part of medical care, since the results may have implications for their children's disease risks. We hypothesize that this may be due to a parental desire to know the information, but discomfort in deciding what information should be important to their children and possibly fear of what the results may predict about their future leading them to feel less inclined to actively make the decision. We do not know of any past studies that have explored how familial variables influence decision role preferences and this is an area for future work.

This study has several limitations. It is known that role preferences can vary by race and ethnicity and education level [10, 25, 26, 45, 46, 48] and our study population is limited in this regard. Our population is reflective of the breast cancer patient population in general [49] and consistent with populations in prior studies, but there is evidence that responses to genomic risk information do vary by race and ethnicity [50] and role preferences and

associations may be different if studied in a more diverse population. There may also be important differences in role preference for return of actionable versus non-actionable results. Though the larger survey did distinguish between actionable and non-actionable results, the questions specifically regarding decision role preferences did not so this cannot be assessed in this study. Also, this survey asks the participants to imagine hypothetical scenarios and it is not known if they would want the same role if they were faced with this decision in actual practice or research. Future work should address this by looking at role preferences in people who are approached for genome sequencing in clinical and research settings. It would also be interesting to reassess preferences among participants from this study who go on to receive genome sequencing to see if their preferences were stable. It is not clear if these results would translate to people making decisions about genome sequencing who are healthy or have a history of other conditions.

4.2 Conclusion

What these data very clearly show is that decision making preferences are not easily categorized and reinforce the idea that preferred role is not easily predicted based on demographic factors alone [13, 15, 25]. It is critical for providers to recognize that because someone is young (or old) or well-educated (or not) or has children (or doesn't), they can't assume what role the patient wants to play in the decision-making process in a particular context. Models of patient-centered care should incorporate questions about decision role preferences across contexts, recognizing that this is a dynamic preference that needs to be re-addressed regularly. This is not a preference that can be easily known without direct questioning. Elicitation of decision role preferences is not routinely done which contributes to discordance between patient and provider perceptions of role. Aligning perceptions and helping patients achieve their preferred role could improve care delivery in as yet undefined ways. This is also an area for future exploration.

4.3 Practice Implications

There are many types of genetic variants that can be discovered through genome sequencing. Patients prefer different roles in deciding what information they would like to receive and this does vary by the context of the testing. We advocate for practice guidelines that incorporate discussion of decision role as an integral part of engaging in patient-centered care and promoting shared decision-making. This study demonstrates that most patients do want to play a collaborative or active role in these decisions, but there is variability in role preferences for return of results. This provides important data that can be used by providers, researchers, and policy makers when developing policies and procedures for returning results from genetic tests.

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HIGHLIGHTS

- Availability and use of whole genome sequencing is expanding
- Patients prefer different roles when deciding what results to receive
- There is no consistent variable that predicts what role a person prefers

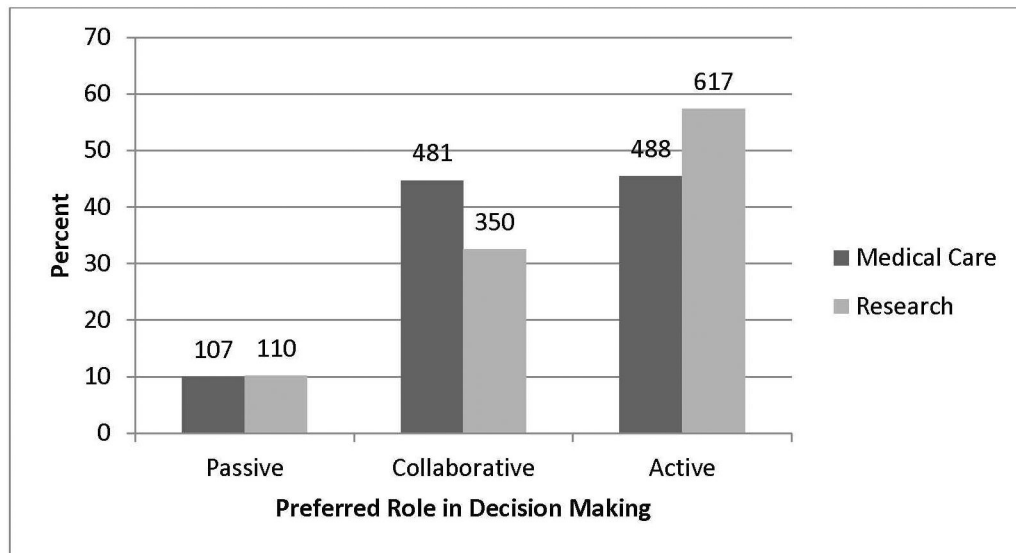


Figure 1. Preferred role of women diagnosed with breast cancer at a young age in decisions related to return of results from whole genome sequencing as part of medical care versus research.

Table 1.

Study participant characteristics.

	N	%
Genetic related beliefs		
Genetic make-up affects getting breast cancer (<i>n=1079</i>)		
High genetic causal belief*	299	27.21
Low genetic causal belief	780	72.29
Family history affects getting breast cancer (<i>n=1079</i>)		
High family history causal belief	394	36.52
Low family history causal belief	685	63.48
Received genetic testing - clinical record (<i>n=1080</i>)		
No	245	22.69
Yes	835	77.31
Family history of breast cancer (<i>n=1078</i>)		
Unknown or Low	568	52.69
Moderate	207	19.20
Strong	303	28.11
Mutation on BRCA1 or BRCA2 (<i>n=1080</i>)		
Positive	120	11.11
Negative/Variant	712	65.93
Unknown	248	22.96
Mutation / family history categories (<i>n=1080</i>)		
No mutation and strong family history	190	17.59
No mutation and low or moderate family history	525	48.61
Mutation positive	120	11.11
No genetic testing	245	22.69
Biological children (<i>n=1080</i>)		
Yes	740	68.52
No	340	31.48
Siblings related by blood (<i>n=1080</i>)		
Yes	999	92.50
No	81	7.50
Race (<i>n=1080</i>)		
Non-Hispanic White	994	92.04
Other	86	7.96
Education (<i>n=1079</i>)		
Some college or less	222	20.57
College degree	376	34.85
Beyond college	481	44.58
Marital status (<i>n=1077</i>)		
Married/Living as married	839	77.90
Widowed/Divorced/Separated	132	12.26

	N	%
Never been married	106	9.84
Annual household income (<i>n=947</i>)		
< \$100,000	468	49.42
\$100,000 and over	479	50.58
General perception of health (<i>n= 1080</i>)		
Poor/Fair/Good	458	42.41
Very good/Excellent	622	57.59
Medical professional has told that have a genetic condition (<i>n=1044</i>)		
No	843	80.75
Yes	201	19.25
More than one primary cancer (<i>n=1079</i>)		
No other cancer	907	84.06
Yes, breast cancer recurrence or second breast Cancer	113	10.47
Continuous Covariates		
	Mean (Range)	SD
	N	%
Age - current (<i>n=1080</i>)	45.93 (26–82)	9.15
Age - at diagnosis (<i>n=959</i>)	34.88(21–41)	4.17
Time since diagnosis (<i>n=959</i>)	10.43(0–57)	7.87
Genetic self-efficacy (<i>n= 1080</i>)	2.89(1.00–5.00)	1.08
Subjective numeracy scale (<i>n=1080</i>)	4.92(1.25–6.00)	0.84
Ability subscale (<i>n=1080</i>)	4.95(1.00–6.00)	1.09
Preference subscale (<i>n=1079</i>)	4.90(1.25–6.00)	0.91

* Stronger beliefs in genetic causes over environmental causes in the development of breast cancer

Table 2.

Factors associated with preferred role for decisions related to return of results from whole genome sequencing as part of medical care.

Preferred Role (passive as reference)	Active Role		Collaborative Role	
Bivariate Analysis				
	OR (95% CI)	p-value	OR (95% CI)	p-value
High genetic causal belief	0.627 (0.398 – 0.988)	0.044	0.926 (0.592–1.449)	0.737
Have more than one primary cancer	0.840 (0.592–1.193)	0.330	0.672 (0.467 – 0.966)	0.032
Biological children	0.567 (0.354–0.911)	0.019	0.902 (0.558–1.458)	0.673
General perception of health: Very good/Excellent	1.906 (1.250–2.905)	0.003	1.515 (0.995–2.307)	0.053
Genetic self-efficacy	1.375 (1.132–1.669)	0.001	1.055 (0.870–1.278)	0.588
Time since diagnosis	1.037 (1.005–1.071)	0.023	1.024 (0.992–1.057)	0.149
Multivariable analysis¹				
	OR (95% CI)	p-value	OR (95% CI)	p-value
Biological children	0.573 (0.349 – 0.940)	0.028	0.893 (0.540–1.475)	0.658
General perception of health: Very good/Excellent	1.730 (1.107–2.705)	0.016	1.563 (1.001 –2.441)	0.049
Time since diagnosis	1.039 (1.006–1.072)	0.020	1.021 (0.989–1.054)	0.209

¹Model controlling for genetic causal belief and subjective numeracy

Table 3.

Factors associated with preferred role for decisions related to return of results from whole genome sequencing as part of research.

Preferred Role (passive as reference)	Active Role	p-value	Collaborative Role	p-value
Bivariate Analysis				
	OR (95% CI)	p-value	OR (95% CI)	p-value
General perception of health: Very good/Excellent	2.141 (1.418–3.233)	<0.001	1.877 (1.216–2.897)	0.005
Genetic self-efficacy	1.285 (1.066–1.549)	0.009	1.160 (0.953–1.412)	0.138
Age	1.055 (1.027–1.084)	<0.001	1.063 (1.034–1.093)	<0.001
Age at diagnosis	1.062 (1.013–1.113)	0.013	1.092 (1.037–1.150)	<0.001
Time since diagnosis	1.072 (1.032–1.113)	<0.001	1.074 (1.034–1.117)	<0.001
Subjective numeracy scale	1.317 (1.043–1.661)	0.021	1.061 (0.834–1.351)	0.630
Multivariable Analysis^I				
	OR (95% CI)	p-value	OR (95% CI)	p-value
General perception of health: Very good/Excellent	1.858 (1.199–2.878)	0.006	1.673 (1.051–2.665)	0.030
Age at diagnosis	1.063 (1.011–1.117)	0.016	1.089 (1.032–1.149)	0.002
Time since diagnosis	1.071 (1.031–1.113)	<0.001	1.073 (1.032–1.117)	<0.001
Subjective numeracy preference subscale	1.309 (1.042–1.646)	0.021	1.095 (0.863–1.390)	0.454

^I Model controlling for genetic causal belief and biological children