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Implementation of a Patient Decision Aid to Facilitate the Selection of a Pre-Test Genetic Counseling Model

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IMPLEMENTATION OF A PATIENT DECISION AID TO FACILITATE THE SELECTION OF A PRE-TEST GENETIC COUNSELING MODEL

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Abstract

Genetic testing is being increasingly offered to patients that have a personal or family history of cancer. This has led to a higher demand of patients needing to meet with genetic counselors (GCs) for pre-test counseling and education, which has led to increased wait times for appointments. In order to ameliorate the long wait times patients now face, alternatives such as providing patient education through a series of online educational materials are now being offered. Patient decision aids (PtDAs) are tools created by a team of healthcare professionals that assist patients in making important decisions regarding their healthcare. For this study, we created a PtDA for patients at Memorial Sloan Kettering Cancer Center (MSK) to assist them in choosing between a traditional pre-test genetic counseling pathway and a novel online Direct-To-Test (DTT) pathway. Eligible participants were asked to complete a baseline survey before viewing the PtDA, and then a post-survey after using the PtDA. Participants voluntarily self-reported information, such as demographics, reason for pursuing genetic testing, anxiety levels using the GAD-2 scale, and understanding of genetics using in the baseline survey. The post-survey asked participants to report which pathway they would choose, factors that influenced their decision, decisional regret using the SURE scale, satisfaction with the PtDA using a 5-item Likert scale, and evaluation of their knowledge of genetics using the KnowGene scale. The results indicated that participants were highly satisfied with the PtDA, and had little regret over their final decision. Satisfaction with the PtDA was also found to be correlated with KnowGene scores. This study can be used as a guide for a larger investigation into the use and effectiveness of PtDAs in regards to patient preference concerning genetics education.

Introduction

There has been an uptake in genetic testing amongst individuals who are suspected to be at risk for hereditary cancer syndromes (Williams et al., 2008). Such syndromes arise due to a change, or 'mutation,' in a known gene that increases the individual's risk for developing certain cancers over their lifetime. With approximately 5-10% of all cancers caused by inherited genetic mutations, more opportunities for cancer surveillance and prevention, as well as tailored treatment for patients with active cancer, are now possible through the identification of these mutations via genetic testing (Garber & Offit, 2005). This allows affected individuals to participate in additional screening methods which can ultimately contribute to the earlier detection and treatment of cancer (Lewis, 2014).

Relatives of affected individuals are also at an increased risk for having the same mutation, and the sharing of this information amongst families has prompted an increase in genetic testing (Garber & Offit, 2005). Pursuing testing in search of a familial mutation has shown to provide relevant information for such family members and has the potential to alleviate uncertainties and anxieties (Lewis, 2014). An unaffected patient may qualify for genetic testing if a known familial mutation is found in a family member, or if there is a substantial family history of cancer (Meiser, 2005). Although testing has become increasingly available to these individuals, the decision to undergo genetic testing is not a one-size-fits-all approach (Stacey et al., 2008).

Historically, a 'paternalistic approach' has been utilized by physicians as they made decisions without consulting patients about their values or preferences concerning their healthcare (Stacey et al., 2008). This approach often left the patient in the dark about which tests were being run, including genetic testing. The decision to undergo genetic testing for hereditary

cancer syndromes is one that should be made with informed consent from the patient. Informed consent is when a healthcare provider reviews the purpose, risks, benefits, limitations, and clinical utility of the potential results that the genetic testing can yield (Lewis, 2014). Informed consent has been typically facilitated in a pre-test counseling session by a genetic counselor who is trained to educate and counsel patients about these concerns (Elwyn et al., 2000). As the genetic counselor works with the patient towards a decision, the patient's values are explored and discussed in order to serve as the basis for the final decision the patient makes (O'Connor et al., 2004). This is known as 'shared decision making,' (SDM) and it complements genetic counseling's non-directiveness by allowing genetic counselors to offer their professional assessment while respecting the patient's values in regards to making a final decision (Elwyn et al., 2000).

With the increase in demand for genetic testing services, the need for genetic counselors to provide pre-test education and participate in SDM with patients is at an all-time high. Patients are now finding themselves waiting for available appointments for several months, or even up to a year. To ameliorate these wait times and allow patients to move forward with genetic testing sooner, educational interventions have been implemented at various institutes. At Memorial Sloan Kettering Cancer Center (MSK), a new direct-to-test (DTT) option is being offered to eligible patients. Patients that elect the DTT pathway can alternatively consent to genetic testing without pre-test counseling with a genetic counselor. This DTT pathway helps streamline the process of scheduling patients with a personal or family history of a cancer diagnosis for genetic testing. While the DTT pathway is now an option provided to many patients, they can still choose to instead meet with a genetic counselor for pre-test counseling to make an individualized choice regarding genetic testing. In order to help patients discern which option will best meet

their needs, we have developed a patient decision aid (PtDA) to be used to support any preexisting knowledge, ameliorate uncertainties or anxiety, and educate patients about both options.

PtDAs are non-directive tools that are designed to help patients make informed choices about a clinical decision (Elwyn et al., 2006). They are not meant to take the place of a healthcare professional; rather, they are meant to encourage patient participation by exploring the most important values and preferences the patient has about their healthcare (O'Conner et al., 2007). One study evaluated 46 PtDAs from the 2017 Cochrane review and the use of PtDAs in a cancer setting significantly improved patient knowledge and participation during the decision-making process (McAlpine et al., 2018). This ultimately led to a decrease in decisional conflict, as well as physician-controlled decisions.

In this study, a PtDA was designed to assist patients at MSK who are considering genetic testing for hereditary cancer syndromes choose between the DTT pathway and pre-test counseling. Patients were asked to complete surveys before and after being presented with the PtDA. Their responses were used to evaluate anxiety towards genetic testing, decisional conflict, general knowledge about hereditary cancer, and satisfaction with PtDA.

History of PtDA Development

Patient decision-aids have started to become more commonplace within medical settings; however, there has not always been standardized guidance for ensuring quality measures. In 1998, the Ottawa Hospital Research Institute proposed a conceptual framework – the Ottawa Decision Support Framework - for assisting patients in decision making. The Ottawa Hospital Research Institute decided to evaluate patient decision aids with the Decisional Conflict Scale

(DCS) and found that many decision aids, both that were and were not created according to the Ottawa Decision Support Framework, increased patient knowledge while reducing decisional conflict (O'Connor, 1995 and O'Connor et al., 1999). However, it was found that in 1999 there were only 15 PtDAs that had been created by academic intuitions – meaning that they were carefully crafted and heavily cited with proper sources (Elwyn et al., 2006). By 2004, more than 500 decision aids had been created, by both nonprofit and commercial companies (Elwyn et al., 2006). Without proper international standards, it was feared that the market would become saturated with PtDAs that lacked quality and could ultimately harm patients.

In 2003, at the 2nd International Shared Decision-Making Conference, the International Patient Decision Aids Standards (IPDAS) was established with the aim to create an internationally agreed upon framework for quality criteria when creating PtDAs (Elwyn et al., 2006). Decision aids must include reputable sources and have balanced viewpoints with patients and providers in mind. The original study reviewed the perspectives of patients, providers, policyholders, and researchers from different countries using a Delphi consensus to encourage reflection and agreement which resulted in the *IPDAS Patient Decision Aid Checklist for Users* (Elwyn et al. 2006). The original publication has been used to ensure quality control of PtDAs as a supplemental tool for patient-provider encounters.

The Ottawa Hospital Research Institute currently has an accessible inventory of patient decision aids that help support decisional conflict when patients are unsure about a course of action. The structure and format of a decision aid needs to meet the specific criteria by *IPDAS* in terms of its content, development process, and effectiveness.

Methods

Study Design

The implementation of the PtDA provided the opportunity for patients to be educated on the two options and clarify patient values that are aligned with their pre-testing consenting modality which was further evaluated using quantitative methods of inquiry in the form of validated and investigator-developed surveys. This study was submitted to the MSK Institutional Review Board (IRB) and was determined to be a quality improvement project which did not require IRB oversight.

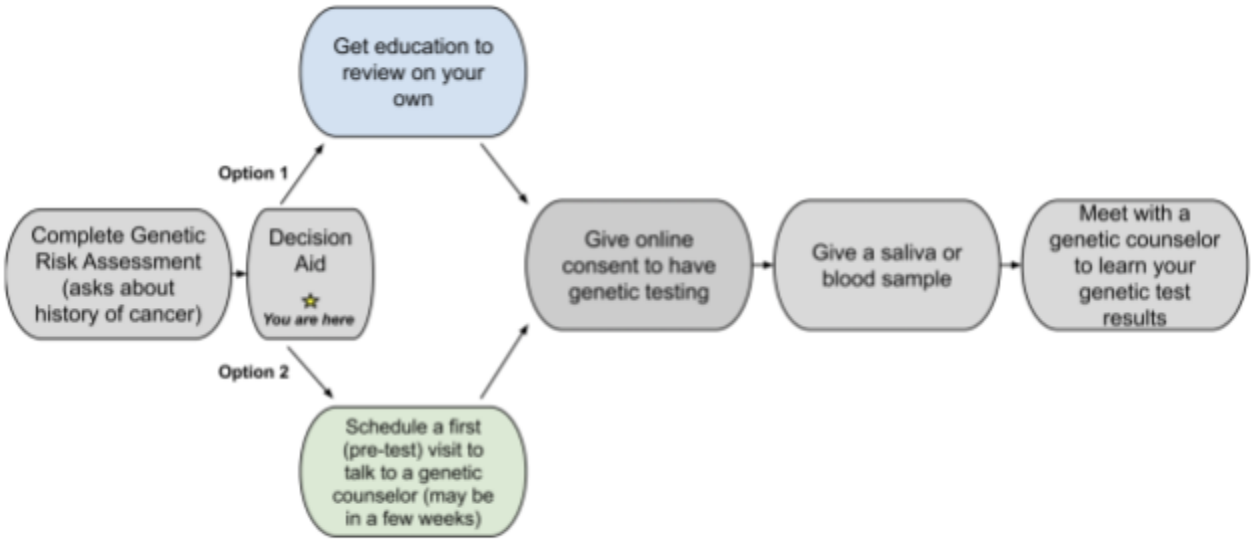

Figure 1: Patient Decision Aid Workflow

PtDA Design

A prototype of the decision aid was created after reviewing cancer-related patient decision aids that were listed on the Ottawa Hospital Research Institute's inventory. A non-profit developer, Healthwise Knowledgebase, established a patient decision aid to help clarify a participant's breast cancer risk and refine whether or not the option of testing for the *BRCA* genes is indicated. This decision aid served as a model for constructing a decision aid for MSK's pre-test consenting options. The prototype was developed using Research Electronic Data

Capture's (REDCap) features and evaluated with informal/open-ended feedback from approximately 9 professionals in the field of genetics, genetic counseling, oncology, and behavioral psychology at MSK. The decision aid content was adapted based on feedback to ensure that all pertinent information was portrayed accurately. It was implemented into clinical practice for patients who were eligible for the DTT pathway from November 2022 until April 2023.

	Option 1: Reviewing online education before genetic testing	Option 2: Meeting with a genetic counselor before genetic testing
What is the process?	Online education (fact sheet + video) will be emailed or sent via the MyMSK Patient Portal for you to review. You will also get a genetic test consent form to fill out	You will first have a telehealth (video) or in-person visit with a genetic counselor. You will then get a genetic test consent form to fill out
How long does it take?	You can review the online education at your own speed. You should review the education and fill out the genetic test consent form within 2 weeks of getting them	The genetic counseling visit usually lasts about 1 hour
Will I need an appointment?	No, you will review this information on your own	Yes, you will need to schedule a visit, which may be in a few weeks depending on availability
What are the limitations of this option?	- If questions come up, it may take a few days for the Clinical Genetics team to get back to you - You won't benefit from having a genetic counselor explain information in a way that you can best understand	- You will have to schedule a visit to meet with a genetic counselor, which may take a few weeks
What happens when genetic test results are ready?	No matter which option you choose, you will talk to a genetic counselor during a telehealth (video) or in-person visit about your results when they are ready	
Does insurance cover the cost of genetic counseling and testing?	Many insurance companies cover genetic counseling and testing at MSK but each carrier has different rules for coverage. If you have questions about your health plan or want to make sure that your health plan covers genetic counseling and testing, contact your insurance company. We can help answer questions about your insurance coverage. Please go to Frequently Asked Questions about Genetic Counseling or call MSK's insurance specialists at 646-497-9176.	

Figure 2: Differences between the pathway choices as outlined in the PtDA

Participant Recruitment

Eligible participants were invited to participate in the study between November 2022 to April 2023. Participants were eligible if they met the following criteria: were existing MSK patients, were between the ages of 25-99 years of age, and were eligible to participate in the direct-to-test (DTT) pathway. Potential participants were invited to participate by the team at MSK via email or through their patient portal (MyMSK). Consent was implied if participants completed any portion of the surveys.

Survey Methodology

All surveys were created using REDCap and were informally evaluated by 9 professionals in the field of genetics, genetic counseling, oncology, and behavioral psychology. Eligible participants were encouraged to complete two voluntary surveys, one prior to and one following the utilization of the PtDA. The first survey provided a standardized baseline assessment to report patient demographics, personal medical/family history, and anxiety levels.

Anxiety levels were assessed by using the Generalized Anxiety Disorder Scale (GAD-2) that has been implemented in the Kroenke et al. study. This validated survey consists of two statements that help measure anxiety symptoms based on the corresponding participant responses on a 4-point Likert scale. Baseline assessment of the participant's anxiety was used to evaluate whether preexisting anxiety plays a role in how participants choose to review pre-testing information.

Upon completion of viewing the PtDA, participants were then presented with the post-PtDA survey which collected information on which option they wanted to proceed with: the DTT pathway, pre-test counseling with a genetic counselor, not proceeding with genetic testing,

or, ‘Don’t know.’ They were also asked whether they would prefer to provide a saliva or blood sample. They were asked about factors influencing their decisions regarding their choice between educational pathways. Participants were asked which factors influenced their decision regarding the pathway that they chose. They were able to choose from the following: Time, Preferred Learning, GC support, Feelings, and Other. Participants were allowed to choose more than one factor. The second survey also collected information regarding decisional conflict (SURE test) and an assessment of the knowledge patients had concerning hereditary cancer (KnowGene), as well as questions designed by the study team concerning the overall satisfaction of the participant regarding the use of the PtDA.

The KnowGene scale was implemented to measure general knowledge about hereditary cancer and genetic testing methodology. In the original study (Underhill et al., 2019) the scale was cut down from a 24-item knowledge scale to a validated 16-item knowledge scale. For the purpose of our study, we further removed two items as they were less relevant to our participant population, and only used 14 items. The two items that were omitted were items 19 and 24, which stated, ‘If a person does not have a mutation found on genetic testing (negative result), interpreting results will depend on whether someone in the family has a known gene mutation associated with cancer risk (positive result),’ and ‘Multi-gene panel testing could find a mutation in a gene that is not clearly associated with the pattern of cancer in the family,’ respectively. The remaining questions were posed after the patient had interacted with the PtDA to measure their baseline understanding of cancer genetics prior to receiving either their educational interventions or meeting with a genetic counselor, and to determine if knowledge correlated with pathway choice.

Overall satisfaction with use of the PtDA was assessed on a 5-item Likert scale of investigator designed statements modeled from validated surveys. The statements included whether (1) It was easy to access the patient decision aid, (2) The patient decision aid was useful in making a decision about how to get genetic testing, (3) The patient decision aid presented information in an understandable way, (4) The patient decision aid presented information about both options, (5) The patient decision aid presented information in a *biased* way. Responses were recorded based on a Likert scale of Strongly agree, Agree, Neutral, Disagree, and Strongly disagree. The scoring scale was 4, 3, 2, 1, and 0 respectively for the first 4 statements, and the final statement was reverse scored. The omission of the final satisfaction item (5) was decided after examining the reliability during preliminary analysis after finding that Cronbach's alpha was increased from 0.33 to 0.90 when this item was removed. The overall satisfaction of the patient decision aid was clarified based on the values of the first four statements out of 16 points, therefore higher values indicate a higher satisfaction.

In order to collect additional feedback regarding the usability of the survey, we ended it with an open-ended question, 'Is there anything else you want to share about your experience using the patient decision aid', as a way to address any improvements that could be made to future survey developments. Unfortunately it did not yield any responses that could clarify any benefits or limitations to utilizing the PtDA so this portion of the survey was excluded from data analysis.

Data Analysis

Data analysis was performed by a statistician at MSK. Decisional conflict, anxiety levels, knowledge of genetic testing, and satisfaction were all analyzed through proper algorithms. For

the KnowGene scale, a ‘correct’ variable was set as ‘know’ and had a score of 1, while 'incorrect' or 'don’t know' were scored as 0. The sum of all 14 items was calculated, with the possible final score being 0-14. The SURE test was scored with no = 0, and yes = 1. A total of 4 items were scored, with the final scores ranging from 0-4. A score of 3 or less indicated decisional conflict. Satisfaction was measured by scoring the first four items listed in the survey, and assigning 'strongly agree' = 4, and 'strongly disagree' = 0. The GAD-2 was a sum of two items, with total scores ranging from 0-6. Each possible response was scored from 0-3. Higher scores indicated higher anxiety. Multiple independent t-tests and ANOVA were performed to assess for any correlations between the results that were obtained from the analyses above.

Results

Demographics

Seventy-one patients initially participated in the study and 54 completed *both* the baseline survey and the post-PtDA survey. It was suspected that 4 participants completed the pre-survey multiple times due to the concordance of answer choices. The replicated surveys were not included in the overall data analysis. The majority of participants were assigned female at birth (79.63%) and reported their ethnicity to be Caucasian (66.67%), and were not Hispanic or Latino (92.59%). The median age was 51 [26-74]. Most participants reported no previous experience having genetic counseling (96.30%), nor had they had prior genetic testing (83.33%). Almost half of participants had >4-year college degree (46.30%), followed by a 4-year college degree (44.44%) (see Table 1 in Supplemental Information).

Reason for pursuing testing

There were mixed reasons participants self-reported seeking genetic testing, with the majority being due to only family history of cancer (37.04%) or both personal history and family history of cancer (35.19%). This was followed by only personal history of cancer (16.67%). A few participants reported having a family member with a known pathogenic variant as well as family history of cancer (9.26%). Pathogenic variants patients reported included: *BRCA1, MSH2,* or did not specify. One participant (1.85%) reported personal history of cancer and 'other,' giving the reason that their breast surgeon recommended testing.

Baseline self-reported understanding of genetics

Baseline understanding of genetics was self-reported by participants. The 54 participants who answered both surveys were found to have reported varying levels of understanding. Most participants indicated that they felt they understood genetics 'Very' well (n=18, 33.33%) with 9 of those participants reporting having received a >4 year college education. The following levels of understanding were 'Somewhat' (n=16, 29.63%), and 'Slightly' (n=15, 27.78%). The options with the least amount of responses were 'Not at All' (n=2, 3.70%), 'Extremely' (n=2, 3.70%), and Did Not Answer (n=1, 1.85%). Independent sample t-tests found no significant relationship between baseline understanding of genetics and choice of educational pathway type, choice in DNA sample collection, or satisfaction.

Anxiety (GAD-2 scale)

Results from the GAD-2 scale found that the majority of participants (92.59%) had a score of 0-2, indicating that they had little to no pre-existing anxiety. Four participants scored 3

or above, indicating that pre-existing anxiety was present in those individuals (7.41%), which could be due to any degree of extraneous circumstances. All four participants with an increased GAD-2 were shown to prefer a blood draw, but were equally split in their choice of pathway type. Independent sample t-tests found no significant relationship between GAD-2 score and pathway type ($t(30)=0.50$, ns), or sample type ($t(28)=-1.42$, ns).

Decisional Conflict (SURE scale)

Forty-nine participants had a decisional conflict score of 4, indicating that they likely did not regret their decision. Five participants scored less than 4, which indicates the higher likelihood that they did feel regret over their decision. Decisional conflict scores were found to be significantly correlated with satisfaction scores ($r=0.296$, $p=0.030$). Decisional conflict scores were not found to be significantly correlated with baseline reports of understanding of genetics or validated genetics knowledge scores. One participant (1.85%) was unable to make a decision after the use of the PtDA. Independent sample t-test found no significant relationship between SURE scores and pathway type ($t(29)=1.18$, ns) or sample type $t(27)=0.53$, ns).

Genetics knowledge (KnowGene scale)

The average genetics knowledge score amongst participants was 9.30 out of a maximum of 14 points, with a standard deviation of +/- 2.93. The highest score was 14/14 (n=1, 1.89%) and the lowest score was 2/14 (n=1, 1.89%), both from participants with a 4-year degree.

There were three questions that were answered correctly by most participants on the KnowGene scale. Question 1, 'Knowing about inherited risk (passed down within a family) can affect choices about cancer treatments (for example, medications or surgery),' had an accuracy

rate of 93%. Question 8, 'In the future, more information could become available that could alter the meaning of genetic testing results,' had an accuracy rate of 94%. Question 12, 'The blood relatives (for example, sister, father, or child) of a person with a mutation in a cancer risk gene might share the same gene mutation,' had an accuracy rate of 92%. A question that was frequently answered incorrectly was question 21, 'A Variant of Uncertain Significance (VUS) will not likely influence recommendations for screening or prevention,' with only an accuracy of 18%.

Participant-reported baseline understanding of genetics was significantly correlated with their KnowGene score ($r=0.432$, $p=0.002$). Thus, a higher reported baseline of understanding was seen with higher KnowGene scores. Independent sample t-tests found no significant relationship between KnowGene score and pathway type or sample type.

Satisfaction

The results found that participant satisfaction with the decision aid was overall very high (14.77 ± 1.9, out of a total of 16). Satisfaction scores were found to be significantly correlated with KnowGene scores ($r=0.324$, $p=0.017$) but not baseline self-reported understanding. Independent sample t-tests found no significant relationship between satisfaction and pathway choice or sample type. The original satisfaction measure, which consisted of 5 items, had a Cronbach's alpha of 0.33. The omission of the last reverse-scored item inquiring whether the 'the patient decision aid presented information in a biased way' was able to increase Cronbach's alpha to 0.90, thus increasing reliability considerably. Of the 54 participants who completed the post survey, 33 (61.11%) scored 16/16, indicating maximum satisfaction, while 11 (20.37%) scored 12/16, indicating that they were mostly satisfied with the PtDA.

Factors Influencing Decision

Participants were asked which factors influenced their decision regarding the pathway that they chose. They were able to choose from the following: Time, Preferred Learning, GC support, Feelings, and Other (see Figure 3 in Supplemental Information). Participants were allowed to choose more than one factor. The responses were divided into three groups: DTT (n=38), Pre-Test Counseling (n=15), and Did Not Answer (n=1). There were 30 participants that chose preferred learning as one of the factors that influenced their pathway decision, and of those participants, 21 chose to move forward with online education.

The highest response was Preferred Learning (n=13 for DTT; n=6 for Pre-Test Counseling), followed by Time (n=11 for DTT; n=3 for Pre-Test Counseling). It should be noted that both Time and Preferred Learning were recorded by 7 participants who chose the DTT pathway. There were 2 participants who chose the Pre-Test Counseling who marked three factors that influenced their decision: with one reported 'Time, Preferred Learning, and GC Support,' and the other, 'Feelings, Time, and GC Support.'Feelings were not found to be a popular factor, as only 1 participant each from DTT and Pre-Test Counseling selected this; as well as 1 DTT participant who reported Feelings and Preferred Learning.

The 1 participant who Did Not Answer selected GC support, although they did not state which pathway they would choose. Surprisingly, only 2 participants from the Pre-Test Counseling group marked GC Support, as well as 1 participant from the DTT group. 2 participants selected Other and gave the reasons as 'transportation/safety,' and 'insurance coverage.' Participants who reported they were seeking genetic testing due to a known familial mutation were significantly more likely to choose Pre-Test Counseling over DTT($LR(2, 54)=9.057$, $p=.011$).

Sample Type

Participants had the option to select the method of sample collection they would prefer. Twenty-six participants opted for a blood sample, 24 opted for a saliva sample, and 4 participants were unsure of their choice. A blood sample was chosen by all 4 participants with a GAD-2 score of 3. It was noted that 12 participants that chose to meet with a GC selected blood as their preferred sample type. Conversely, 21 participants who chose DTT as their pathway, selected saliva as their preferred sample.

Discussion

The aim of this study was to create, implement, and assess a PtDA for patients at MSK to use when choosing between the DTT pathway, or pre-test counseling in regards to genetic testing for hereditary cancer syndromes.

Satisfaction with the PtDA

Overall, participants were found to be highly satisfied with using the PtDA to assist them in choosing between pre-test counseling or online patient education. By having high satisfaction rates, it has been shown that patients are more likely to participate in the decision-making process, as seen in a systematic review of 23 trials of PtDAs in a cancer setting (Stacey et al., 2008). As also supported in the subanalysis by McAlpine et al., PtDAs are shown to improve the overall quality of decisions made by patients because they increase knowledge and are aligned with patients' values (2018). High satisfaction with the PtDA was found to be correlated with high KnowGene scores. This leads us to hypothesize that if a better understanding of genetics prior to testing correlates with a higher satisfaction rate, then providing genetics knowledge to

patients in advance can be beneficial. Neuman et al. found that in randomized controlled studies, patients' knowledge regarding their condition increased after using cancer-related PtDAs (2007). High decisional conflict scores (SURE) were found to be significantly correlated with high satisfaction scores, indicating that participants who were more satisfied with the use of the PtDA tended to have less decisional regret. This supports that PtDAs are useful in a clinical setting, as they can contribute to lower levels of decisional regret amongst patients.

Baseline understanding of genetics was not found to be correlated with satisfaction scores. This may suggest that self-assumed knowledge of genetics does not influence the overall satisfaction with using a PtDA. This may make the PtDA more accessible or approachable to a larger population of patients.

Decisional Conflict (SURE) Scores

Low decisional conflict scores (SURE scores) amongst participants were found in both pathways, thus supporting that the PtDA enabled high decisional satisfaction regardless of pathway choice. This also suggests that participants were able to make a decision that was in-line with their own personal values. One study that examined a group of breast cancer patients who used a PtDA concerning treatment were observed to be more empowered because they were more confident in their beliefs and less likely to need a formal recommendation for treatment by physicians when compared to a control group (Whelan et al., 2003).

SURE scores, however, were not found to be significantly correlated with the baseline understanding of genetics, satisfaction, or KnowGene scores. This may suggest that patients are intrinsically equipped to make the best decision for themselves, regardless of their prior knowledge of genetics. This would support the move away from paternalistic medicine.

KnowGene Scores

The average KnowGene score was found to be 9.30. High KnowGene scores were found to be correlated with high satisfaction scores, thus indicating that understanding of genetics influenced satisfaction levels. Another study that integrated the use of KnowGene scores assessed satisfaction between face-to-face genetic counseling and pre-test video genetic education for patients with prostate cancer. The results suggested no difference in satisfaction using the KnowGene scale for participants who received video genetic education (Greenberg, 2022), further establishing that alternative pre-test counseling modalities should be considered for patients at risk for hereditary cancers. The high accuracy rates of questions 1, 8, and 12 demonstrate that participants had a great understanding of the effect inherited risks have on cancer treatments, how there is still the potential to learn more information which could change the interpretation of genetic testing result, and how blood relatives might share the same genetic mutation as an affected person. Question 21 had the lowest accuracy rate amongst participants, which suggests that education concerning variants of uncertain significance (VUS) needs improvement in order to clear up any misconceptions patients may have about this type of result.

Reasons for pursuing geneting testing

Family history of cancer, personal history of cancer, and both family and personal history of cancer were found to be the top reasons participants amongst all groups had for pursuing genetic testing. More physicians and patients are becoming aware of how a family history of cancer may indicate the presence of a familial pathogenic variant being passed through generations. This had led more patients to discuss their family histories with their primary care

providers (PCPs), as well as PCPs to inquire about a family history of cancer and refer these patients for genetic testing.

Oncologists are also ordering germline genetic testing for participants with a personal history of cancer due to the fact that certain genotypes respond better to specific treatments. It also gives a better idea of the recurrence risk, as well as possible other cancers that patients could be at risk for.

Sample Type

Although both blood and saliva samples yield the same genetic results, we asked participants on their preferred method of sample collection. A prior study showed that saliva collection compared with other invasive procedures such as phlebotomy is more favorable for patients with higher anxiety levels (Soo-Quee Koh, 2007). However, all four participants who had a GAD-2 score of 3 chose a blood sample which may indicate they do not trust the accuracy of saliva, are not comfortable sending DNA samples through mail, or are potentially anxious about doing the testing on their own.

Participants that chose to meet with a GC primarily chose blood as their preferred method of sample collection. This could be attributed to the convenience of meeting with a genetic counselor at an MSK facility and potentially obtaining a blood draw within the same visit. Conversely, participants who chose the DTT pathway primarily opted for saliva. This can be due to the fact that saliva collection is quick and easy. For patients who want to review educational materials without a genetic counselor, this form of sample collection is also an independent task that does not require engaging with any medical professionals.

Decision of Preferred Educational Pathway

Factors that were not found to have any significant impact on decision making were: baseline self-reported understanding genetics, knowledge of cancer genetics (KnowGene score), decisional conflict (SURE score), overall satisfaction with the PtDA, and pre-existing anxiety (GAD-2 score). Factors that were not found to influence choice between the two educational pathways were: age, race, and sample type chosen. This suggests that the use of PtDAs and online educational options may be useful across different ages and races, and may raise awareness of saliva as an alternative to blood. There was a significant correlation between participants seeking genetic testing due to a known familial mutation and educational pathway choice, as it was more likely for them to choose Pre-Test Counseling with a GC over the DTT pathway. This could be due to a number of psychological reasons, as the threat of having a hereditary cancer syndrome may be increasingly perceived as a possible reality for these participants.

Study Limitations

The timeframe for data collection was limited, thus contributing to a small sample size that may be limited in generalizability about the trends observed in this cohort. Despite this small sample size, there was an overall 76.06% response rate to both the pre and post-survey. In attempts to avoid survey fatigue, we aimed to keep surveys short to encourage engagement and completion. It is possible there was a degree of respondent burden that contributed to a lower overall response rate in the post-decision survey or it could have been due to survey user error in which participants did not know how to progress to the next survey. This could be attributed to a lack of time or priority to complete the survey. At the time when the study was initiated, the DTT

pathway was newly launched therefore the reason behind the smaller sample size was because the volume of eligible patients was only observed to increase in the later months of the study.

A drawback to having the decision aid surveys anonymized to protect patient identities was that the surveys could be opened by a respondent multiple times via the single survey link that was sent to them. Once a survey was completed, there were no safeguards in place to prevent participants from taking it a second time. REDCap tracks participant responses, but because the data is anonymized, it does not report whether specific participants submit more than one survey.

A survey-related effect pertains to the likelihood of acquiescence bias in the surveys. This has the potential to limit the accuracy of the results as participants may choose to agree with statements or answer in a way they think researchers want them to answer without reflecting their true opinion. This may be due to limited time reviewing the statement and its content. With the inclusion of a Likert scale assessing patient satisfaction, respondents may enact extreme responding, meaning that they answer either the highest or lowest response available. This was demonstrated in a lot of participant responses throughout the satisfaction scale who elicited 'Strongly agree' across all statements because it seems like the most favorable option. However, while taking response bias into consideration, we initially integrated an opposing statement interpreting the PtDA as *biased* to report if there was a shift in answer choice by respondents. This last satisfaction measure was the only reverse-scored item. It is unclear whether patients truly felt differently about this particular item, the scoring played a role in the inconsistent responses, or if there was a problem with the item itself, therefore its inclusion was omitted.

A survey constraint pertaining to the KnowGene scale in the post-decision survey, is that two of the sixteen validated statements were not included. Ideally, to ensure a reliable and

accurate assessment, validated questionnaires should not be modified. The purpose of the KnowGene scale was to review how genetic knowledge plays into the decision-making process. In attempts to motivate participation, this survey was shortened and therefore this will need to be taken into account when comparing our results to other papers using the same KnowGene survey.

The other limitation was the lack of diversity amongst participants. All participants were exclusively internal MSK patients. The data and overall trends we see may not be applicable to other healthcare systems who have more diverse patient populations. The vast majority of participants identified as Caucasian females and had at least a 4-year college degree. Another limitation that was identified was that the PtDA was only created and offered to participants in English which contributes to a linguistic barrier and hinders accessibility for all patient populations.

Conclusion

Overall, participants on average had very high satisfaction and low decisional conflict while using the PtDA. Majority of participants elected the novel DTT pathway providing insight that its integration could benefit the workflow of genetic testing. There were no significant relationships between choosing a pathway type based on anxiety, KnowGene score, SURE score, or satisfaction. A relationship was found between pathway choice and influence for genetic testing, especially for those who reported having a family member with a confirmed predisposition or diagnosis. Those who had a family member with a variant were more likely to choose to meet with a genetic counselor.

References

Elwyn, G., Gray, J., & Clarke, A. (2000). Shared decision making and non-directiveness in genetic counselling. *Journal of medical genetics*, *37*(2), 135–138. https://doi.org/10.1136/jmg.37.2.135

Elwyn, G., O'Connor, A., Stacey, D., Volk, R., Edwards, A., Coulter, A., Thomson, R., Barratt, A., Barry, M., Bernstein, S., Butow, P., Clarke, A., Entwistle, V., Feldman-Stewart, D., Holmes-Rovner, M., Llewellyn-Thomas, H., Moumjid, N., Mulley, A., Ruland, C., Sepucha, K., … International Patient Decision Aids Standards (IPDAS) Collaboration (2006). Developing a quality criteria framework for patient decision aids: online international Delphi consensus process. *BMJ (Clinical research ed.)*, *333*(7565), 417. https://doi.org/10.1136/bmj.38926.629329.AE

Garber, J. E., & Offit, K. (2005). Hereditary Cancer Predisposition Syndromes. *Journal of Clinical Oncology*, *23*(2), 276–292. https://doi.org/10.1200/JCO.2005.10.042

Greenberg, Samantha, Elizabeth Woelmer, Morgan Devlin, Sara Low, Amanda Anson, Wendy Kohlmann, Bob Wong, et al. (2022). "Impact of Pretest Video Genetic Education in Prostate Cancer Patients: Do Patients Need Us?" *Journal of Clinical Oncology* 40, no. 6_suppl: 68–68. https://doi.org/10.1200/JCO.2022.40.6_suppl.068.

Kroenke, K., Spitzer, R. L., Williams, J. B., Monahan, P. O., & Löwe, B. (2007). Anxiety disorders in primary care: prevalence, impairment, comorbidity, and detection. *Annals of internal medicine*, *146*(5), 317–325. https://doi.org/10.7326/0003-4819-146-5-200703060-00004

Légaré, F., Kearing, S., Clay, K., Gagnon, S., D'Amours, D., Rousseau, M., & O'Connor, A. (2010). Are you SURE?: Assessing patient decisional conflict with a 4-item screening test. *Canadian family physician Medecin de famille canadien*, *56*(8), e308–e314.

Lewis K. M. (2014). Identifying hereditary cancer: genetic counseling and cancer risk assessment. *Current problems in cancer*, *38*(6), 216–225. https://doi.org/10.1016/j.currproblcancer.2014.10.002

McAlpine, K., Lewis, K. B., Trevena, L. J., & Stacey, D. (2018). What Is the Effectiveness of Patient Decision Aids for Cancer-Related Decisions? A Systematic Review Subanalysis. *JCO Clinical Cancer Informatics*, *2*, 1–13. https://doi.org/10.1200/CCI.17.00148

Meiser, B. (2005). Psychological impact of genetic testing for cancer susceptibility: An update of the literature. *Psycho-Oncology*, *14*(12), 1060–1074. https://doi.org/10.1002/pon.933

Neuman, H. B., Charlson, M. E., & Temple, L. K. (2007). Is there a role for decision aids in cancer-related decisions? *Critical Reviews in Oncology/Hematology*, *62*(3), 240–250. https://doi.org/10.1016/j.critrevonc.2006.12.006

O'Connor, A. M., Bennett, C., Stacey, D., Barry, M. J., Col, N. F., Eden, K. B., Entwistle, V., Fiset, V., Holmes-Rovner, M., Khangura, S., Llewellyn-Thomas, H., & Rovner, D. R. (2007). Do patient decision aids meet effectiveness criteria of the international patient decision aid standards collaboration? A systematic review and meta-analysis. *Medical decision making : an international journal of the Society for Medical Decision Making*, *27*(5), 554–574. https://doi.org/10.1177/0272989X07307319

O'Connor, A. M., Llewellyn-Thomas, H. A., & Flood, A. B. (2004). Modifying unwarranted variations in health care: Shared decision making using patient decision aids. *Health Affairs (Project Hope)*, *Suppl Variation*, VAR63-72. https://doi.org/10.1377/hlthaff.var.63

O'Connor, A. M., Fiset, V., DeGrasse, C., Graham, I. D., Evans, W., Stacey, D., Laupacis, A., & Tugwell, P. (1999). Decision aids for patients considering options affecting cancer outcomes: Evidence of efficacy and policy implications. Journal of the National Cancer Institute. Monographs, 25, 67–80 https://doi.org/10.1093/oxfordjournals.jncimonographs.a024212

O'Connor, A. M. (1995). Validation of a decisional conflict scale. Medical Decision Making: An International Journal of the Society for Medical Decision Making, 15(1), 25–30. https://doi.org/10.1177/0272989X9501500105

Soo-Quee Koh, D., & Choon-Huat Koh, G. (2007). The use of salivary biomarkers in occupational and environmental medicine. *Occupational and environmental medicine*, *64*(3), 202–210. https://doi.org/10.1136/oem.2006.026567

Stacey, D., Samant, R., & Bennett, C. (2008). Decision Making in Oncology: A Review of Patient Decision Aids to Support Patient Participation. *CA: A Cancer Journal for Clinicians*, *58*(5), 293–304. https://doi.org/10.3322/CA.2008.0006

Underhill-Blazey, M., Stopfer, J., Chittenden, A., Nayak, M. M., Lansang, K., Lederman, R., Garber, J., & Gundersen, D. A. (2019). Development and testing of the KnowGene scale

to assess general cancer genetic knowledge related to multigene panel testing. *Patient education and counseling*, *102*(8), 1558–1564. https://doi.org/10.1016/j.pec.2019.04.014

Whelan, T., Sawka, C., Levine, M., Gafni, A., Reyno, L., Willan, A., Julian, J., Dent, S., Abu-Zahra, H., Chouinard, E., Tozer, R., Pritchard, K., & Bodendorfer, I. (2003). Helping Patients Make Informed Choices: A Randomized Trial of a Decision Aid for Adjuvant Chemotherapy in Lymph Node-Negative Breast Cancer. *JNCI Journal of the National Cancer Institute*, *95*(8), 581–587. https://doi.org/10.1093/jnci/95.8.581

Williams, L., Jones, W., Elwyn, G., & Edwards, A. (2008). Interactive patient decision aids for women facing genetic testing for familial breast cancer: A systematic web and literature review. *Journal of Evaluation in Clinical Practice*, *14*(1), 70–74. https://doi.org/10.1111/j.1365-2753.2007.00798.x

Supplemental Information

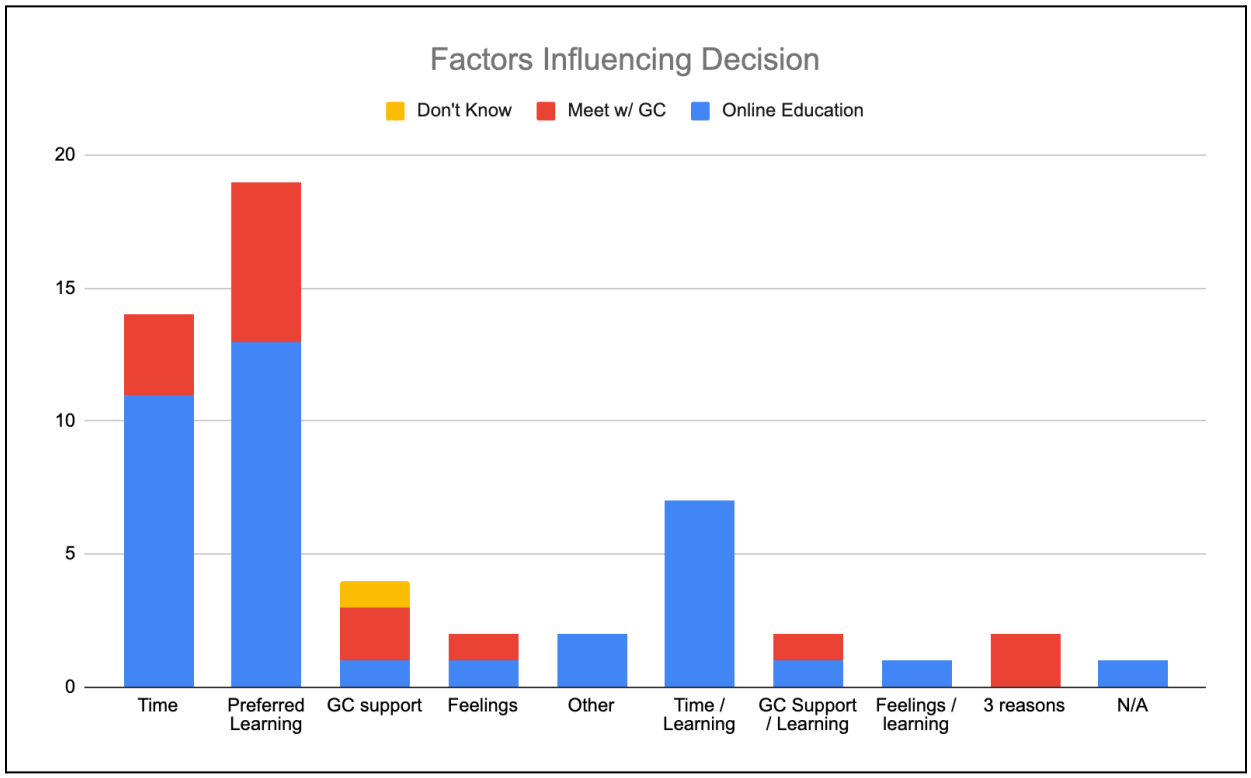

Figure 3: Factors influencing decision for pretest pathway

Characteristic	Group	n=54	%
Sex	Male	10	18.52%
	Female	43	79.63%
	Undisclosed	1	1.85%
Ethnicity	American Indian/Native Alaskan	0	0.00%
	Asian	6	11.11%
	Black/African American	7	12.96%
	Caucasian	36	66.67%
	Middle Eastern/North African	1	1.85%
	Other	1	1.85%
	Bi/Multiracial	1	1.85%
	Prefer not to answer	2	3.70%
Hispanic or Latino?	Yes	2	3.70%
	No	50	92.59%
	Prefer not to answer	2	3.70%
Education	Some college or 2-year degree	5	9.26%
	4-year degree	24	44.44%
	>4-year degree	25	46.30%
Pathway Choice	Online Education	38	70.37%
	Meet with GC	15	27.78%
	Don't Know	1	1.85%
Sample Type	Saliva	24	44.44%
	Blood	26	48.15%
	Unsure	4	7.41%

Table 1: Demographics