



## Investigation of interest in and timing preference for cancer predisposition testing and expanded carrier screening among women of reproductive age

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### ABSTRACT

**Objective:** To examine cognitive, relational, and social predictors of interest in and timing preference for cancer predisposition testing (CPT) and expanded carrier screening (ECS) offered in routine gynecologic care for women of reproductive age.

**Methods:** Women between 20 and 35 years old who were currently pregnant or had a prior pregnancy ( $N = 351$ ) completed an online survey. Bivariate and multivariable analyses were used to identify significant predictors of women's interest in and timing preference for CPT and ECS.

**Results:** Most respondents reported high interest in CPT and ECS and preferred to have them when planning for a pregnancy. Perceived importance of genetic information and negative attitude towards uncertainty predicted interest in CPT and ECS in multivariable models. Genetic knowledge predicted preference for CPT or ECS when planning for a pregnancy.

**Conclusion:** Educational and decision support tools should be developed to enhance women's knowledge and awareness of CPT and ECS and to provide them with strategies to manage uncertainty.

**Innovation:** We examined women's timing preference for CPT and ECS and the impact of partner support and trust with gynecologist. A context-specific attitudes toward uncertainty scale was used to investigate women's particular perceptions of uncertainty in genetic testing.

### 1. Introduction

As next-generation sequencing technologies continue to advance and precision medicine becomes more accessible for patients and health care providers, greater considerations have been given to DNA-based screening of unaffected individuals at a population level [1]. Pre-pregnancy is a crucial time window to engage women and their reproductive partners in acquiring information about genetic risks that may affect their own health and that of their biological children [2]. Compared with genetic testing offered during or after pregnancy, pre-pregnancy genetic testing may offer a better timing and unique opportunity to present couples with maximized genetic risk information, helping them make informed decisions about their health and reproductive options [3]. Two widely used genetic tests that are clinically relevant and salient for women of reproductive age are expanded carrier screening (ECS) and cancer predisposition testing (CPT).

ECS allows for multi-disease and pan-ethnic carrier screening, which can maximize couples' knowledge about inherited genetic risks and thus help them make informed reproductive decisions [4,5]. CPT, which is used for detecting inherited cancer syndromes [6,7], is also a viable test for women of reproductive age to inform their cancer risks and, if relevant, cancer risk management.

Research on patient preferences for genetic testing has shown that patients are often interested in receiving multiple types of genetic results [8-10]. For example, prior research suggests that CPT results have impacts on family planning decisions and approaches [11]. Additionally, women with family planning needs generally would be willing to pay for ECS that provides additional information about risks for other health conditions including cancer [12]. Taken together, integrating CPT and ECS into pre-pregnancy genetic testing may be a feasible and cost-effective approach to providing women and their partners with adequate

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information to inform reproductive decisions and decisions about their own health.

Despite its benefits, pre-pregnancy genetic testing that integrates CPT and ECS is not yet offered in the United States [13], and clinical use of both tests is less than optimal [14,15]. The inadequate utilization of both tests may be due to provider-level and patient-level factors, such as inconsistent clinical recommendations for cancer genetic testing [15], individuals' concerns about cost [16], and lack of knowledge about and interest in the testing before pregnancy [4]. Routine gynecologic care may provide a convenient and viable context in which CPT and ECS are offered to women of reproductive age. However, little research has examined women's timing preference for these tests in consideration of pregnancy, thus limiting effective communication and implementation of the tests at a population level.

Research to date has identified several correlates of women's willingness and preferences for learning about different types of genetic test results, such as health literacy and numeracy [8,17,18]. Another factor that may potentially affect women's interest is interpersonal dynamics with health care providers and romantic partners [15]. Patient trust with health care providers (e.g., gynecologists) regarding their knowledge and skills in genetic counseling and testing may affect patients' intention and motivation to undergo health screenings [4,19]. Additionally, romantic partner's support for and involvement in genetic testing is crucial to women's genetic testing decision making [20]. This is especially the case for ECS, as both partners need to be tested for autosomal recessive conditions that may not influence their health but may influence the health of their biological children [2]. Qualitative research has identified partner resistance as a major barrier to couple-based ECS [21,22]. As a whole, more research is needed to unveil a wide range of factors that influence women's interest in and timing preference for CPT and ECS offered together in the context of pregnancy.

The purpose of this study was to examine predictors of interest in population-based ECS and CPT offered as part of routine gynecologic care as well as timing preference among women of reproductive age. To better understand a wide range of predictors of interest and timing preference, we drew upon extant research on this topic as well as relevant theoretical frameworks on uncertainty management [23] and medical uncertainty [7,24], theory of planned behavior (TPB) [25], and dyadic coping [26]. Specifically, we focused on three categories of predictors encompassing cognitive, relational, and social dimensions that may account for interest in and timing preference for ECS and CPT of women of reproductive age.

## 2. Methods

### 2.1. Procedure and participants

In August 2022, we conducted an online English-language survey administered through Qualtrics Panel to recruit respondents who self-identified as meeting the eligibility criteria: female, between the ages of 20-35 years, having a prior pregnancy or being pregnant at the time of data collection. Respondents were removed from the final sample if they did not meet the eligibility criteria ( $n = 559$ ), did not provide consent at the beginning of the survey ( $n = 46$ ), or did not pass the data quality checks ( $n = 92$ ). Three metrics were used to check data quality. First, at the beginning of the survey we asked respondents if they would commit to thoughtfully provide their best answers to each question. Second, we inserted two questions in the survey that provided specific instruction for respondents to select answers. Third, we carefully examined participants' responses to two open-ended questions. The final sample size was 351; valid respondents were compensated by Qualtrics. The survey was approved as an exempt protocol by University of Utah Institutional Review Board.

### 2.2. Measures

After providing informed consent for participation, eligible respondents were asked to first read through information about multiple types of

testing, including ECS and CPT. Then, they were directed to complete a set of measures that assessed their interest in and timing preference for CPT and ECS as well as cognitive, social, and relational factors related to having genetic testing, followed by a set of demographic questions (Supplemental material).

#### 2.2.1. Interest outcome variables

Based on a prior study [18], respondents' interest in ECS and CPT were assessed by two questions using a 7-point Likert scale from "not at all interested" to "very interested." Due to the skewed distribution, we dichotomized the answers as "very interested" vs. all other categories to characterize a high level of interest. Respondents also indicated their timing preference for ECS and CPT by answering two questions with the following answer options: when planning for a pregnancy, during pregnancy, after pregnancy, not sure, and should not be offered. Given small cell counts for some options, we recoded the timing preference variable into three levels: when planning for a pregnancy, during/after pregnancy, and not sure/should not be offered.

#### 2.2.2. Predictor variables

We explored a set of cognitive, relational, and social variables that may predict women's interest in and timing preference for ECS and CPT. Scale internal consistency reliability was assessed using Cronbach's  $\alpha$ . Measured on a 0-1 scale, Cronbach's  $\alpha$  quantifies whether several items measuring the same general construct produce consistent responses; values  $\geq 0.70$  are acceptable in most fields [27,28].

**2.2.2.1. Cognitive variables.** Worry about genetic risks was assessed by three items used in prior research (Cronbach's  $\alpha = 0.86$ ) [29]. Higher scores indicated greater genetic worry. An 18-item validated measure was used to assess respondents' genetic knowledge [30]. The sum of correct answers for each respondent was calculated (Cronbach's  $\alpha = 0.72$ ); higher scores indicated greater genetic knowledge.

We assessed respondents' perceived importance of health information by using the 8-item health information orientation measure (Cronbach's  $\alpha = 0.88$ ) [31]. We also measured respondents' perceived importance of genetic information about cancer and carrier status (Cronbach's  $\alpha = 0.73$ ) [32,33]; responses were dichotomized as "very important" vs. other categories.

To measure respondents' cancer risk perceptions, we used three items to assess respondents' risk perceptions of breast, ovarian, and colon cancer compared with other people. Responses were dichotomized as "somewhat" or "a lot" more likely vs. other categories.

Respondents' attitudes toward uncertainty about genetic testing were measured by a modified 7-item measure utilized in prior research about genomic testing (Cronbach's  $\alpha = 0.83$ ) [34]. Higher scores indicated greater negative attitudes toward uncertainty and thus a greater tendency to reduce uncertainty via genetic testing. A 4-item validated measure was used to assess respondents' ability to cope with genetic test results (Cronbach's  $\alpha = 0.72$ ) [34]; higher scores indicated greater perceived efficacy to cope with genetic test results.

Respondents' subjective numeracy was measured by an 8-item validated subjective numeracy scale that assessed both numeracy preference (Cronbach's  $\alpha = 0.71$ ) and ability (Cronbach's  $\alpha = 0.88$ ) [35]. We used a 3-item measure to assess health literacy [36]; the sum of these responses was calculated.

**2.2.2.2. Relational variables.** We used a 10-item modified measure to assess respondents' perceived partner emotional support, which asked about perceived romantic partner's reactions when discussing ECS (Cronbach's  $\alpha = 0.91$ ) [37]. Perceived trust with gynecologist was assessed by a 13-item patient-provider trust scale [38]. The sum of responses for each participant was calculated, and as prior research indicates the scale has a moderate ceiling effect, a square root transformation was used to normalize the sum score [38]. Higher score indicated greater trust with gynecologist (Cronbach's  $\alpha = 0.94$ ).

2.2.2.3. *Social variables.* To assess one’s perceived social influence about genetic testing, we used four items to measure respondents’ injunctive norms and motivation to comply related to staying healthy and having genetic testing (Cronbach’s  $\alpha = 0.74$ ) [39]. Based on the theorization of social norms from TPB [25], we also used three items to measure respondents’ descriptive norms that tapped into their perceptions of family members’ and romantic partner’s interest in having genetic testing.

2.3. *Data analysis*

Respondent characteristics were reported using means and standard deviations for continuous measures and using counts and percentages for categorical measures. We used t-tests and chi-squared tests to evaluate differences in interest levels and timing preference by pregnancy status. To identify potential predictors of interest levels and timing preference, we used t-tests, analysis of variance, and chi-squared tests. Predictors with a bivariate association of  $p < 0.10$  were included in multivariable regression models [40]. Sociodemographic characteristics were also assessed in these models and retained in final adjusted models if  $p < 0.10$ . Additionally, we also tested potential interactions between pregnancy status and the cognitive, relational, and social predictors. Models predicting interest in CPT and ECS were built using multivariable logistic regression. Models predicting timing preference for CPT and ECS were built using multivariable multinomial logistic regression. Final adjusted odds ratios are presented with corresponding 95% confidence intervals. R [41] was used to perform statistical analyses with a significance level set at  $p < 0.05$ .

3. Results

3.1. *Participant characteristics*

Table 1 and Table 2 display the sociodemographic and psychosocial characteristics of the sample. The mean age of the participants was 29.6 years ( $SD = 3.9$ ). Most participants were non-Hispanic/non-Latino (90%), White (74%), and did not have Ashkenazi Jewish ancestry (96%). About 25% of the participants had a college degree or higher level of education. Most participants (80%) lived in urban areas and about half (53%) had a household income less than \$50,000. Most participants (85%) had a prior pregnancy and the remaining 15% were pregnant at the time of data collection. Of those that had a prior pregnancy, 19% planned to become pregnant in the next year. Most participants (90%) had biological children, and about 89% of the participants had a romantic partner. Approximately a quarter of the participants (26%) had prior experience with genetic testing; 68% reported having family history of cancer and 3% reported having personal history of cancer. Most respondents reported cancer genetic information (56%) and carrier status information (72%) as being very important. A perceived higher risk for breast cancer, ovarian cancer, and colon cancer was reported by 39%, 38%, and 18% of respondents respectively. Less than half of the respondents reported high levels of injunctive norms (31%) and motivation (38%) related to staying healthy, and only 19% and 25% reported high levels of injunctive norms and motivation for having genetic testing, respectively.

**Table 1**  
Sociodemographic characteristics of respondents overall and by pregnancy status.

Characteristics	Total	Had a prior pregnancy	Currently pregnant	p-value
	N = 351 n (%)	n = 297 n (%)	n = 54 n (%)	
Education (n = 351)				0.05
JHS/HS/GED	120 (34.2)	102 (34.3)	18 (33.3)	
Some college/Associate	144 (41.0)	128 (43.1)	16 (29.6)	
College/Graduate	87 (24.8)	67 (22.6)	20 (37.0)	
Race (n = 350)				0.17
White/Caucasian	260 (74.3)	225 (75.8)	35 (66.0)	
Black/African-American	45 (12.9)	34 (11.4)	11 (20.8)	
A/PI/NH/NA/AN/MR/Other	45 (12.9)	38 (12.8)	7 (13.2)	
Non-Hispanic/non-Latino/Other (n = 351)	317 (90.3)	270 (90.9)	47 (87.0)	0.53
Have Ashkenazi Jewish ancestry (n = 351)	14 (4.0)	12 (4.0)	2 (3.7)	1.00
Have a romantic partner (n = 351)	313 (89.2)	263 (88.6)	50 (92.6)	0.52
Current relationship status (n = 313)				0.65
Married	161 (51.4)	138 (52.5)	23 (46.0)	
Living as married	84 (26.8)	70 (26.6)	14 (28.0)	
Separated/Serious/Casual/Other	68 (21.7)	55 (20.9)	13 (26.0)	
Have biological children (n = 350)	315 (90.0)	267 (90.2)	48 (88.9)	0.96
Planning to become pregnant in the next year (n = 297)	53 (17.8)	53 (17.8)		-
Geographic location (n = 351)				0.19
Urban	279 (79.5)	232 (78.1)	47 (87.0)	
Rural	72 (20.5)	65 (21.9)	7 (13.0)	
Household income (n = 345)				0.45
<\$25,000	75 (21.7)	63 (21.6)	12 (22.2)	
\$25,000–\$49,999	109 (31.6)	93 (32.0)	16 (29.6)	
\$50,000–\$74,999	72 (20.9)	64 (22.0)	8 (14.8)	
>\$74,999	89 (25.8)	71 (24.4)	18 (33.3)	
Health insurance (n = 351)				0.57
None	25 (7.1)	21 (7.1)	4 (7.4)	
Public	160 (45.6)	132 (44.4)	28 (51.9)	
Private	166 (47.3)	144 (48.5)	22 (40.7)	
Have had genetic testing (n = 351)	91 (25.9)	63 (21.2)	28 (51.9)	<0.001
Have personal history of cancer (n = 351)	11 (3.1)	6 (2.0)	5 (9.3)	0.017
Have family history of cancer (n = 351)	238 (67.8)	203 (68.4)	35 (64.8)	0.72
	Mean (SD)	Mean (SD)	Mean (SD)	
Current age (n = 351)	29.6 (3.9)	29.9 (3.7)	27.9 (4.2)	<0.001

JHS - junior high school; HS - high school; GED - General Equivalency Diploma; A/PI/NH/NA/AN/MR - Asian, Pacific Islander, Native Hawaiian, Native American, Alaska Native, Multiracial; Bold p-values indicate  $p < 0.05$ ; SD - Standard Deviation; p-value by t-test or Chi-Square test

**Table 2**  
Psychosocial characteristics of respondents overall and by pregnancy status.

Characteristics	Total N = 351	Had a prior pregnancy	Currently pregnant	p-value
	n (%)	n = 297 n (%)	n = 54 n (%)	
High importance of cancer genetic information (n = 351)	196 (55.8)	169 (56.9)	27 (50.0)	0.43
High importance of carrier status information (n = 351)	253 (72.1)	215 (72.4)	38 (70.4)	0.89
Risk perception (Somewhat more likely/a lot more likely)				
Breast cancer (n = 351)	138 (39.3)	114 (38.4)	24 (44.4)	0.49
Ovarian cancer (n = 351)	132 (37.6)	112 (37.7)	20 (37.0)	1.00
Colon cancer (n = 351)	63 (17.9)	56 (18.9)	7 (13.0)	0.40
Strongly agree that the people who mean the most to me think				
I should learn more about ways I can keep myself healthy (n = 351)	109 (31.1)	92 (31.0)	17 (31.5)	1.00
I should learn more about genetic testing (n = 351)	65 (18.5)	54 (18.2)	11 (20.4)	0.85
Very motivated to do what these people want me to do regarding				
Keeping myself healthy (n = 351)	133 (37.9)	111 (37.4)	22 (40.7)	0.75
Genetic testing (n = 351)	87 (24.8)	70 (23.6)	17 (31.5)	0.29
	Mean (SD)	Range	Mean (SD)	Mean (SD)
Genetic worry (n = 351)	5.3 (1.5)	1-7	5.2 (1.5)	5.5 (1.3)
Genetic knowledge (n = 351)	12.3 (3.1)	0-18	12.3 (3.1)	11.9 (3.4)
Attitude toward uncertainty (n = 351)	4.0 (0.7)	1-5	4.0 (0.8)	4.0 (0.7)
Coping efficacy (n = 351)	3.9 (0.7)	1-5	3.9 (0.7)	3.9 (0.7)
Health orientation (n = 351)	4.1 (0.6)	1-5	4.0 (0.7)	4.1 (0.6)
Partner emotional support (n = 313)	4.1 (0.8)	1-7	4.1 (0.8)	4.1 (0.8)
Descriptive norms (n = 351)	5.0 (1.5)	1-7	5.0 (1.5)	5.4 (1.3)
Patient-provider trust (n = 351)	5.3 (2.7)	0-7	5.1 (2.8)	6.2 (1.7)
Numeracy preference (n = 351)	4.5 (1.1)	1-6	4.5 (1.1)	4.6 (0.9)
Numeracy ability (n = 351)	3.9 (1.4)	1-6	3.9 (1.4)	4.1 (1.3)
Health literacy (n = 351)	8.5 (2.2)	1-5	8.4 (2.1)	9.1 (2.5)

Bold p-values indicate  $p < 0.05$ ; SD - Standard Deviation; p-value by t-test or Chi-Square test

Overall, respondents had relatively high worry about their genetic risks ( $M = 5.3, SD = 1.5$ ) and a high level of health information orientation ( $M = 4.1, SD = 0.6$ ). They had a moderate level of genetic knowledge ( $M = 12.3, SD = 3.1$ ), subjective numeracy (for numeracy preference,  $M = 4.5, SD = 1.1$ ; for numeracy ability,  $M = 3.9, SD = 1.4$ ), and health literacy ( $M = 8.5, SD = 2.2$ ). They also reported a moderate degree of descriptive norms related to genetic testing ( $M = 5.0, SD = 1.5$ ), negative attitudes toward uncertainty ( $M = 4.0, SD = 0.7$ ) and coping efficacy ( $M = 3.9, SD = 0.7$ ), as well as partner emotional support ( $M = 4.1, SD = 0.8$ ) and trust with gynecologists ( $M = 5.3, SD = 2.7$ ). Half (50%) of the respondents were very interested in both types of testing, 11% were very interested in CPT only, 11% were very interested in ECS only, and 28% were not interested in both programs. Most respondents preferred to have CPT (70%) and ECS (70%) when planning for a pregnancy, followed by during/after pregnancy (for CPT, 19%; for ECS, 20%), and not sure/should not be offered (for CPT, 11%; for ECS, 11%).

### 3.2. Multivariable predictors of interest in CPT and ECS

Table 3 displays results of bivariate analyses for the predictors of women's interest in CPT and ECS. As indicated in Table 4 that shows multivariable predictors of women's interest in CPT and ECS, women who were very interested in CPT perceived greater importance of cancer genetic information ( $OR = 3.84, 95\% CI: 1.83-8.21$ ), had greater genetic worry ( $OR = 1.32, 95\% CI: 1.03-1.69$ ), had greater negative attitudes toward uncertainty ( $OR = 2.68, 95\% CI: 1.51-4.93$ ), and had greater numeracy preference ( $OR = 1.47, 95\% CI: 1.04-2.10$ ), compared with women with less interest in CPT. However, women who were very interested in CPT reported lower health literacy than those with less interest in CPT ( $OR = 0.79, 95\% CI: 0.67-0.93$ ). As for interest in ECS in a multivariable model, women with greatest interest in ECS perceived greater importance of carrier status information ( $OR = 3.91, 95\% CI: 1.84-8.49$ ), had greater negative attitudes toward uncertainty ( $OR = 1.92, 95\% CI: 1.11-3.44$ ), and perceived stronger efficacy of coping with genetic test results

( $OR = 2.04, 95\% CI: 1.18-3.61$ ), compared with women with less interest in ECS. Similar to women with high interest in CPT, women with substantive interest in ECS had lower health literacy than those with less interest in ECS ( $OR = 0.84, 95\% CI: 0.72-0.99$ ).

### 3.3. Multivariable predictors of timing preference for CPT and ECS

Table 5 displays results for bivariate predictors of women's timing preference for CPT and ECS. Multivariable analyses for predictors of timing preference (Table 6) suggested that women who preferred CPT when planning for a pregnancy perceived weaker injunctive norms related to genetic testing ( $OR = 0.27, 95\% CI: 0.09-0.79$ ), had more genetic knowledge ( $OR = 1.13, 95\% CI: 1.01-1.26$ ), had greater negative attitudes toward uncertainty ( $OR = 2.71, 95\% CI: 1.48-4.97$ ), and were less likely to have some college/associate education ( $OR = 0.42, 95\% CI: 0.19-0.95$ ), compared with women who preferred CPT during or after pregnancy. Additionally, results indicated that breast cancer risk perception significantly moderated the association between pregnancy status and timing preference for CPT. Specifically, pregnant women with lower perceived breast cancer risk were less likely to prefer CPT when planning for a pregnancy ( $OR = 0.32, 95\% CI: 0.11-0.93$ ). Having a higher perceived breast cancer risk increased this association by a factor of 7.98 ( $95\% CI: 1.39-45.80$ ). In other words, pregnant women with higher perceived breast cancer risk were more likely to prefer CPT when planning for a pregnancy.

Regarding women's timing preference for ECS, in a multivariable model, those who preferred ECS when planning for a pregnancy perceived weaker injunctive norms related to genetic testing ( $OR = 0.26, 95\% CI: 0.09-0.74$ ), were less likely to live as married than married ( $OR = 0.35, 95\% CI: 0.16-0.81$ ), had more genetic knowledge ( $OR = 1.13, 95\% CI: 1.01-1.26$ ), and were more likely to have a household income between \$25,000 and \$49,999 than below \$25,000 ( $OR = 3.16, 95\% CI: 1.20-8.32$ ), compared with women who preferred ECS during or after pregnancy. We did not observe any significant moderation in this model.

**Table 3**  
Bivariate predictors of interest in cancer predisposition testing and expanded carrier screening.

Characteristics	Cancer predisposition testing		p-value	Expanded carrier screening		p-value
	<i>Very interested</i>	<i>Other categories</i>		<i>Very interested</i>	<i>Other categories</i>	
	<i>n = 214</i>	<i>n = 137</i>		<i>n = 216</i>	<i>n = 135</i>	
	<i>n (%)</i>	<i>n (%)</i>		<i>n (%)</i>	<i>n (%)</i>	
Pregnancy status			0.67			0.49
Had a prior pregnancy	183 (85.5)	114 (83.2)		180 (83.3)	117 (86.7)	
Currently pregnant	31 (14.5)	23 (16.8)		36 (16.7)	18 (13.3)	
High importance of cancer genetic information	161 (75.2)	35 (25.5)	<0.001	157 (72.7)	39 (28.9)	<0.001
High importance of carrier status information	188 (87.9)	65 (47.4)	<0.001	194 (89.8)	59 (43.7)	<0.001
Risk perception (Somewhat more likely/a lot more likely)						
Breast cancer	83 (38.8)	55 (40.1)	0.89	90 (41.7)	48 (35.6)	0.30
Ovarian cancer	85 (39.7)	47 (34.3)	0.36	85 (39.4)	47 (34.8)	0.46
Colon cancer	40 (18.7)	23 (16.8)	0.76	43 (19.9)	20 (14.8)	0.29
Strongly agree that the people who mean the most to me think						
I should learn more about ways I can keep myself healthy	84 (39.3)	25 (18.2)	<0.001	90 (41.7)	19 (14.1)	<0.001
I should learn more about genetic testing	48 (22.4)	17 (12.4)	0.027	50 (23.1)	15 (11.1)	0.007
Very motivated to do what these people want me to do regarding						
Keeping myself healthy	102 (47.7)	31 (22.6)	<0.001	105 (48.6)	28 (20.7)	<0.001
Genetic testing	71 (33.2)	16 (11.7)	<0.001	68 (31.5)	19 (14.1)	<0.001
	Mean (SD)	Mean (SD)		Mean (SD)	Mean (SD)	
Genetic worry	5.6 (1.4)	4.7 (1.5)	<0.001	5.7 (1.4)	4.7 (1.4)	<0.001
Genetic knowledge	12.7 (2.8)	11.5 (3.5)	<0.001	12.6 (2.8)	11.8 (3.5)	0.018
Attitude toward uncertainty	4.3 (0.6)	3.5 (0.7)	<0.001	4.3 (0.6)	3.5 (0.7)	<0.001
Coping efficacy	4.1 (0.7)	3.6 (0.7)	<0.001	4.1 (0.7)	3.5 (0.6)	<0.001
Health orientation	4.2 (0.6)	3.8 (0.6)	<0.001	4.2 (0.7)	3.8 (0.5)	<0.001
Partner emotional support	4.2 (0.8)	3.9 (0.7)	<0.001	4.3 (0.8)	3.8 (0.7)	<0.001
Descriptive norms	5.3 (1.4)	4.7 (1.4)	<0.001	5.4 (1.4)	4.4 (1.5)	<0.001
Patient-provider trust	5.4 (2.8)	5.1 (2.7)	0.34	5.4 (2.7)	5.0 (2.8)	0.11
Numeracy preference	4.7 (1.1)	4.2 (1.1)	<0.001	4.7 (1.1)	4.2 (1.0)	<0.001
Numeracy Ability	4.0 (1.4)	3.7 (1.3)	0.06	4.0 (1.4)	3.7 (1.3)	0.06
Health literacy	8.4 (2.0)	8.7 (2.3)	0.22	8.5 (2.2)	8.5 (2.1)	0.86

Bold p-values indicate p < 0.05; SD - Standard Deviation; p-value by t-test or Chi-Square test

**Table 4**  
Multivariable logistic models showing predictors of interest in cancer predisposition testing and expanded carrier screening.

Tested predictors	Cancer predisposition testing	Expanded carrier screening
	<i>n = 313</i>	<i>n = 313</i>
	OR (95% CI)	OR (95% CI)
Currently pregnant <sup>a</sup>	0.69 (0.30, 1.61)	1.28 (0.56, 2.98)
High importance of cancer genetic information <sup>b</sup>	<b>3.84 (1.83, 8.21)</b>	1.60 (0.76, 3.34)
High importance of carrier status information <sup>b</sup>	1.80 (0.81, 3.99)	<b>3.91 (1.84, 8.49)</b>
Strongly agree that the people who mean the most to me think I should learn more about ways I can keep myself healthy <sup>b</sup>	0.98 (0.42, 2.32)	2.18 (0.92, 5.46)
Strongly agree that the people who mean the most to me think I should learn more about genetic testing <sup>b</sup>	1.01 (0.34, 3.09)	0.77 (0.26, 2.35)
Very motivated to do what these people want me to do regarding keeping myself healthy <sup>b</sup>	1.75 (0.83, 3.74)	2.08 (1.00, 4.42)
Very motivated to do what these people want me to do regarding genetic testing <sup>b</sup>	1.82 (0.67, 5.22)	0.70 (0.27, 1.84)
Genetic worry	<b>1.32 (1.03, 1.69)</b>	1.20 (0.95, 1.51)
Genetic knowledge	1.11 (0.99, 1.25)	1.04 (0.94, 1.16)
Attitude toward uncertainty	<b>2.68 (1.51, 4.93)</b>	<b>1.92 (1.11, 3.44)</b>
Coping efficacy	1.34 (0.77, 2.36)	<b>2.04 (1.18, 3.61)</b>
Health orientation	0.83 (0.42, 1.62)	0.63 (0.31, 1.24)
Partner emotional support	0.94 (0.57, 1.52)	0.99 (0.61, 1.56)
Descriptive norms	0.91 (0.68, 1.21)	1.26 (0.96, 1.67)
Numeracy preference	<b>1.47 (1.04, 2.10)</b>	1.37 (0.97, 1.94)
Numeracy ability	1.13 (0.85, 1.51)	0.97 (0.73, 1.29)
Health literacy	<b>0.79 (0.67, 0.93)</b>	<b>0.84 (0.72, 0.99)</b>
Have Ashkenazi Jewish ancestry <sup>c</sup>	0.26 (0.06, 1.14)	

OR: Odds Ratio; CI: Confidence Interval

Significant results are bolded.

<sup>a</sup> Compared with had a prior pregnancy.

<sup>b</sup> Compared with other categories.

<sup>c</sup> Compared with No Ashkenazi Jewish ancestry.

**Table 5**  
Bivariate predictors of timing preferences of cancer predisposition testing and expanded carrier screening.

Characteristics	Cancer predisposition testing			p-value	Expanded carrier screening			p-value
	When planning for a pregnancy	During/After pregnancy	Not sure/Should not be offered		When planning for a pregnancy	During/After pregnancy	Not sure/Should not be offered	
	<i>n</i> = 246	<i>n</i> = 65	<i>n</i> = 40		<i>n</i> = 244	<i>n</i> = 70	<i>n</i> = 37	
	<i>n</i> (%)	<i>n</i> (%)	<i>n</i> (%)		<i>n</i> (%)	<i>n</i> (%)	<i>n</i> (%)	
Pregnancy status				0.24				0.14
Had a prior pregnancy	210 (85.4)	51 (78.5)	36 (90.0)		210 (86.1)	54 (77.1)	33 (89.2)	
Currently pregnant	36 (14.6)	14 (21.5)	4 (10.0)		34 (13.9)	16 (22.9)	4 (10.8)	
High importance of cancer genetic information	148 (60.2)	29 (44.6)	19 (47.5)	<b>0.043</b>	149 (61.1)	33 (47.1)	14 (37.8)	<b>0.008</b>
High importance of carrier status information	194 (78.9)	37 (56.9)	22 (55.0)	<b>&lt;0.001</b>	190 (77.9)	45 (64.3)	18 (48.6)	<b>&lt;0.001</b>
Risk perception (Somewhat more likely/a lot more likely)								
Breast cancer	91 (37.0)	31 (47.7)	16 (40.0)	0.29	91 (37.3)	34 (48.6)	13 (35.1)	0.20
Ovarian cancer	98 (39.8)	25 (38.5)	9 (22.5)	0.11	98 (40.2)	25 (35.7)	9 (24.3)	0.17
Colon cancer	41 (16.7)	14 (21.5)	8 (20.0)	0.62	45 (18.4)	14 (20.0)	4 (10.8)	0.47
Strongly agree that the people who mean the most to me think								
I should learn more about ways I can keep myself healthy	79 (32.1)	22 (33.8)	8 (20.0)	0.27	79 (32.4)	24 (34.3)	6 (16.2)	0.11
I should learn more about genetic testing	39 (15.9)	19 (29.2)	7 (17.5)	<b>0.047</b>	37 (15.2)	23 (32.9)	5 (13.5)	<b>0.003</b>
Very motivated to do what these people want me to do regarding								
Keeping myself healthy	103 (41.9)	21 (32.3)	9 (22.5)	<b>0.038</b>	97 (39.8)	28 (40.0)	8 (21.6)	0.10
Genetic testing	64 (26.0)	17 (26.2)	6 (15.0)	0.31	59 (24.2)	24 (34.3)	4 (10.8)	<b>0.026</b>
	Mean (SD)	Mean (SD)	Mean (SD)		Mean (SD)	Mean (SD)	Mean (SD)	
Genetic worry	5.4 (1.5)	5.4 (1.2)	4.5 (1.6)	<b>0.001</b>	5.4 (1.5)	5.4 (1.4)	4.4 (1.6)	<b>&lt;0.001</b>
Genetic knowledge	12.8 (2.9)	11.6 (3.1)	10.3 (3.7)	<b>&lt;0.001</b>	12.7 (2.9)	11.7 (3.0)	10.1 (4.0)	<b>&lt;0.001</b>
Attitude toward uncertainty	4.1 (0.7)	3.7 (0.8)	3.6 (0.8)	<b>&lt;0.001</b>	4.1 (0.7)	3.8 (0.7)	3.4 (0.7)	<b>&lt;0.001</b>
Coping efficacy	3.9 (0.7)	3.8 (0.7)	3.7 (0.7)	<b>0.027</b>	4.0 (0.7)	3.9 (0.7)	3.4 (0.8)	<b>&lt;0.001</b>
Health orientation	4.1 (0.6)	4.0 (0.6)	3.9 (0.7)	0.06	4.1 (0.6)	4.1 (0.5)	3.7 (0.8)	<b>&lt;0.001</b>
Partner emotional support	4.2 (0.7)	3.9 (0.7)	3.7 (0.9)	<b>0.001</b>	4.2 (0.8)	4.0 (0.6)	3.7 (0.6)	<b>0.007</b>
Descriptive norms	5.1 (1.4)	5.1 (1.5)	4.5 (1.5)	0.07	5.0 (1.5)	5.3 (1.4)	4.5 (1.5)	<b>0.019</b>
Patient-provider trust	5.5 (2.6)	5.5 (2.4)	3.3 (3.3)	<b>&lt;0.001</b>	5.4 (2.7)	5.1 (2.7)	4.2 (3.0)	<b>0.030</b>
Numeracy preference	4.6 (1.1)	4.4 (1.0)	4.1 (1.2)	<b>0.042</b>	4.6 (1.1)	4.5 (1.1)	4.0 (1.1)	<b>0.017</b>
Numeracy Ability	3.9 (1.3)	4.0 (1.4)	3.4 (1.5)	0.06	3.9 (1.4)	4.0 (1.5)	3.6 (1.4)	0.35
Health literacy	8.3 (2.0)	9.1 (2.7)	8.7 (2.3)	<b>0.034</b>	8.4 (1.9)	9.1 (2.9)	8.4 (1.9)	0.06

Bold p-values indicate  $p < 0.05$ ; SD - Standard Deviation; p-value by ANOVA or Chi-Square test

## 4. Discussion and conclusion

### 4.1. Discussion

This study aimed to identify and examine cognitive, relational, and social predictors of interest in and timing preference for CPT and ECS among women of reproductive age. Overall, respondents reported relatively high levels of interest in both types of testing and largely preferred to have CPT and ECS when planning for a pregnancy. These findings provide empirical support for integrating CPT and ECS in routine gynecologic care for women of reproductive age.

We found that perceived importance of genetic information and negative attitudes toward uncertainty are two consistent predictors of women's interest in CPT and ECS. These findings corroborate with prior research that speak to the significance of perceived importance of genetic information on women's interest in multiple types of testing [8,27]. According to TPB, behavioral beliefs affect one's attitudes towards the behavior, which in turn have implications for behavioral intentions [25]. As perceived importance of genetic information is an essential component of behavioral beliefs, attaching greater importance to genetic information may prompt women to have more favorable attitudes towards genetic testing and thus increase their intentions to utilize it. Hence, decision support tools for genetic testing should highlight the importance and utility of genetic information. Theoretical frameworks on medical uncertainty [7,24] and uncertainty management [23] posit

that uncertainty as a subjective perception of information being probabilistic, ambiguous, or complex, may induce different management and coping strategies. Individuals perceiving uncertainty as a threat may be inclined to manage it via information seeking, whereas those who perceive uncertainty optimistically may choose to maintain it via information avoidance. Our findings are in line with the conceptualizations of medical uncertainty [7] and underscore the implications of the propensity of managing uncertainty via genetic testing on women's interest in seeking genetic testing [34,42]. Additionally, we found a significant association between perceived coping efficacy and interest in ECS, as greater perceived efficacy to cope with uncertainty in genetic test results could enhance women's interest in pursuing ECS. Taken together, these findings illuminated that clinical education and decision support tools should acknowledge uncertainty as an important concern for people undergoing genetic testing and provide tangible resources to equip them with information and ability to cope with uncertainty.

Prior research supports that women's genetic knowledge affects their interest in genetic testing [2,18]. In line with it, we found that genetic knowledge is a consistent predictor of preferring CPT and ECS when planning for a pregnancy. This finding highlights the importance of developing educational programs to enhance women's genetic knowledge as a prerequisite for implementing genetic testing in routine gynecologic care [4]. The only significant interaction effect of pregnancy status and perceived breast cancer risks on timing preference for CPT illustrated that pregnant women who have heightened concerns about breast cancer risks may need further

**Table 6**  
Multivariable multinomial models showing predictors of timing preferences of cancer predisposition testing and expanded carrier screening.

Tested predictors	Ref: During/After pregnancy			
	Cancer predisposition testing		Expanded carrier screening	
	n = 313		n = 313	
	When planning for a pregnancy	Not sure/Should not be offered	When planning for a pregnancy	Not sure/Should not be offered
	OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)
Currently pregnant <sup>a</sup>	<b>0.32 (0.11, 0.93)</b>	0.15 (0.02, 1.45)	0.60 (0.26, 1.37)	0.48 (0.12, 1.98)
High importance of cancer genetic information <sup>b</sup>	0.92 (0.37, 2.24)	1.99 (0.49, 8.19)	1.69 (0.73, 3.87)	2.27 (0.53, 9.85)
High importance of carrier status information <sup>b</sup>	2.28 (0.92, 5.69)	0.75 (0.19, 2.97)	1.52 (0.63, 3.67)	0.67 (0.16, 2.83)
Strongly agree that the people who mean the most to me think I should learn more about ways I can keep myself healthy <sup>b</sup>	1.03 (0.39, 2.73)	1.03 (0.23, 4.71)	2.01 (0.78, 5.23)	1.74 (0.32, 9.50)
Strongly agree that the people who mean the most to me think I should learn more about genetic testing <sup>b</sup>	<b>0.27 (0.09, 0.79)</b>	0.58 (0.10, 3.42)	<b>0.26 (0.09, 0.74)</b>	0.30 (0.04, 2.27)
Very motivated to do what these people want me to do regarding keeping myself healthy <sup>b</sup>	1.39 (0.61, 3.16)	0.81 (0.23, 2.88)	0.88 (0.40, 1.95)	0.45 (0.11, 1.85)
Very motivated to do what these people want me to do regarding genetic testing <sup>b</sup>	0.98 (0.38, 2.51)	0.99 (0.20, 4.93)	0.81 (0.33, 2.00)	0.78 (0.15, 4.18)
Genetic worry	0.92 (0.69, 1.22)	0.69 (0.47, 1.02)	0.98 (0.75, 1.29)	0.73 (0.49, 1.08)
Genetic knowledge	<b>1.13 (1.01, 1.26)</b>	0.90 (0.77, 1.06)	<b>1.13 (1.01, 1.26)</b>	0.90 (0.76, 1.07)
Attitude toward uncertainty	<b>2.71 (1.48, 4.97)</b>	1.29 (0.55, 3.03)	1.81 (0.99, 3.31)	0.90 (0.37, 2.22)
Coping efficacy	0.93 (0.52, 1.67)	1.06 (0.45, 2.51)	1.02 (0.57, 1.83)	0.76 (0.30, 1.90)
Health orientation	1.10 (0.53, 2.29)	2.50 (0.85, 7.39)	0.58 (0.27, 1.25)	1.14 (0.34, 3.87)
Partner emotional support	1.26 (0.74, 2.12)	0.71 (0.35, 1.46)	1.32 (0.81, 2.15)	1.09 (0.49, 2.45)
Descriptive norms	0.78 (0.58, 1.05)	0.80 (0.52, 1.23)	0.83 (0.62, 1.12)	0.80 (0.49, 1.28)
Numeracy preference	1.17 (0.81, 1.69)	0.96 (0.57, 1.62)	1.21 (0.85, 1.72)	0.75 (0.42, 1.34)
Numeracy ability	0.92 (0.68, 1.26)	0.87 (0.55, 1.38)	0.95 (0.70, 1.28)	1.49 (0.87, 2.56)
Education <sup>c</sup>				
Some college/Associate	<b>0.42 (0.19, 0.95)</b>	<b>0.22 (0.07, 0.72)</b>	0.52 (0.23, 1.18)	<b>0.12 (0.03, 0.48)</b>
College/Graduate	0.61 (0.23, 1.59)	0.47 (0.12, 1.80)	0.58 (0.21, 1.63)	0.24 (0.05, 1.10)
Current relationship status <sup>d</sup>				
Living as married	0.45 (0.20, 1.01)	0.45 (0.13, 1.56)	<b>0.35 (0.16, 0.81)</b>	0.29 (0.08, 1.07)
Separated/Serious/Casual/Other	0.78 (0.34, 1.80)	1.27 (0.40, 4.08)	0.69 (0.29, 1.64)	0.39 (0.10, 1.56)
Breast cancer risk perception (Somewhat more likely/a lot more likely)	0.53 (0.25, 1.11)	0.40 (0.13, 1.21)		
Currently pregnant*Breast cancer risk perception	<b>7.98 (1.39, 45.80)</b>	<b>14.81 (0.78, 28.99)</b>		
Hispanic/Latino <sup>e</sup>			0.68 (0.24, 1.99)	2.51 (0.52, 12.06)
Household income <sup>f</sup>				
\$25,000–\$49,999			<b>3.16 (1.20, 8.32)</b>	<b>5.48 (1.27, 23.59)</b>
\$50,000–\$74,999			1.56 (0.58, 4.19)	1.57 (0.30, 8.28)
Greater than \$74,999			1.26 (0.44, 3.61)	0.97 (0.16, 5.88)
Age			1.02 (0.93, 1.11)	0.89 (0.77, 1.02)

OR: Odds Ratio; CI: Confidence Interval

Significant results are bolded.

<sup>a</sup> Compared with had a prior pregnancy.

<sup>b</sup> Compared with other categories.

<sup>c</sup> Compared with Junior High School/High School/General Education Diploma.

<sup>d</sup> Compared with Married.

<sup>e</sup> Compared with Non-Hispanic/non-Latino/Other.

<sup>f</sup> Compared with Less than \$25,000.

support for making decisions about CPT and ECS during routine gynecologic care [43].

Results of multivariable models indicated that women who preferred CPT and ECS during or after pregnancy had greater injunctive norms about genetic testing (e.g., perceiving stronger approval of genetic testing from others) than those who preferred them when planning for a pregnancy. One possible explanation for this finding is that respondents might confuse CPT and ECS with more common types of genetic testing (e.g., prenatal testing) that are normally conducted during or after pregnancy [19]. Thus, clinical education programs should inform women about the differences among divergent types of testing while taking into consideration information complexity and overload [18]. Notably, bivariate associations suggested that the group answering not sure or should not be offered regarding timing for genetic testing had the lowest levels of genetic knowledge, genetic worry, social influence, as well as partner support and trust with gynecologists. Although this group may be heterogeneous in nature, the findings suggest that more efforts are needed to address these women's concerns and lack of knowledge about genetic testing by effectively communicating information about the procedure and nature of

genetic testing and its potential benefits for informing risk management and reproductive choices [2].

These findings should be interpreted in light of several limitations. First, the convenience sample may limit the generalizability of the findings to certain sub-populations characterized by race, ethnicity, and geographic locations. Respondents may also be more comfortable with genetic testing in general than individuals who did not opt to participate in the study. Second, despite that the sample included women with a prior or current pregnancy, the sample sizes were imbalanced regarding pregnancy status. Additionally, most respondents already experienced pregnancy, which may reduce their interest in CPT and ECS for future reproduction. Third, respondents' reports of interest in and timing preference for CPT and ECS were not based on their actual utilization of genetic testing. Hence, a prospective cohort study that uses a diverse and balanced sample in terms of pregnancy experience and examines women's actual experience with CPT and ECS will provide information about the causal links between a range of factors and women's interest in and timing preference for CPT and ECS. Fourth, we combined "not sure" and "should not be offered" responses due to small cell counts. Future studies with an adequate number of responses

from each category are needed to better understand the differences between these two populations. Fifth, we acknowledge that although we provided education about the different types of genetic testing to respondents, this information may not be adequately comprehended by respondents with low genetic knowledge. Lastly, we only examined patient-level factors that accounted for women's interest in and timing preference for CPT and ECS. Provider-level factors, such as clinicians' recommendations for genetic testing, should be further examined as potential barriers to the integration of CPT and ECS in routine gynecologic care.

#### 4.2. Innovation

Given the important consideration of implementing population-level CPT and ECS for women of reproductive age, one innovation of this research is the investigation of predictors of women's timing preference for CPT and ECS in the context of pregnancy. This research is also one of the first to explore the potential impact of interpersonal dynamics on women's interest in and timing preference for genetic testing. Perceived partner support and trust with gynecologists were significant bivariate predictors of timing preference for CPT and ECS. These findings have merits in that dyadic coping with romantic partners and provider-level factors could potentially affect women's decisions in seeking genetic testing that have implications on their own health and that of their biological children.

We also examined if attitudes toward uncertainty is associated with interest in and timing preference for CPT and ECS. We utilized an instrument that specifically taps into one's attitudes toward uncertainty reduction via genetic testing, allowing us to examine respondents' perceptions of uncertainty that is particularly relevant to genetic testing. This is a different approach from previous research in which general measures such as intolerance of uncertainty were used in the context of genetic testing [44]. The significant effects of negative attitudes toward uncertainty on interest in CPT and ECS suggest that it is methodologically important to assess specific aspects of uncertainty rather than general individual differences in approaching uncertain situations [18,45].

#### 4.3. Conclusion

In conclusion, we investigated cognitive, relational, and social factors that are related to interest in and timing preference for CPT and ECS among women of reproductive age. We found that perceived importance of genetic information and negative attitudes toward uncertainty are two consistent predictors of interest in CPT and ECS, whereas genetic knowledge is a consistent predictor of preferring CPT and ECS when planning for a pregnancy. Our findings provide empirical support for developing educational programs and decision support tools to enhance knowledge and awareness of genetic testing among women of reproductive age and to provide them with coping mechanisms to effectively manage uncertainty. Future research should further explore women's perceptions of uncertainty associated with CPT and ECS and examine the actual utilization of CPT and ECS for women with varied experiences with pregnancy.

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#### Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

## Appendix A. Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.pecinn.2023.100128>.

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