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Original Research

Hereditary Cancer Risk Using a Genetic Chatbot Before Routine Care Visits

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OBJECTIVE: To examine user uptake and experience with a clinical chatbot that automates hereditary cancer risk triage by collecting personal and family cancer history in routine women's health care settings.

METHODS: We conducted a multicenter, retrospective observational study of patients who used a web-based chatbot before routine care appointments to assess their risk for hereditary breast and ovarian cancer, Lynch syndrome, and adenomatous polyposis syndromes. Outcome measures included uptake and completion of the risk-assessment and educational section of the chatbot interaction and identification of hereditary cancer risk as evaluated against National Comprehensive Cancer Network criteria.

RESULTS: Of the 95,166 patients invited, 61,070 (64.2%) engaged with the clinical chatbot. The vast majority completed the cancer risk assessment (89.4%), and most completed the genetic testing education section (71.4%), indicating high acceptability among those who opted to engage. The mean duration of use was 15.4 minutes (SD 2 hours, 56.2 minutes) when gaps of inactivity longer than 5 minutes were excluded. A personal history of cancer was reported by 19.1% (10,849/56,656) and a family history of cancer was reported by 66.7% (36,469/54,652) of patients who provided the relevant information. One in four patients (14,850/54,547) screened with the chatbot before routine care appointments met National Comprehensive Cancer Network criteria for genetic testing. Among those who were tested, 5.6% (73/1,313) had a disease-causing pathogenic variant.

CONCLUSION: A chatbot digital health tool can help identify patients at high risk for hereditary cancer syndromes before routine care appointments. This scalable intervention can effectively provide cancer risk assessment, engage patients with educational information, and facilitate a path toward preventive genetic testing.

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Pathogenic variants in hereditary breast and ovarian cancer susceptibility genes can increase an affected individual's lifetime risk of developing cancer from approximately 11% to between 20% and 80%.[1,2] Genetic testing to identify such variants can reduce this risk by prompting health care professionals and patients to undertake risk-reducing measures, including increased surveillance, early detection, medication, and surgical interventions.[3–6] Unfortunately, less than 20% of women with a family history of *BRCA*-related cancers report having a conversation

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about genetic testing with their health care professional,[7] and only 10–20% of high-risk women are reported to be tested.[8,9] Moreover, Black women are 16 times less likely and Spanish-speaking Hispanic women are two times less likely to discuss genetic testing with a health care professional compared with White women.[10] Equitable and widely available cancer risk assessments and subsequent genetic testing for high-risk individuals are needed to reduce cancer incidence and address disparities in cancer detection and outcomes.

The American College of Obstetricians and Gynecologists recommends that obstetrician–gynecologists regularly perform hereditary cancer risk assessments of their patients.[11] Family history collection remains the gold standard for triaging patients into cancer-risk categories, [12] but this method is time-consuming, inaccurate, and generally underused.[13–16] Scalable and integrated solutions are needed to aid health care professionals in identifying at-risk patients.

Digital health technologies are a promising avenue for improving the efficiency and accessibility of genetic health care,[17] and have been shown to provide health information that is noninferior to physician consult.[18] To explore the acceptance and feasibility of a digital solution for conducting cancer risk assessment in routine women's health care, we examined user uptake and experience with a web-based chatbot that assesses an individual's risk for hereditary breast and ovarian cancer, Lynch, and adenomatous polyposis (hereafter, polyposis) syndromes. In addition, we present findings on risk status among largely healthy individuals using the platform, including genetic testing results, where available.

ROLE OF THE FUNDING SOURCE

The authors had access to relevant aggregated study data and other information (such as study protocol, analytic plan and report, validated data table, and clinical study report) required to understand and report research findings. The authors take responsibility for the presentation and publication of the research findings, have been fully involved at all stages of publication and presentation development, and are willing to take public responsibility for all aspects of the work. All individuals included as authors and contributors who made substantial intellectual contributions to the research, data analysis, and publication or presentation development are listed appropriately. The role of the sponsor in the design, execution, analysis, reporting, and funding is fully disclosed. The authors' personal interests, financial or nonfinancial, relating to this research and its publication have been disclosed.

METHODS

We conducted a retrospective, observational study of technology usage metrics and risk assessment outcomes in patients invited to use a chatbot (a computer program that simulates human conversation via text or voice) that was part of a digital health platform for collecting personal and family cancer history, assessing hereditary cancer risk, providing genetic testing education, and flagging high-risk patients for health care professionals.

Between January 2019 and January 2021, a total of 180 U.S. clinics distributed across all four U.S. Census regions agreed to use the digital platform for preappointment assessment of hereditary cancer risk. These included clinics providing obstetrics and gynecology care, primary care, GI specialty care, oncology care, and surgical services. Each clinic signed a Health Insurance Portability and Accountability Act (HIPAA)–compliant Business Association Agreement or a corresponding Master Service Agreement to ensure protected health information was handled securely. Clinic staff were trained on how to implement the platform within their existing practice workflow.

Patients with appointments scheduled at participating clinics were invited to engage with the chatbot through a link sent to their mobile phone or email, typically 5 days before an upcoming appointment. Only patients with a valid email address or mobile phone number on file were invited. Patients who did not open or complete the chat after the initial invitation were automatically sent reminders 72 hours and 24 hours before the appointment. Data entered into the digital health platform (which included the patient-facing chatbot and a clinician portal) were deidentified and approved for use in this study by an independent institutional review board (Western IRB 20161796). Pregnant patients were excluded because genetic testing for adult-onset disorders is not typically offered as a part of prenatal management.[19]

The clinical chatbot, Gia (Genetic Information Assistant), was designed to simulate patient and health care professional conversation regarding various hereditary disease risks, including inherited cancers.[20] The chatbot used both a scripted conversation and natural language processing that allowed patients to ask their own questions. The chatbot content was developed by genetic counselors to collect personal and family histories, and present users with education through interactive, conversational text exchanges. The chatbot was trained on hundreds of concerns,

questions, and responses (ie, intents) related to clinical genetics, including privacy and billing questions. The platform is HIPAA-compliant and SOC2-certified to safeguard the integrity, confidentiality, and accessibility of health information. The tool is web-based and therefore not stored on a user's device.

The chatbot offered a decision tree of questions and provided information to users who could select or input responses via a smartphone, tablet, or computer. For most users, the chat comprised three main sections: 1) introductory screens, 2) cancer-risk assessment (Fig. 1A and B), and 3) genetic testing education (Fig. 1C and D). Users could navigate the direction of the exchange, but some content was mandatory regardless of which conversational path was selected; for example, educational information on the type of results a patient might receive from genetic testing was always presented. Users could suspend their sessions and return without loss of previously entered information. The chatbot directed all minors (younger than 18 years) and pregnant women down a different conversational path that skipped the cancer-risk assessment and pretest education; these users were told to speak directly with their health care professionals but counted among those who completed the chat when they engaged with options presented to them. All chats were enabled in English and Spanish. If users asked a question that could not be answered by the chatbot, they were prompted to ask their health care professional and provided with a clinic phone number.

To identify patients eligible for hereditary cancer genetic testing, the chatbot was programmed to assess patient-reported personal and family history against National Comprehensive Cancer Network guidelines for hereditary breast and ovarian cancer, Lynch, and polyposis syndromes. Over the 2-year duration of this study, National Comprehensive Cancer Network guidelines for[1] hereditary breast and ovarian cancer syndrome included v2.2019, v3.2019, and v1.2020; and for[2] Lynch and polyposis syndromes included v1.2018, v3.2019, and v1.2020. Optional Tyrer-Cuzick analysis was included for health care professionals who wanted lifetime breast cancer risk assessment of patients. The clinical content was developed by a team of experienced American Board of Genetic Counseling-certified genetic counselors with expertise in hereditary cancer syndromes. Patient data collected by the chatbot and a transcript of the interaction between the chatbot and the patient were made available to health care professionals through the clinician portal, and the composite data could be transferred to the patient's chart. Patients were not informed of their risk status via the chatbot; rather, their health care professional was alerted through the clinician portal and assumed responsibility for discussing next steps with the patient, which could include ordering genetic testing for high-risk patients from any commercial laboratory.

Patients who clicked on the invite link to the chatbot were presented with an introductory screen that included a HIPAA badge, followed by the option to engage with the chatbot by clicking a button labeled "Hi, Gia!" or an icon with a "thumbs up" symbol. Patients who clicked either of these buttons were considered to have engaged with the chatbot and are hereafter called "users." Patients had to complete their chat before 12:01 am of the day after their scheduled appointment and duration of use was measured based on the time between first and last interaction.

A satisfaction question was presented to users on completion of the chat. Users could rank their satisfaction by selecting one to five stars or one of three emoji faces with sad, neutral, and happy expressions. The two scales were converted to numerical scores (one point for each star or one point, three points, or five points for sad, neutral, and happy emoji faces, respectively) and the resulting values were averaged for an overall satisfaction score.

Health care professionals working in five clinics were given the option to order genetic testing directly through the physician portal of the digital platform. Data from the patient chat were used to flag high-risk patients and was collated to facilitate health care professional completion of an electronic test requisition form. Testing using large multi-gene panels of at least 30 cancer predisposition genes was conducted at one of two Clinical Laboratory Improvement Amendments–certified diagnostic testing laboratories that offered full sequencing, deletion–duplication analysis, and copy number variant detection of coding regions and surrounding intronic sequence with high analytic validity. Test results as reported by the testing laboratories (positive, uncertain, or negative) were available to health care professionals in the digital portal and de-identified and converted into data tables for this study.

Information collected from patients by the chatbot included age, race and ethnicity, personal medical history, and family medical history. Race and ethnicity were included in this study to explore potential disparities in chatbot usability and health care history. Age was also provided by participating clinics for some patients. Additional data, including clinic type, chatbot invitation date, chatbot usage duration, and genetic testing results (where applicable), were

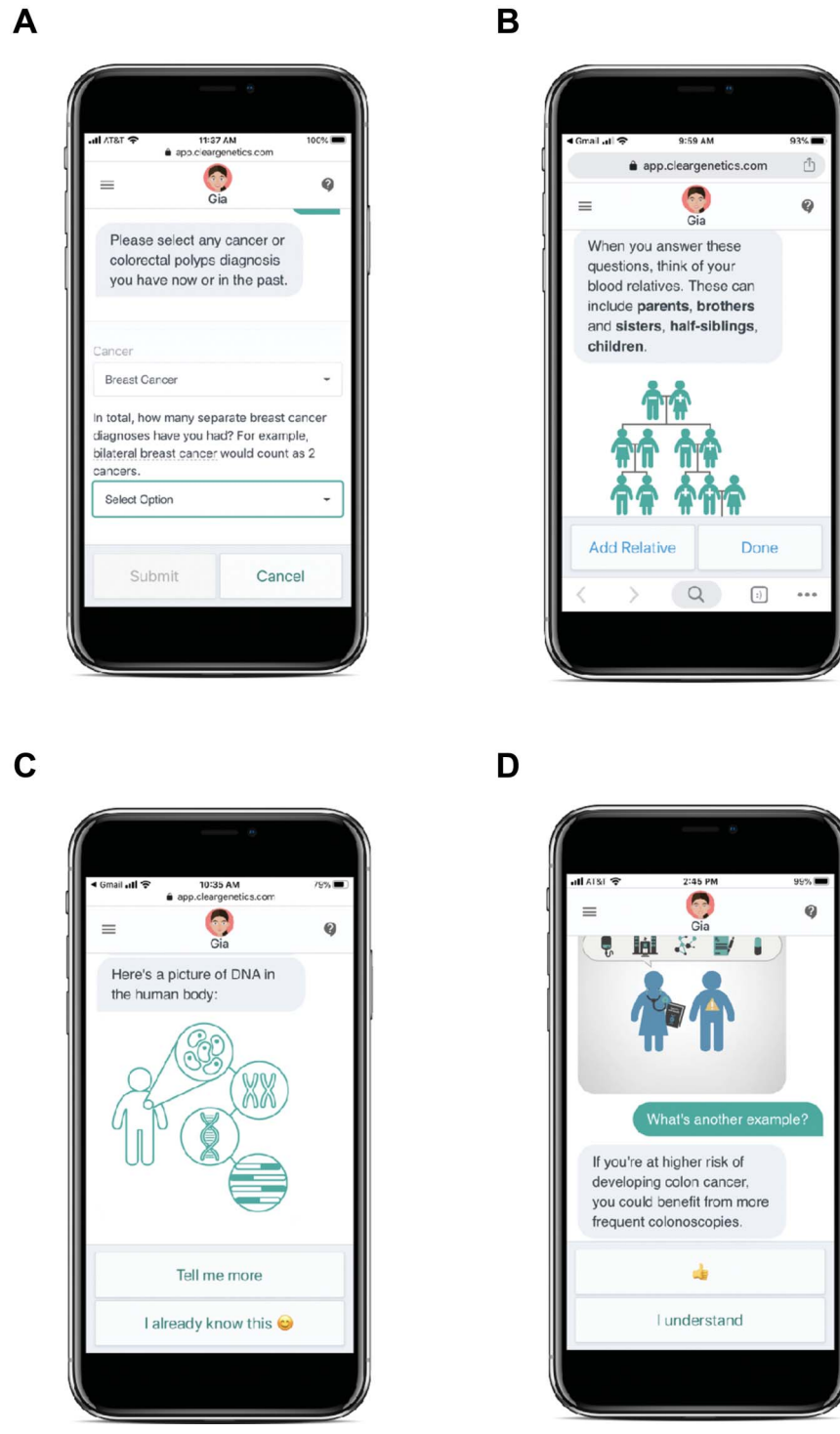

Fig. 1. Examples of chatbot interactions. Representative screenshots of chatbot interactions: a question about the user's personal medical history **(A)**; guidance on providing family cancer history information **(B)**; genetic testing education with the option to get additional details **(C, D)**. Images courtesy of Invitae. Used with permission.

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extracted from the chatbot platform. Missing data (presented as "Unknown" throughout) resulted from patient behavior (eg, incomplete chats, skipped questions) and platform updates, including questions added during the study period and changes to risk-assessment rules after National Comprehensive Cancer Network guideline updates. Analyses were conducted using statistical software R. A one-sided *t* test was used to compare the average age among users and nonusers. Outcomes included whether patients met

National Comprehensive Cancer Network criteria for hereditary breast and ovarian cancer, Lynch, or polyposis syndromes, self-reported familial variants for cancer risk, Tyrer-Cuzick status, and genetic testing results. Participants were stratified by age groups (based on cut-offs relevant to National Comprehensive Cancer Network guidelines), race and ethnicity, clinic type, personal or family history of cancer, National Comprehensive Cancer Network criteria status, and whether or not they completed the chat.

RESULTS

A total of 95,166 individuals scheduled for appointments at 180 clinics across the United States were invited to engage with the chatbot. Most clinics (n=96) provided obstetrics and gynecology care; the remaining clinics provided oncology (n=48), primary,[13] gastroenterology,[11] imaging,[8] and unknown[4] care. Most invited patients (85.9%, n=81,792) were scheduled for appointments with obstetrics and gynecology, primary care, or routine cancer screening clinics. Overall, 61,070 (64.2%) patients invited to use the chatbot engaged with the link, inputting at least one response, which minimally included clicking on an introductory response. Among these responders, self-reported demographics indicated that most (96.3%) were female and nearly half (45.5%) were 40–60 years of age (Table 1). By self-report, the majority (58.8%) were White, 10.9% were Hispanic, and 9.7% were Black. A small proportion (1.9%) reported being adopted, and less than 1% engaged with the chatbot in Spanish. Patient age was provided by the clinics for 27,972 nonusers. These nonusers were significantly older on average than users (53.5 years vs 48.4 years, $P<.001$).

Of the 61,070 users who engaged with the chatbot, 54,547 (89.3%) completed the risk-assessment section (Fig. 2). Users excluded from the assessment were 2,895 pregnant women (4.7% of users) and 445 minors younger than age 18 years (0.7% of users). An additional 2,621 users started but did not complete the assessment (4.3% of users) and 562 (1.0% of users) declined the assessment. The educational section of the chat was completed by 43,575 (71.4%) of users.

On average, the mean duration of use was 10 hours and 17.9 minutes (Appendix 1, available online at http://links.lww.com/AOG/C487). However, some users had lengthy periods of inactivity lasting multiple days. To limit the effect of these long gaps in activity on usage metrics, we excluded all gaps of inactivity longer than 5 minutes. When activity gaps were excluded, the mean duration of use was 15.4 minutes (SD 2 hours, 56.2 minutes) (Appendix 1, http://links.lww.com/AOG/C487). Among users who completed the assessment, those with a personal and family history of cancer had the longest mean usage duration (18.3 minutes) (excluding gaps) compared with those without personal or family history of cancer (11.0 minutes) (Appendix 2, available online at http://links.lww.com/AOG/C487).

Users who completed the chat (both the risk-assessment and education sections) were prompted to rate their satisfaction with their chatbot experience. On a scale of one to five, where five was the highest satisfaction score, the average score among 39,215 responders was 4.6.

Among the 56,656 individuals who provided personal history information, 2,923 (5.2%) reported a personal history of colon polyps and 10,849 (19.1%) reported a personal history of cancer, most frequently colorectal cancer (8.8%), breast cancer (3.4%), and skin cancer (2.0%) (Appendix 3, available online at http://links.lww.com/AOG/C487). Among the 54,652 users who provided family history, 36,469 (66.7%) reported a family history of cancer, most frequently breast cancer (34.3%), colorectal cancer (18.7%), and lung cancer (14.4%) (Appendix 3, http://links.lww.com/AOG/C487). Some users were also asked whether they or a member of their family had previously been tested for cancer risk genes. Of the 21,493 users who were presented with this question, 10.6% (n=2,269) indicated that they or a family member had been tested and 3.6% (n=772) reported a positive result, or "familial variant."

National Comprehensive Cancer Network criteria were met by 14,850 (27.2%) individuals who completed the risk-assessment section of the chat. Of these, 11,126 users (20.4% of risk assessment completers) met hereditary breast and ovarian cancer criteria (only), 1,300 (2.4%) users met Lynch criteria (only), 41 (0.08%) users met polyposis criteria (only), and 443 (0.8%) reported a hereditary breast and ovarian cancer-, Lynch-, or polyposis syndrome-associated familial variant (only) (Fig. 2). An additional 1,488 (2.7%) users met multiple criteria, and another 452 (0.8%) users met criteria but data were missing on criteria type (Appendix 4, available online at http://links.lww.com/AOG/C487). Among the 14,850 high-risk individuals were 3,649 women younger than age 40 years (25.0% of individuals meeting National Comprehensive Cancer Network criteria) who were identified as high-risk before a routine visit (mostly annual well-woman visits) (Table 2). Additional characteristics of patients who met National Comprehensive Cancer Network criteria for Lynch and hereditary breast and ovarian cancer syndrome and Tyrer-Cuzick criteria are shown in Table 2. More

Table 1. Self-Reported Characteristics of Chatbot Users

Characteristic	Users (n=61,070)	Completed Chat (n=43,575)
Age (y)		
Younger than 18	440 (0.7)	312 (0.7)
18–24	3,476 (5.7)	2,370 (5.4)
25–39	13,697 (22.4)	9,824 (22.5)
40–60	27,759 (45.5)	19,958 (45.8)
Older than 60	15,279 (25.0)	10,871 (24.9)
Unknown	419 (0.7)	240 (0.6)
Sex or gender		
Female	58,781 (96.3)	41,978 (96.3)
Male	1,242 (2.0)	868 (2.0)
Unknown	1,047 (1.7)	729 (1.7)
Race and ethnicity		
Ashkenazi Jewish	269 (0.4)	174 (0.4)
Asian	2,548 (4.2)	1,856 (4.3)
Black	5,927 (9.7)	4,367 (10.0)
Hispanic	6,626 (10.9)	4,570 (10.5)
Mediterranean	39 (0.06)	28 (0.06)
Native American	87 (0.1)	67 (0.2)
White	35,910 (58.8)	25,872 (59.4)
Other*	1,085 (1.8)	741 (1.7)
Multiple†	4,220 (6.9)	2,960 (6.8)
Unknown	4,359 (7.1)	2,940 (6.7)
Adopted		
No	55,854 (91.5)	39,885 (91.5)
Yes	1,150 (1.9)	818 (1.9)
Unknown	4,066 (6.7)	2,872 (6.6)

Data are n (%).
* Other includes groups with fewer than 160 users, including but not limited to: French Canadian, Middle Eastern, Pacific Islander, Portuguese, and Sephardic Jewish.
† Multiple includes individuals who selected two or more of the groups identified in the table.

than two thirds of users who met National Comprehensive Cancer Network criteria for genetic testing also completed the education component of the chat (Appendix 4, http://links.lww.com/AOG/C487).

Where applicable, users were also assessed for Tyrer-Cuzick 8.0 criteria, which are distinct from National Comprehensive Cancer Network criteria and assess lifetime breast cancer risk. A total of 3,844 individuals met the Tyrer-Cuzick risk threshold of 20% for lifetime breast cancer risk (Table 2), and of these, 1,551 (40.3%) did not meet any National Comprehensive Cancer Network guideline. The discordance between Tyrer-Cuzick criteria and National Comprehensive Cancer Network guidelines was especially pronounced among Asian women, as 56.5% of Asian women who met Tyrer-Cuzick criteria did not meet National Comprehensive Cancer Network criteria (Appendix 5, available online at http://links.lww.com/AOG/C487).

For a subset of users, their health care professional could directly order genetic testing through the associated clinician portal, alleviating the need to complete a separate test requisition. Genetic testing was ordered for 29.0% (1,622/5,594) with this option. Of these, 1,313 had a result available within the study period, of whom 73 (5.6%) were found to have a positive result for a pathogenic or likely pathogenic variant, 342 (26.0%) had a variant of uncertain significance, and 898 (68.4%) were negative for variants in the hereditary cancer genes tested (Table 3). Among users with both personal and family history, 30 (6.6%) had a positive result and 121 (26.8%) had a variant of uncertain significance (Table 3). Among users who met National Comprehensive Cancer Network criteria for genetic testing related to hereditary breast and ovarian cancer risk, 5.8% had a positive result and 26.9% had a variant of uncertain significance.

DISCUSSION

This study describes the real-world experience of 61,070 users of a digital platform that provides individual risk evaluation for hereditary cancers and genetic testing education. Within this largely healthy cohort, 27% of individuals were triaged as high-risk for hereditary breast and ovarian cancer, Lynch, or

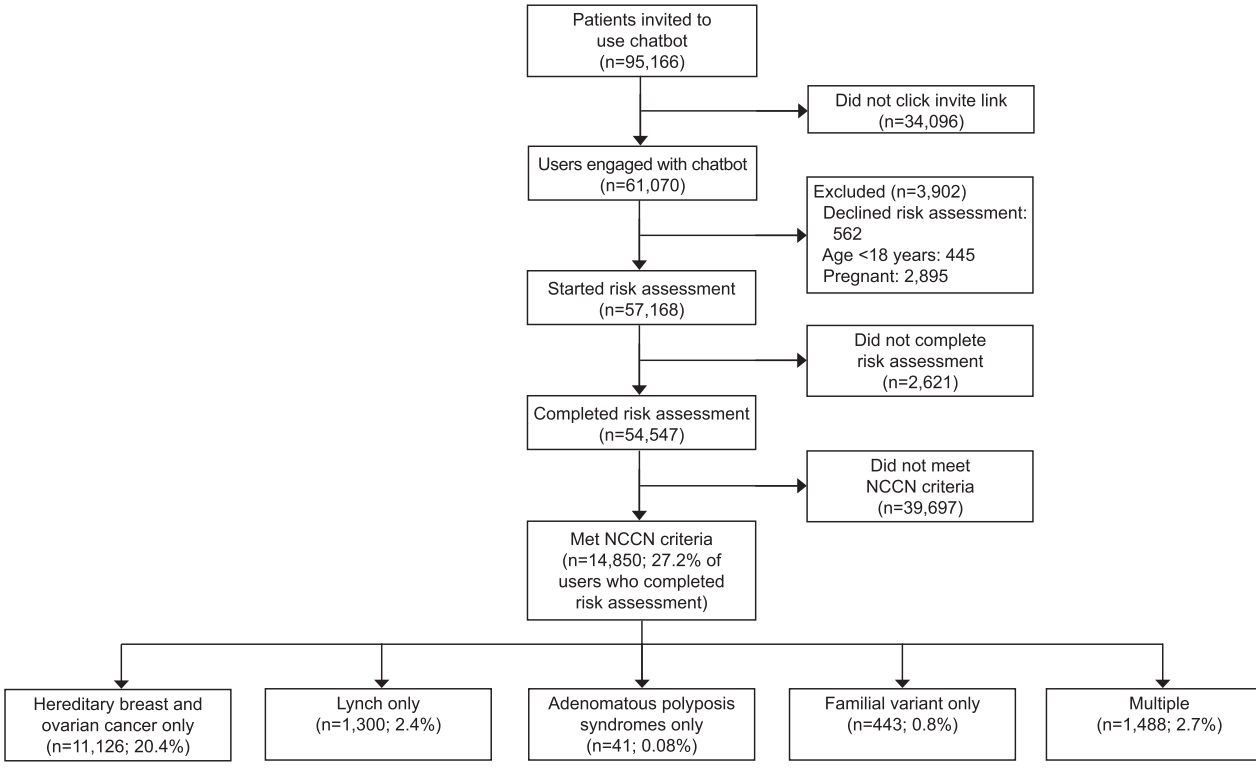

Fig. 2. Flow diagram of patient participation, chatbot uptake, and cancer risk outcomes. Percentages shown in the bottom row are calculated from among the 14,850 users who met National Comprehensive Cancer Network (NCCN) criteria for genetic testing. Multiple includes: hereditary breast and ovarian cancer and Lynch syndromes (n=1,137); hereditary breast and ovarian cancer syndrome and familial variant (n=267); hereditary breast and ovarian cancer, Lynch, and familial variant (n=42); Lynch and familial variant (n=19); hereditary breast and ovarian cancer and polyposis (n=12); Lynch and polyposis (n=6), hereditary breast and ovarian cancer, Lynch, and polyposis (n=4); hereditary breast and ovarian cancer, polyposis, and familial variant (n=1). An additional 452 patients met criteria, but data were missing on the specific syndrome.
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polyposis syndromes based on personal or family history and should have been offered genetic testing. However, less than 11% of users who were asked about previous genetic testing had reported that they had received it, suggesting a missed opportunity for cancer prevention measures. Among users with genetic testing results available through the chatbot platform, 5.6% had a positive variant. Thus, hereditary cancer risk is common enough among individuals receiving obstetrics and gynecology care or cancer screenings to warrant routine evaluation.

The low percentage of patients who had previously been offered testing may suggest a gap in familiarity with genetics among health care professionals, among other previously reported barriers,[21] which calls for greater professional education on medical genetics. Although genetic counselors can provide patient pretest education and testing services, the specialty is understaffed and overburdened.[22–24] A timestamp study of genetic counselor activities found that 64% of their time was spent in case preparation, follow-up, and administrative tasks, and only 20% of time was spent face-to-face with patients.[25] Interventions such as the chatbot could release some of the overencumbered genetic counselor time from administrative duties such as intake forms for complex genetics services and patient interaction.

As demonstrated in this study, digital platforms can also streamline clinic workflow by automatically flagging high-risk patients and collating data for a genetic test requisition form. Although the chatbot-collected data in this study was not automatically added to a patient's electronic medical record (EMR), health care professionals were provided composite data (and a transcript of the chatbot conversation) that could be transferred to the EMR. In the future, automatic transfer of discrete data to an EMR could save additional clinical time. For patients whose clinicians had access to the service, 29.0% had genetic testing ordered on their behalf through the digital platform

Table 2. Characteristics of Users Who Met Criteria for Further Cancer-Risk Evaluation

Characteristic	Criteria Met*		
	Lynch	Hereditary Breast and Ovarian Cancer	Tyrer-Cuzick
Total[†]	2,508 (100)	12,589 (100)	3,844 (100)
Age (y)			
18–24	42 (1.7)	507 (4.0)	132 (3.4)
25–39	387 (15.4)	2,642 (21.0)	746 (19.4)
40–60	1,276 (50.9)	6,007 (47.7)	2,612 (68.0)
Older than 60	799 (31.9)	3,392 (26.9)	336 (8.7)
Unknown	4 (0.2)	41 (0.3)	18 (0.5)
Race or ethnicity			
White	1,713 (68.3)	8,266 (65.7)	2,780 (72.3)
Black	226 (9.0)	1,300 (10.3)	382 (9.9)
Hispanic	243 (9.7)	1,110 (8.8)	209 (5.4)
Asian	56 (2.2)	323 (2.6)	69 (1.8)
Other[‡]	56 (2.2)	389 (3.1)	102 (2.7)
Multiple	214 (8.5)	1,201 (9.5)	302 (7.9)
Clinic type			
Obstetrics and gynecology	838 (33.4)	5,068 (40.3)	643 (16.7)
Cancer clinic	507 (20.2)	2,205 (17.5)	1,084 (28.2)
Gastrointestinal clinic	77 (3.1)	250 (2.0)	1 (0.03)
Primary care	31 (1.2)	225 (1.8)	22 (0.6)
Imaging clinic	1,048 (41.8)	4,814 (38.2)	2,091 (54.4)
Unknown	7 (0.3)	27 (0.2)	3 (0.08)

Data are n (%).
* Users who met both hereditary breast and ovarian cancer and Lynch syndrome criteria are included in both columns.
† Includes individuals younger than age 18 years or with unknown age.
‡ Other includes self-reported ancestry groups with limited numbers of users, including but not limited to users self-reported as French Canadian, Pacific Islander, Portuguese, Sephardic Jewish, and unknown.

during their appointment. The rate of uptake of the portal-based ordering was higher than has been previously reported, as prior studies found less than 20% of U.S. women with breast or ovarian cancer who met National Comprehensive Cancer Network criteria were tested,[9] and less than 10% of high-risk women referred to a genetic specialist by a physician actually followed through within 1 year.[22]

Family history collection is a well-established screening tool for evaluating hereditary cancer risk in patients, but it is time consuming and can be difficult to obtain in families with limited communication.[13–16] Although 70% of Americans report discussing family cancer history with at least one family member, fewer than one third report knowing their family cancer history well.[26] Prior findings indicate that digital tools can increase the odds of communicating with family about shared risks.[27] An advantage of a preappointment assessment tool is that users can leave the assessment to call relatives and then return to the assessment to complete it. Among users in our study who completed the risk assessment, 30% had gaps of inactivity longer than 5 minutes, sometimes longer than a day, which may represent time allotted for information gathering. Further, automating family history collection could 1) help standardize the information collected, including potentially overlooked history such as the occurrence of prostate cancer in the family; 2) address the need to repeat the assessment regularly to capture changing information (eg, family member diagnoses that occur after an initial assessment)[28]; and 3) remain current with changing guidelines. These reductions in clinical burden free up more time for health care professional–directed conversations about a patient's genetic risk.

Our findings that 27.2% of individuals assessed for cancer risk met National Comprehensive Cancer Network criteria for genetic testing and most had not previously had genetic testing are consistent with prior reports that most individuals at risk for hereditary breast and ovarian cancer or Lynch syndromes remain unidentified. A recent assessment of National Comprehensive Cancer Network guidelines found 28% of patients remain untested even after meeting criteria and are thus unable to receive preventive cancer care.[29] The U.S. Preventive Services Task Force has identified digital health as a means to make preventive care recommendations shareable and

Table 3. Genetic Testing Outcomes Among Users Who Received Genetic Testing After Platform-Based Test Referral

		Test Result*		
	Patients With Genetic Testing Results	Positive	Uncertain	Negative
Total†	1,313	73 (5.6)	342 (26.0)	898 (68.4)
By personal and family history‡				
Personal and family history	452	30 (6.6)	121 (26.8)	301 (66.6)
Personal history only	16	0 (0)	2 (12.5)	14 (87.5)
Family history only	844	43 (5.1)	219 (25.9)	582 (69.0)
By NCCN risk met				
Hereditary breast and ovarian cancer	993	58 (5.8)	267 (26.9)	668 (67.3)
Lynch	134	6 (4.5)	29 (21.6)	99 (73.9)
Hereditary breast and ovarian cancer and Lynch	137	8 (5.8)	33 (24.1)	96 (70.1)
Other or unknown§	49	1 (2.0)	13 (26.5)	35 (71.4)

NCCN, National Comprehensive Cancer Network.
Data are n or n (%).
* Positive, Uncertain, and Negative indicate results provided by genetic testing laboratories.
† Table excludes one user with negative results who had neither personal nor family history of cancer.
‡ No patients who met risk criteria for adenomatous polyposis syndromes or who reported a familial variant had genetic testing ordered within the study period.
§ The majority of patients in "Other or unknown" (46/49) met NCCN criteria but have missing data on the associated syndrome. The remaining three did not meet criteria.

consumable for clinicians and patients. Prior reports on internet-based family history collection tools and hereditary cancer risk assessment support this approach with demonstrations of acceptability and clinical validity.[30–33] A previously reported focus group of users of the chatbot found the technology to be acceptable for preappointment consent and genetic testing results follow-up.[20] Future work should explore barriers to implementation, which is perhaps the greatest challenge to broad utilization of such technologies.[34]

Strengths of this study include its large, U.S.-dispersed cohort of patients scheduled for routine care. In addition, unlike other studies on digital tools for risk assessment,[32] this study observed real-world experiences. Thus, our findings may be generalizable to many U.S.-based patients seeking routine care. However, the majority of patients were women, likely a result of the emphasis on women's health clinics and concerns such as the Tyrer-Cuzick risk screening. Although 10.9% of users identified as Hispanic, few (less than 1%) interacted with the chatbot in Spanish, which may have been a limitation of how this option was presented to patients (language preference is now required at the start of the chat). Another limitation was the lack of a comparison group that was not invited to engage with the chatbot and, therefore, the inability to determine whether chatbot engagement and associated genetic findings affected downstream preventive measures compared with standard of care. Further, our data on the rate of genetic testing among high-risk individuals could not account for testing undertaken outside of the digital portal. In addition, our observations regarding Tyrer-Cuzick criteria may overestimate user risk as our assessment did not take into account real-time breast density or unaffected female relatives. Finally, the study was limited to patients who had given an email address or smartphone number to their health care professional and responded to the digital invite, which may have introduced nonresponse bias for older age (as observed) or lesser digital literacy among nonusers compared with users. Future studies with broader sampling procedures could address this issue.

As calls for routine genetic testing for all breast and ovarian cancer patients grow,[35,36] there is a moral imperative to ensure that access is not limited to the most privileged. U.S. federal programs such as Healthy People 2030 and the Biden Cancer Initiative have set objectives to increase access to genetic counseling and intervention,[37] and more broadly identify patients at high risk of developing a hereditary cancer.[38] These goals may be accomplished with the adoption of vetted digital technologies that ensure equitable access to information across diverse patient populations.

REFERENCES

1. Kuchenbaecker KB, Hopper JL, Barnes DR, Phillips KA, Mooij TM, Roos-Blom MJ, et al. Risks of breast, ovarian, and contra-

lateral breast cancer for BRCA1 and BRCA2 mutation carriers. JAMA 2017;317:2402–16. doi: 10.1001/jama.2017.7112

2. Fan X, Wynn J, Shang N, Liu C, Fedotov A, Hallquist MLG, et al. Penetrance of breast cancer susceptibility genes from the eMERGE III network. JNCI Cancer Spectr 2021;5:kab044. doi: 10.1093/jncics/pkab044
3. Buchanan AH, Manickam K, Meyer MN, Wagner JK, Hallquist MLG, Williams JL, et al. Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. Genet Med 2018;20:554–8. doi: 10.1038/gim.2017.145
4. Buchanan AH, Kirchner HL, Schwartz MLB, Kelly MA, Schmidlen T, Jones LK, et al. Clinical outcomes of a genomic screening program for actionable genetic conditions. Genet Med 2020;22:1874–82. doi: 10.1038/s41436-020-0876-4
5. Nelson HD, Fu R, Zakher B, McDonagh M, Pappas M, Stillman L. Medication use for the risk reduction of primary breast cancer in women: a systematic review for the U.S. Preventive Services Task Force. Agency for Healthcare Research and Quality (US); 2019.
6. Domchek SM, Friebel TM, Singer CF, Evan DG, Lynch HT, Isaacs C, et al. Association of risk-reducing surgery in BRCA1 or BRCA2 mutation carriers with cancer risk and mortality. JAMA 2010;304:967–75. doi: 10.1001/jama.2010.1237
7. Hull LE, Haas JS, Simon SR. Provider discussions of genetic tests with U.S. women at risk for a BRCA mutation. Am J Prev Med 2018;54:221–8. doi: 10.1016/j.amepre.2017.10.015
8. Bellcross CA, Peipins LA, McCarty FA, Rodriguez JL, Hawkins NA, Hensley Alford S, et al. Characteristics associated with genetic counseling referral and BRCA1/2 testing among women in a large integrated health system. Genet Med 2015;17:43–50. doi: 10.1038/gim.2014.68
9. Childers CP, Childers KK, Maggard-Gibbons M, Macinko J. National estimates of genetic testing in women with a history of breast or ovarian cancer. J Clin Oncol 2017;35:3800–6. doi: 10.1200/JCO.2017.73.6314
10. Cragun D, Weidner A, Lewis C, Bonner D, Kim J, Vadaparampil ST, et al. Racial disparities in BRCA testing and cancer risk management across a population-based sample of young breast cancer survivors. Cancer 2017;123:2497–505. doi: 10.1002/cncr.30621
11. Hereditary breast and ovarian cancer syndrome. Practice Bulletin No. 182. American College of Obstetricians and Gynecologists. Obstet Gynecol 2017;130:e110–26. doi: 10.1097/AOG.0000000000002296
12. Lu KH, Wood ME, Daniels M, Burke C, Ford J, Kauff ND, et al. American Society of Clinical Oncology expert statement: collection and use of a cancer family history for oncology providers. J Clin Oncol 2014;32:833–40. doi: 10.1200/JCO.2013.50.9257
13. Vig HS, Armstrong J, Egleston BL, Mazar C, Toscano M, Bradbury AR, et al. Cancer genetic risk assessment and referral patterns in primary care. Genet Test Mol Biomarkers 2009;13:735–41. doi: 10.1089/gtmb.2009.0037
14. Armel SR, McCuaig J, Gojska N, Demsky R, Maganti M, Murphy J, et al. All in the family: barriers and motivators to the use of cancer family history questionnaires and the impact on attendance rates. J Genet Couns 2015;24:822–32. doi: 10.1007/s10897-014-9813-5
15. Lynn B, Hatry A, Burnett C, Kan L, Olatunbosun T, Bluman B. Identifying primary care physicians continuing education needs by examining clinical practices, attitudes, and barriers to screening across multiple cancers. J Cancer Educ 2018;33:1255–62. doi: 10.1007/s13187-017-1240-5
16. Campbell-Salome G, Rauscher EA, Freytag J. Patterns of communicating about family health history: exploring differences in family types, age, and sex. Health Educ Behav 2019;46:809–17. doi: 10.1177/1090198119853002
17. Nazareth S, Nussbaum RL, Siglen E, Wicklund CA. Chatbots & artificial intelligence to scale genetic information delivery. J Genet Couns 2021;30:7–10. doi: 10.1002/jgc4.1359
18. Bibault J-E, Chaix B, Guillemassé A, Cousin S, Escande A, Perrin M, et al. A chatbot versus physicians to provide information for patients with breast cancer: blind, randomized controlled noninferiority trial. J Med Internet Res 2019;21:e15787. doi: 10.2196/15787
19. Prenatal diagnostic testing for genetic disorders. Practice Bulletin No. 162. American College of Obstetricians and Gynecologists. Obstet Gynecol 2016;127:e108–22. doi: 10.1097/AOG.0000000000001405
20. Schmidlen T, Schwartz M, DiLoreto K, Kirchner HL, Sturm AC. Patient assessment of chatbots for the scalable delivery of genetic counseling. J Genet Couns 2019;28:1166–77. doi: 10.1002/jgc4.1169
21. Douma KFL, Smets EMA, Allain DC. Non-genetic health professionals' attitude towards, knowledge of and skills in discussing and ordering genetic testing for hereditary cancer. Fam Cancer 2016;15:341–50. doi: 10.1007/s10689-015-9852-6
22. Kne A, Zierhut H, Baldinger S, Swenson KK, Mink P, McCarthy Veach P, et al. Why is cancer genetic counseling underutilized by women identified as at risk for hereditary breast cancer? Patient perceptions of barriers following a referral letter. J Genet Couns 2017;26:697–715. doi: 10.1007/s10897-016-0040-0
23. Swink A, Nair A, Hoof P, Matthews A, Burden C, Johnson K, et al. Barriers to the utilization of genetic testing and genetic counseling in patients with suspected hereditary breast and ovarian cancers. Proc 2019;32:340–4. doi: 10.1080/08998280.2019.1612702
24. Boothe E, Greenberg S, Delaney CL, Cohen SA. Genetic counseling service delivery models: a study of genetic counselors' interests, needs, and barriers to implementation. J Genet Couns 2021;30:283–92. doi: 10.1002/jgc4.1319
25. Attard CA, Carmany EP, Trepanier AM. Genetic counselor workflow study: the times are they a-changin'? J Genet Couns 2019;28:130–40. doi: 10.1002/jgc4.1041
26. Krakow M, Rising CJ, Trivedi N, Yoon DC, Vanderpool RC. Prevalence and correlates of family cancer history knowledge and communication among US adults. Prev Chronic Dis 2020;17:E146. doi: 10.5888/pcd17.200257
27. Wang C, Sen A, Plegue M, Ruffin MT, O'Neill SM, Rubinstein WS, et al. Impact of family history assessment on communication with family members and health care providers: a report from the Family Healthware™ Impact Trial (FHITr). Prev Med 2015;77:28–34. doi: 10.1016/j.ypmed.2015.04.007
28. Hereditary cancer syndromes and risk assessment. ACOG Committee Opinion No. 793. American College of Obstetricians and Gynecologists. Obstet Gynecol 2019;134:e143–9. doi: 10.1097/AOG.0000000000003562
29. Alberty-Oller JJ, Weltz S, Santos A, Pisapati K, Ru M, Weltz C, et al. Adherence to NCCN guidelines for genetic testing in breast cancer patients: who are we missing? Ann Surg Oncol 2021;28:281–6. doi: 10.1245/s10434-020-09123-z
30. Cohn WF, Ropka ME, Pelletier SL, Barrett JR, Kinzie MB, Harrison MB, et al. Health Heritage© a web-based tool for the collection and assessment of family health history: initial user experience and analytic validity. Public Health Genomics 2010;13:477–91. doi: 10.1159/000294415

31. Baumgart LA, Postula KJV, Knaus WA. Initial clinical validation of Health Heritage, a patient-facing tool for personal and family history collection and cancer risk assessment. Fam Cancer 2016;15:331–9. doi: 10.1007/s10689-015-9863-3

32. Bucheit L, Johansen Taber K, Ready K. Validation of a digital identification tool for individuals at risk for hereditary cancer syndromes. Hered Cancer Clin Pract 2019;17:2. doi: 10.1186/s13053-018-0099-8

33. Li W, Murray MF, Giovanni MA. Obtaining a genetic family history using computer-based tools. Curr Protoc Hum Genet 2019;100:e72. doi: 10.1002/cphg.72

34. Powell J. Trust Me, I'm a chatbot: how artificial intelligence in health care fails the Turing test. J Med Internet Res 2019;21:e16222. doi: 10.2196/16222

35. Stoll K, Kubendran S, Cohen SA. The past, present and future of service delivery in genetic counseling: keeping up in the era of precision medicine. Am J Med Genet C Semin Med Genet 2018;178:24–37. doi: 10.1002/ajmg.c.31602

36. US Preventive Services Task Force, Owens DK, Davidson KW, Krist AH, Barry MJ, Cabana M, et al. Risk assessment, genetic counseling, and genetic testing for BRCA-related cancer: US Preventive Services Task Force recommendation statement. JAMA 2019;322:652–65. doi: 10.1001/jama.2019.10987

37. U.S. Department of Health and Human Services, Office of Disease Prevention and Health Promotion. Increase the proportion of people with colorectal cancer who get tested for Lynch syndrome–C-R03. Accessed March 23, 2021. https://health.gov/healthypeople/objectives-and-data/browse-objectives/cancer/increase-proportion-people-colorectal-cancer-who-get-tested-lynch-syndrome-c-r03

38. Siyahian K. The Biden Cancer Initiative: an interview with Gregory C. Simon, JD, about the Biden Cancer Initiative's strategies to improve cancer care. Accessed March 22, 2021. https://www.jons-online.com/issues/2019/march-2019-vol-10-no-3/2324-the-biden-cancer-initiative

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